MPS

Summer 2010

Society for Mucopolysaccharide Diseases

What's Inside...

New recommendations for Fabrazyme
plus a round up of recent events organised by the MPS Society

The MPS Society

Founded in 1982, the Society for Mucopolysaccharide Diseases (the MPS Society) is the only national charity specialising in MPS and Related Diseases in the UK, representing and supporting over 1200 affected children and adults, their families, carers and professionals. The MPS Society:

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

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

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

MPS & Related Diseases

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

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

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

Where does your money go?

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

Access to clinical management and palliative care

MPS Regional Specialist clinics

Support with disability benefits

Paving a child's way in accessing education

Upholding rights in employment

Advising on home adaptations

Bereavement support

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

Front cover photo: Isaac Turner (MPS I BMT) attends Scouting Parade at Windsor Castle. For full story turn to page 10.

Society for Mucopolysaccharide Diseases

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

Staff

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

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

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

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

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

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

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

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

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

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

Magazine Deadlines

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

Friend of MPS

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

The articles in this magazine do not necessarily reflect the opinions of the MPS Society or its Management Committee. The MPS Society reserves the right to edit content as necessary. Products advertised in this newsletter are not necessarily endorsed by the Society.

IN THIS ISSUE...

Summer 2010

NEW! Children's Newsletter

We are delighted to be able to include our first **MPS Children's Newsletter** alongside our Summer 2010 issue of the MPS Magazine.

We hope that you all enjoy reading through the stories that have been sent to us so far from children that are affected by MPS or related diseases.

We hope that it will encourage all the children including brothers and sisters who read it to get in touch with us. Tell us what you think about what we have done so far and what you would like to see in the next edition.

We would love to hear from you all so why not get in touch and tell us all about your life at home and at school. Tell us about the people that you love spending time with. Do you or your brothers and sisters have an MPS or a related disease? Tell us all about them and what you do together.

You can even send us your drawings, a short story or a poem. **We can't wait to hear from you!**

You can get in touch via email: newsletter@mpssociety.co.uk

Or via post: MPS Society, MPS House, Repton Place, White Lion Road, Amersham, HP7 9LP

Please get permission from your parents first!

TRUSTEES' REPORT

I am pleased to present the Trustees' Report for the year ending 31 October 2009.

The past year has for the MPS Society and its Board of Trustees proved to be a mixture of unexpected challenges combined with the rewards of achieving our aims and serving the public interest of the charity.

The MPS Society can be proud of the services it has delivered to those affected by these devastating diseases. The support offered within the individual advocacy service is necessarily wide-ranging with the aim of meeting the needs of a diverse group of 1200 affected children and adults and their families. The men, women and children with Fabry or Morquio disease have completely different support needs to those with Sanfilippo disease or Mucolipidosis. In Fabry disease it is not unusual to have three or four generations of one family suffering from the condition which untreated leads to progressive renal failure, cardiac involvement and susceptibility to life-threatening TIAs and strokes. Since 2001, when Enzyme Replacement Therapy (ERT) was approved in Europe, our 250 Fabry members have been able to look forward to their disease stabilising and an improved quality of life. Bone Marrow Transplant (BMT) and Cord Blood Transplant has also provided an imperfect treatment option for MPS I, Hurler disease whilst ERT has also changed the course of disease for those suffering from MPS I, Hurler Scheie and Scheie, MPS II, Hunter, attenuated form and MPS VI, Maroteaux Lamy.

This is all good news but where does that leave our members affected by the other seventeen diseases? In 2009 a Phase I/II clinical trial for ERT in MPS IVA, Morquio disease started at three specialist centres in England. The safety trial has continued into 2010 when it is hoped a Phase III/IV clinical trial will prove efficacy. The other sixteen diseases all present huge difficulties for the patients who bear them, the doctors who manage the clinical manifestations, and the research scientists looking for potential breakthroughs. Many of these diseases are associated with progressive neurodegenerative brain disease where the only treatment is palliative. In 2008, funded by the Roald Dahl Foundation, our families were able to rely on the support of a dedicated palliative care and bereavement advocacy worker. Whilst 40 to 50 babies and young children will be diagnosed with MPS each year, another 20 or so will lose their life to MPS. Supporting our 300 bereaved families, many of whom have stayed with the MPS Society for one or two decades, is also important. Families have the opportunity to plant a tree in the Childhood Wood following the death of a loved one with an MPS or Related Disease and each year will be invited to a Remembrance Day in the Wood.

Support activities and events are an important part of the MPS calendar and included a week at Boreatton Park for MPS siblings last August, a London Zoo family day last July, a weekend in London for MPS young adult sufferers, and a family day at the Hilton Hotel Templepatrick in Belfast. Every two years the MPS Society holds a Weekend Family Conference. In June 2009 we held our biggest one yet. It was so big with over 350 adults and 100 children attending that the gala dinner had to be held in a marquee in the grounds of the Hilton Hotel Northampton.

The Society is a respected key player in supporting the development of new strategies to clinically manage affected adults and children. During 2009 the Chief Executive Officer and Senior Advocacy Officer led discussions on the transition of children from paediatric to adult clinical care. Many adult clinics are far from suited to the needs of young adults with no self-help skills, no mobility, no communication and doubly incontinent, all the result of MPS neurodegenerative brain disease. Equally, we have a number of wholly independent young adults less than one metre tall, wheelchair dependent, not needing a carer but unable to be accommodated with dignity when being seen in an adult clinical setting. These are all serious transition issues that affect our members and indeed many others with rare diseases.

Public awareness and communications are paramount to getting the MPS message out. During this reporting period the Society appointed its first full time Communications Officer who is working on improving the accessibility of the website, producing the quarterly MPS Magazine and devising new communication tools for our members. On-going communication with our fundraising supporters has been key to us in these difficult economic times. We have learnt that our fundraisers want to have fun and do something for themselves in supporting the MPS Society. As a result many of the fundraising events offered by the Society in the past year have created a sense of fun or encouraged personal achievement.

Funding research, after providing support and advocacy for our members, is the most important objective. The MPS Society first started funding basic research in 1985 and over the past 25 years has raised nearly £4 million for research that may lead to a therapeutic approach for these diseases. In the last 12 years the MPS Society has received significant funds as a partner in the Jeans for Genes Charity. Regrettably, the net funds available to the Jeans for Genes Charity Trustees to distribute equally between the four partner charities in 2008 and 2009 fell well short of what

might have been expected, due to the high cost / income ratios. As a consequence, the MPS Society apart from its support to the Programme Grant for the MPS Stem Cell Group at the University of Manchester and a second year grant to the Blood Brain Barrier Group at Kings College, London, has only been able to offer support to small research grant initiatives in this financial year.

Mindful of the economic challenges ahead, the MPS Trustees set a prudent budget prior to the beginning of this financial year that provided for all core costs and essential events and allowed for additional support activities to take place as and when restricted funding for that purpose was achieved. Furthermore, as part of our fundraising strategy and following on from a fruitful and successful three years with a fundraising consultant, the Society appointed its first full time Corporate and Trusts Fundraising Officer.

We end this financial year knowing that our efforts to increase the level of unrestricted income have been rewarded and we have been able to reduce the mortgage on MPS House. Most importantly of all we have been able to provide a unique and disease specific support service to all those affected by MPS and related diseases including parents, partners, siblings and extended family. This could not have been achieved without the commitment of dedicated staff, volunteers, our members and MPS supporters.

Barry Wilson
Chairman of Trustees

Highlights from the Management Committee

The Society's Board of Trustees meet regularly. Here is a summary of the key issues that were discussed and agreed at the Management Committee Meeting held on 19 - 20 March 2010.

GOVERNANCE
Trustees approved two new policies, Volunteer Fundraising Policy and the Protection of Vulnerable Adults Policy. The Trustees declined to approve the Fundraising Complaints Procedure until they had had sight of all the guidance paperwork.The Maternity Policy and Policy on Leave for Domestic and Personal Reasons were reviewed and approved.

PERSONNEL
The CEO reported on how well the two new members of staff, the Office Administrator and Special Projects and Communications Assistant had settled in.

RISK MANAGEMENT
Trustees reviewed the Risk Register and the CEO advised on a number of small points. The Finance Officer is compiling a Register of Assets. The Disaster Recovery Plan is still under development and the Staff Handbook is nearing completion.

TREASURER'S REPORT
The Treasurer presented her report and Trustees agreed that the Society is holding its own considering the current financial climate. The Trustees were asked to consider the draft accounts for year ending 31 October 2009 and the Trustees went through them in fine detail. The Chairman explained that there was a change in format because the report has to be comprehensive for grant applications as grant giving bodies will often not read supplementary documents. The Trustees approved the accounts for year ending 31 October 2009 subject to the scrutiny of the Chairman of the ammendments put forward.

MPS Annual Review & Accounts 2009
available online at www.mpssociety.co.uk

JEANS FOR GENES
The MPS Trustees were introduced to Christopher Syrda, the MPS nominated Jeans for Genes Trustee. Mr Syrda said that everyone had been very welcoming at his first J4G Trustees meeting. He reported that the J4G Trustees had agreed to seek professional advice regarding the handover of the J4G trademark post March 2011 and suggested the MPS Trustees might also need legal representation. Christopher Syrda also reported that he had been appointed to the Finance Committee and a discussion was had about costs, the J4G management of income and the campaign costs after 31 October when J4G no longer has the licence to use the J4G trademark. Christopher Syrda also said that he wants to ensure that the transfer of the licence is handled appropriately and wants to ensure that everything is transparent. A detailed discussion took place regarding what the best outcome would be for the MPS Society regarding the future of J4G.

CLINICAL MANAGEMENT
The CEO spoke of Dr Uma Ramaswami's prposal to move from Addenbrooke's Hospital, Cambridge to Great Ormond Street Hospital. This matter is being considered by the National Commissioning Group and any decision will be communicated to all concerned once any appointment has been approved.

COMMUNICATIONS
The Communication Officer's report was received in advance and copies of the new-look Fundraising Magazine were given to all Trustees. The CEO explained that the Communicatiuons Officer had worked very hard on revising and updating the format and it will now be published twice a year and will include fundraisers telling their stories and gving information as to how the money raised is being spent.

WHAT'S ON 2010!

CONFERENCE EVENTS
MPS III Expert Meeting, Northampton
27 - 28 August

Scottish LSD Conference
23 – 25 September

REGIONAL EVENTS
Birmingham Family Day, Cadbury World
12 September

London Family Event, Science Museum
16 October

Last call for family bookings
for the Cadbury World Family Day.
Contact the MPS office for a booking form.
T: 0845 389 9901, Email: mps@mpssociety.co.uk

MPS CLINICS
Birmingham clinic
9 July, 26 November

MPS I Bone Marrow Transplant clinic (under 6's)
16 July, 15 October

MPS I Bone Marrow Transplant clinic (over 6's)
23 July, 22 October

Bristol clinic
8 September

Northern Ireland Clinic
November (tbc)

Cardiff clinic
December (tbc)

Last chance to book your place!

Expert Meeting on MPS III, Sanfilippo Disease

Friday 27th - Saturday 28th August 2010, Hilton Northampton

This expert meeting will offer individuals, parents, partners, carers and professionals the opportunity to hear state of the art talks on clinical management, research and clinical initiatives for those affected by MPS III.

This event will not only give you the opportunity to meet and talk informally with professionals, it will encourage individuals, parents, partners and carers to share their own experiences with others in similar circumstances.

There will also be a children and vulnerable adults activity programme for children and young adults affected by Sanfilippo disease and their siblings aged 17 and under.
This will allow parents and carers to take in the various aspects of the conference as fully as possible knowing that their children will be cared for all the while having fun and making new friends.

Among our many esteemed Professional Speakers, who include Dr. Fiona Stewart, Dr. Brian Bigger, Dr. Ed Wraith, Dr. Brett Crawford, Dr. Chris Hendriksz and Prof. Bryan Winchester (to name but a few) we are also delighted to welcome a few of our MPS Family Members to speak about their personal experiences.

Overnight accommodation comprises of: Friday Lunch, Friday Conference, Friday Gala Dinner and overnight accommodation as well as Saturday breakfast, lunch and the Saturday Conference. The cost for a single room conference package is £60. A Double/Twin room conference package is £120. A family room conference package is £140. The MPS Society has subsidised these costs for members of the Society.

Please contact us for the cost of attendance if attending from outside the UK.

Should you be unable to make the most of the full conference package over the Friday and Saturday duration, alternative arrangements can be made depending on your availability.

For more information and to book your place please contact Fiona Hopson on 0845 389 9901 or email: f.hopson@mpssociety.co.uk. We look forward to hearing from you soon!

New Members

Ms Brown has recently been in contact with the Society. Joseph has a diagnosis of ML. Joseph is 16 months old and the famly live in the South West.

Mr and Mrs Brown have recently been in contact with the Society. Sam has a diagnosis of Morquio disease. Sam is 2 years old in August and the family live in the North of England.

Births

Congratulations to Claire Arrowsmith, sister to Colin (MPS II) and her husband Dan on the birth of their daughter, Sophie, on 6 May 2010.

Deaths

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

Shujah Altaf who suffered from Sanfilippo disease and who passed away on 30 March 2010 aged 16 years.

Jordan Lacey who suffered from Sanfilippo disease and who passed away on 10 April 2010 aged 8 years.

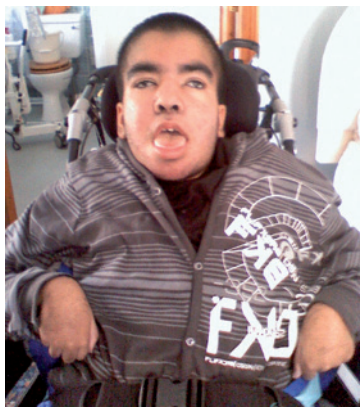
Mohammed Razaq who suffered from Maroteaux Lamy disease and who passed away on 22 May 2010 aged 16 years.

Sad News

Many of you will have been shocked by the deaths of three people, two teenagers on their way home on the school bus in Cumbria on Monday 24 May 2010. This was a double tragedy for the Walker family who lost their daughter Chloe who was celebrating her 16th birthday that Monday. Chloe's brother Jordan died in April 2007 from Sanfilippo Disease. Sharon and John Walker have been members of the MPS Society since Jordan's diagnosis. I am sure that you would want to join with the MPS staff and Trustees in sending our deepest condolences and love at this terribly sad time. At the Society's AGM on Sunday 30 May 2010 a one minute silence was held to remember Chloe, her family and all others who have lost loved ones.

In Memory

SHUJAH ALTAF
5th January 1994 - 31st March 2010



My Amazing Brother
In loving memory of Shujah

My brother is the cutest; he has the perfect smile,
Seeing his face light up makes my life worthwhile,
He is the most special boy I know,
His skin is baby soft and his face has a lovely glow.

My brother was diagnosed with Sanfilippo at the age of three,
I never thought life would turn out like this; it was a huge shock to me,
He would play with his ball like any other kid, he can hear and he can see,
But underneath it all lays pain and misery.

When he was little he could walk and there were even some words he could say,
My mum always played with him and cherished everyday,
That innocent face and those tiny hands really melted my heart,
He was always such a bubbly child; I had nicknames for him from the start,
I call him my bubbly, sugar puffs and cutie pie,
I will love my brother always and forever till the day I die.

He looks around with his beautiful eyes, he tries to communicate,
I talk to him and try to make him smile; he has a horrible fate,
None of us can imagine what he is going through,
His condition has worsened and he is always so blue.

I love seeing him laugh, he never laughs anymore,
I hate the fact that I never see him happy now; I don't know what's in store,
I can't believe this is happening; it's a terrible turn of events,
I feel so bad about this; I wish it was something I could prevent.

I want him to get better; I can't bear to see him in pain,
I want to see him smile; I want to stop the rain,
He is suffering, he's just an innocent child, it's so unfair,
He doesn't deserve to go through all this pain, he deserves love and care.

Ever since I was little I always wanted a brother,
I got what I wanted, he's so precious to me and I want to thank my mother,
Every moment with him counts, all the time spent with him is a joy,
He means the world to me; he is such a sweet boy.

It makes me sad when I think of what he could have become,
He never got the chance; life is taken for granted by some,
I know that he would have been really smart and would have made it far,
He would have been nice and caring, he is a sparkling star.

My brother will be in my heart always and forever,
He has touched the lives of many; he will never be forgotten ever,
He is an inspiration; he is so brave and strong,
I would do anything to see him happy; I would even write him a song.

Some people look at my brother, they don't understand,
They don't know what it feels like to hold his tiny hand,
He is so fantastic, I look at his cute face and he brightens up my day,
If I could, I would make all his pain go away.

It makes me cry when I see him cry,
When that happens, I die inside,
To me, he's like an adorable baby,
One of a kind, my brother is amazing.

Focus on Bereavement

During our last year's conference in Northampton, we looked at the support provided by the MPS Society and particularly about bereavement and how to recognise and understanding the grieving process.

It is important to remember that grief affects everyone differently and will follow many different patterns. There is no 'right way' to grieve as it is a very unique and individual experience. As no two people grieve in the same way, this in itself can place a strain on family relationships.

The death of a family member can be a devastating experience and can bring about stronger emotions than we ever thought possible. This can involve a long and difficult period of grieving and during this time behaviour may vary and feelings pushed to the extremes, further than we ever felt possible.

For some people the grieving process starts at the time of death, for others it will begin at the time of diagnosis.

Suddenly the world in which we were familiar with has changed and feelings of helplessness may occur as the reality begins to set in. Around this time we may also experience a sense of hopelessness as it can be difficult to adapt to the new changes, meeting professionals, dealing with local councils, special educational needs, these all may be so different to the aspirations of parenthood which we once hoped for.

The Grieving Process

As we begin to understand grief is an individual experience and follows many different patterns:

Shock and disbelief: Unable to accept the fact of death, there may be feelings of numbness, panic, anger or it isn't happening. These feelings can last a short time, but it is not unusual for the emotions to last longer.

The Loss: Feelings of loss and loneliness may strike particularly when the network of support (family and friends) return to their own lives. This is particularly common following a funeral, for others life goes on and they return to their routines, yet for the bereaved this may be an extremely lonely time. The world they once knew has been changed and will never be the same again. Feelings of isolation, changes to unfamiliar and new routines, lots of time on their hands, all contribute to the awareness of a loss. Sleep can be disrupted with feelings of wakefulness at certain times as well as experiencing vivid dreams.

Changes: Awareness of changes in family structure, feeling different, lots of time now available and even a sense of feeling insecure as the world which was once so familiar has taken a different direction.

Despair: At times this can be so overwhelming, often described as a huge wave of emotions, which can occur without warning and little understanding of their triggers (could be a song, a smell, favourite food, book, clothing, film…). There may be feelings of yearning for your loved one, total sadness and tears of pain for your loss, even a sense of what's the point? It is usually at this stage (whenever that may be), that support from qualified professionals may ease this and help you to understand the grieving process, either by a referral from your Doctor for counselling or by contacting one of the many organisations who offer support to the bereaved, for example: Child Bereavement Trust, Children's Hospice, Cruse, Samaritans or other local networks in your area.

Re-organisation: Often as you begin the re-building of your life, get new routines and are able to look forward and finding a sense of purpose in life, may come feelings of guilt. Questions such as "I went on holiday, or I went out with friends and had a fabulous time" may often leave you feeling guilty or ashamed that you were able to enjoy yourself. Unfortunately the reality is that life does go on and whilst you can never replace and will never forget that very special person, the past will always be part of you, and by enjoying yourself you will not affect this.

Bereavement is something which we will all experience at some time during our life, and every loss is a unique experience, we need to remember to be gentle with ourselves. **Linda Warner**

Farewell to Linda

It is with a very heavy heart that I say farewell to everyone at the MPS Society as I leave for pastures new. I am retiring to the seaside, where I plan to take up and spend my days painting and walking.

The last 2 and a bit years have been incredible for me and a time which I shall always remember fondly. I have enjoyed working with families, professionals and fellow work colleagues and being part of an incredible team at MPS House. I shall miss everyone very much.

I would also like to say a big thank you to everyone for making me feel so welcome in your homes. **Linda Warner**

I am sure all the families who have been supported by Linda would want to wish Linda every happiness in her retirement. Linda we will miss you lots.
Christine and the MPS Staff Team

Aaryanna celebrates her First Holy Communion

Our daughter Aaryanna has MPS I Hurler Scheie. Aaryanna celebrated her First Holy Communion on Saturday 8th May 2010. It was a very special day for us all and the air was palpable with excitement for many weeks prior! Aaryanna had a starring role in her ceremony as one of the Narrators of the Gospel story that day and was so well rehearsed that she was saying it in her sleep!

Following the ceremony, we were joined at our home by extended members of our family for a big party and despite the joy and excitement of the day, it was clear that Aaryanna was harbouring a degree of disappointment. She had desperately wanted a bouncy castle to play on during the party, alas, as a sufferer of MPS I HS, medical advice would not permit one... so unknown to Aaryanna, we secretly made contact with a wonderful company called The Belfast Circus School, based in Northern Ireland. We explained our situation to them... we wanted an entertainer to attend our family party. The Circus School don't usually provide family party entertainers, however, on hearing about Aaryanna's condition and her physical restrictions, they immediately agreed to help and arranged for a fantastic Fire Juggler, known as "Jitterbug Jackson" to produce a show especially for Aaryanna.

Jitterbug Jackson arrived in secret and set up stage in our garden. The show commenced and immediately the laughter and tears started. He was totally brilliant. The children and adults alike loved him and nearly two weeks later, they are still talking about his show! Aaryanna was so amazed and surprised. All longings and disappointment about bouncy castles immediately disappeared and she sat in awe as Jitterbug Jackson rode on an 8ft high uni-cycle, wearing a 3ft long hat which blazed with purple smoke, while he juggled fire batons! Finally, he presented Aaryanna with a bouquet of flowers. She was totally thrilled to bits!

Speaking to Jitterbug Jackson in our home as he packed his stage equipment away was equally amazing, when, he announced he did not want his fee for his performance and he insisted we spend the money on Aaryanna. Despite arguing with him, he insisted that his mind was already set and he flatly refused to accept his performance fee. Consequently, the Belfast Circus School has also declined to accept their fee. In light of that, Aaryanna has since decided to buy a very special charm for a beautiful charm bracelet she had received earlier that day. The first charm she will choose will be a permanent reminder of the wonderful Jitterbug Jackson and the amazing show he performed for her on her very special day.

In light of Jitterbug Jackson's generosity, his enormous heart and his wonderful compassion for our daughter and in recognition of the generosity of The Belfast Circus School, we wish to donate their fee of £450 to the MPS Society. Further, we have decided to match that fee and have today transferred a total of £900 via online transaction into the MPS Account. Please acknowledge that half of this donation, although carried out by us, is actually on behalf of Jitterbug Jackson and The Belfast Circus School. We want to thank them so very much for their kindness. **Helen and Nigel Lever**

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

Isaac attends Scouting Parade at Windsor Castle

This is the story of an 11 year old MPS child, a noted TV survivalist and the heir to the throne.

Isaac Turner (MPS I Hurler BMT) won the Cornwell Award last year, a very rarely awarded honor in the Scouting movement. Isaac won the award for Scouting in the face of adversity. Despite his many setbacks and medical problems Isaac has always wanted to go to Cubs and Scouts.

As part of the award Isaac and his family were invited to the annual St George's Day Scouting Parade at Windsor Castle. A very proud pair of parents (Adam and Lou) and sister (Eliza) made their way to Windsor on Sunday April 25th not entirely sure what to expect.

After watching the changing of the guard and seeing the huge numbers of scouts in and around Windsor we realised what an event this was going to be. We entered the castle and Isaac was directed to a seating area at the top of the inner quad. Isaac was sitting with the other gallantry award winners, the only person not at all un-nerved by the prospect.

After arranging the seats and discussing how we would all respond and reply to Prince Charles when he came to be introduced to the twenty or so award winners, some people were getting sweaty palms, and not just because the sun had come out. Isaac was unfazed and chatted merrily to other awardees.

After a while, the Chief Scout and famous survivalist Bear Grylls came over to meet us all, and started with Isaac. Bear lent over and began to talk to Isaac and his dad.

By this point Isaac had demolished a chocolate bar, and as Bear's neckerchief dangled invitingly in front of him, Isaac spoke to Bear Grylls, all the while rubbing chocolate all over the Chief Scout's neckerchief.

Undeterred both Bear and Isaac went on. A short time later, Prince Charles came out and received the salute. He made his way round the different Scout troops and bands, finishing off with meeting the Gallantry Award winners.

Bear Grylls introduced Prince Charles to Isaac, and Isaac had two questions for him - firstly 'Are you Charles?' followed up with 'Is your Mum the Queen?' Prince Charles dealt with both questions and learnt about Isaac's award. Isaac then took part in the march down to St George's Chapel, a short service and then a special salute in the Horseshoe Cloister. All in all, Isaac and his family had a lovely day out, met various interesting people and lots of senior scout leaders who treated Isaac and his Cornwell Award with huge respect, and took part in a special event.

As ever Isaac's approach to life and his resilience make us hugely proud of him. **Adam and Louise Turner**

Earlier this year a group of our MPS III Sanfilippo member families (pictured left) got together for a day out at Warwick Castle. It was a cold and damp day but this didn't dampen spirits and a good time was had by all. The team at Warwick Castle were very helpful.

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

The Rocky Road of Transition

I decided to write about my experiences along the rocky road of transition from children's to adult services although we are not yet at the end of all the changes because it is certainly not an easy process. Luckily for us I had decided to take early retirement when my son was seventeen and have needed a lot of my "spare time" to work through the maze. I have an eighteen year old Sanfilippo son called Nathan.

We had a comfortable set up prior to his eighteenth birthday with as much help as we needed. Nathan was at college during term time and had a P.A for three hours per week to help with taking him out if one or other of us wanted to go somewhere. We also had help twice a week from a charitable organisation called Crossroads so that we could go out together and know that Nathan was in good hands. Lastly we had respite from Demelza House Children's Hospice.

Then as he approached his eighteenth birthday people started to say the T word. I was dreading it and the more I voiced my worries to people such as social workers, for example, the more I was reassured that it would all be fine and that all agencies concerned would be there to help and guide us through it. Although I'm afraid that was not to be in all areas and some problems are ongoing although we have also experienced some positives.

Nathan has had or is experiencing all the usual Sanfilippo traits and difficulties which means he requires constant supervision and twenty four hour care. However, being 18 he is now five feet seven inches tall and weighs almost ten stone, I'm sure those of you who have a son or daughter of a similar age will appreciate that as he is not always completely mobile this can cause difficulties in handling him especially as he is not at the hoisting stage.

We were in receipt of direct payments from the age of 17 and I was told these would automatically transfer over to adult services, unfortunately not for us. Firstly our new adult social worker (care manager) was appointed two months before Nathan's birthday and was completely new to the post and the area so was not aware she had to inform the direct payments service. The money stopped on his birthday last November. After many emails and badgering we were finally awarded a new number of hours per week in March at the end of the financial year. We were lucky as Nathan's P.A. was happy to work for no money until it came through and we could give her back pay. We were lucky as the money had previously been paid into an account in my husband's name, this was allowed to continue which is the first time the department had done this. Previously when adult services are involved the money had to go into an account in the recipient's name and as Nathan cannot write that would not be possible.

Together with the finance for direct payments we received a sum of money for respite care without yet being given any indication of where this respite might be found. This is one of the ongoing problems which we are trying to sort out with the help of Mencap. Unfortunately as Nathan is still mobile it rules out some of the possibilities because he would require 1:1 or even 2:1 care. Also he is no longer able to go to any of the centres helping people towards independent living. We have yet to find that elusive place that caters for his needs.

Nathan's medical needs now all come under adult services so we have had to meet a new group of professionals most of whom have little or no idea about the disease and require us to explain all his problems. One of the positives is that we transferred consultants and now attend The National Hospital for neurological diseases. The consultant was extremely nice and helpful. He conducted some tests on Nathan and thoroughly checked him over which means more medication but it is very reassuring. We did have some problems but these were to do with transport to the hospital but after many phone calls to Nathan's GP and to the hospital it has been sorted out.

Some of the outside agencies involved in Nathan's care have been accessed through school/college and where this is the case such as speech therapists etc we have had a lot of help from college and are extremely grateful.

Where do we go from here? We still have to find a replacement for Demelza House so that we can have some respite knowing that Nathan is being cared for properly. Also the question of what happens when Nathan is too old to go to college has yet to be answered, especially now as on becoming an adult he has to pay for or make a contribution towards the costs of a lot of the services. He needs to be around his peers so we will keep looking and pressing people to find a solution. Luckily he has just over a year to go before he has to leave and there are others in the same position although they do not have the same disease, so hopefully we will find somewhere. **Janet Gremo, mum to Nathan (MPS III)**

Bobby's Story...

Bobby is our only child; he was born a healthy baby (9lb 5oz) despite a difficult labour! He hit all his milestones easily until he reached the age of 2. He's always been very boisterous but was very delayed with his speech and fine motor skills. His gross motor skills were fantastic and still are now, he is a fantastic footballer. He was assessed and had hearing tests. He had grommets put in and his adenoids removed because of his frequent colds.

The paediatrician noticed his large liver and he was tested for Soto's syndrome, which was the start of the gene testing. We were absolutely distraught. When the tests came back negative we were over the moon, little did we know we now would have seen Soto's as good news. In 2008 he was fitted with hearing aids which I naively believed was the be all and end all of his problems, until the Neurologist told us otherwise. He told us he strongly believed Bobby had a storage disorder. We did not know what this meant nor did we realise the seriousness of what he had told us.

Bobby was diagnosed with MPS III Type A on 11th December 2008, soon after his 4th birthday. That day we were told they were 95% sure he had MPS III, Sanfilippo Disease and it would be 3 weeks before it was confirmed. I felt surprisingly composed when we left the hospital, considering we had only heard the term MPS a few days before. Once we were home it was a different story.

For the next two days it was like we were grieving uncontrollably, I felt ill with sadness and couldn't even eat. For any parent the most unbearable thing is to be faced with losing your child. You feel like there will never be a time in your life when you can be happy again or even bring yourself to smile. Then Bobby's Dad and I sat down and had a chat. We realised he's not been taken from us yet and who knows what the future holds. It is up to us to give him the best childhood / life we can, we do not want to look back with any regrets of being depressed and miserable when now is the time to enjoy him.

Bobby is such a happy little boy and he loves his life and everyone around him. The only saving grace with his condition is that he is not aware of it. He has no idea or understanding that he's different. He has lots of friends and everyone who meets Bobby loves him, he's so playful and energetic and always smiling.

The first two weeks were a bit of a blur. Friends and family rallied round and were very supportive; they arranged a weekend away for us and to top it off Bobby's dad proposed! We realised we shouldn't put anything off, we wanted Bobby to be part of our special day so we married in August 2009 with Bobby as our page boy, it was a fabulous day and Bobby loved it as he always enjoys a party!

Finally, the three weeks were up and we returned to the hospital for the confirmed results. This time we went armed with our list of questions and both sets of grandparents. At least that would be four less people to repeat the outcome to! We'd done what we shouldn't have and looked far too much on the internet upsetting ourselves even more, so we decided we only wanted to know the important facts for now. The diagnosis was confirmed and we were given lots of information and told about the MPS Society.

I then decided I wanted to do something, it was getting a bit tedious having to explain to everyone in as simple terms as possible what MPS is and what it means for Bobby. I also felt frustrated being the bearer of bad news all the time and you start softening up the details a little so as not to upset people or further upset yourself. A lot of people seemed to get very confused or were unable to grasp the seriousness of what Bobby has. I ordered lots of booklets from the Society and handed those out to friends and relatives, we all purchased wristbands / key rings etc and then we decided, what better way of promoting awareness than a bit of fundraising!

I work for a Law firm called Norton Rose and a work colleague there suggested doing a sponsored run. By this time I felt ready to let everyone at work know about Bobby's condition and the response was amazing. The compassion and interest people have shown is fantastic. It was hard for me to decide whether to tell people or not as I didn't want them to feel I was after sympathy as I'm not. I want everyone to be aware of these diseases, otherwise how will people know to give money? I had never heard of MPS, neither had my doctor and I have found there is a minority of people whose ignorance of Bobby's behaviour is infuriating and I don't always manage to bite my tongue. But I am now starting to feel happy and safe in the knowledge that Bobby is unaware of their judgement so therefore it shouldn't bother me. Days he is able to wear his hearing aids I find strangers are more tolerant and somewhat intrigued by Bobby as they have a visual prompt that he has some form of Special Needs. So I think if I can just spread the word to the few people I will meet in my lifetime I will feel I have helped another family who are yet to face what we have.

A few of us signed up for the London 10k in July 2009. Having never run this distance before I was slightly worried, but soon I started enjoying the training. Very quickly the list of people who wanted to run with us grew until there were 9 of us. I was overwhelmed by how generous everyone was with their donations; even people I hadn't spoken to for years were sending us such lovely messages and more than happy to donate. We were so happy; so many people came up on the day to cheer us on. We all wore matching outfits with pictures of Bobby and the MPS logo on our t-shirts (with MPS Society's consent of course!) and I couldn't have prepared myself for the emotion I felt when I crossed that finish line. We all felt like we'd really achieved something, wearing the t-shirts was fantastic so many people were coming up to read them and asking questions and we even met two other runners who were also fundraising for the MPS Society. It was such a lovely day, we have fantastic memories and Bobby wore my medal all the way home. Between all of us we raised almost £6,000.

Since then the Practice Director of Norton Rose has set up a Charity Committee for my department (Finance) where a group of volunteers meet every so often to discuss fundraising ideas for the MPS Society. We have had lots of successful events, most notably the Golf Day which raised over £4,000. The amount of effort the organisers went to was astounding; there was very impressive attendance and lots of competitions, raffles and auctions. Bobby won the prize for 'Best Man There'!

So the Finance Department were now aware of Bobby's condition and it was raised at the Firm's Charity Committee meeting where it was agreed they would hold a dress down day. I drafted an email briefly explaining MPS and also explaining how the support and research funded by the Society could not continue without ongoing donations. The dress down day was met with enthusiasm by all, as well as the £1 per person donation quite a few employees made additional donations and the messages of support were very heart warming. The Firm's Charitable Foundation made the amount collected up to £2,000.

Since then we have also had a Christmas raffle and a silent auction of signed X Factor pictures as me and Bobby were lucky enough to meet the 2009 contestants through a family friend. One group of colleagues also did a sponsored cycle.

I could never have expected the support I have received from my friends and colleagues. They are always happy to take on a new challenge and always have suggestions of new ways to fundraise, they really have been amazing.

Another important factor we have had to consider carefully is our home. Bobby is so clumsy and now we are aware that this is an attribute he will not grow out of. We have had to 'Bobby-proof' our home! It is all open plan with no steps, sharp edges or even photo frames within reach. However, our main priority was the garden. Bobby lives for playing outdoors, if he's frustrated or upset you can open that backdoor and he's a happy little soul again. We are lucky to have a large garden but there were lots of concrete steps, the fences were unsafe and Bobby had fallen on plenty of occasions, one time needing stitches in his head as he never puts his hands out to break his fall.

Bobby's Nanny took it upon herself to hold a fundraising night at a local restaurant to get works on the garden started. The concrete was removed and new fences fitted. During this time we were just planning to have the garden levelled and turfed to make it safe. Then my cousin contacted me. He owns a sports surfacing company and offered to Astroturf the whole garden as a gift to Bobby. We were ecstatic to say the least! The works were completed 2 weeks ago. Not only is there shock absorbency when he falls, but he no longer gets covered in mud and he can continue to play out there all year round. Bobby's trampoline has been dug into the ground so he can use it as and when he pleases (which is most of the day!) with no hazards. It really was the best gift anyone could have given Bobby. We couldn't thank my cousin and his wife enough, but they told us that seeing Bobby's obvious happiness with the new garden was all the appreciation they needed. The gratitude we feel towards everyone involved is beyond words.

The reason why all this has happened is that everyone who meets Bobby is touched by him. He is such a character! Bobby is such an accepting little boy, he just gets on with life, always happy. He brings smiles and laughter to our faces every single day. To start with you wonder, why us? But Bobby is ours for a reason, he truly is a gift. We have learnt so much from him in the small amount of time we have had him. I have felt things I didn't know I could feel, he has made me so much stronger and everything we do is with him in mind.

A large group of us are now planning to run a half marathon in September, who knows maybe the marathon or 3 peaks challenge next, they have all been discussed! Most importantly though we like to believe that if Bobby understood what we have done and what we still plan to achieve, that he would be extremely proud of us and understand the depth of our love for him.

Liz Gill, Bobby's Mum

It took us five years to conceive, but now we know why? It was God's way to make sure we were ready

Reporter Lee Marlow talks to a Leicester couple who 'burn with pride' for a daughter living with the rare Morquio's syndrome

You might not have heard of Morquio Syndrome. In fact, you have to scour the net to find a crumb of detail on this life-changing, debilitating condition. In all of Britain, only a limited number of people have it and Aisha Seedat is one of them.

A 12-year-old from Leicester, Aisha's life is a daily battle against this genetic disease which affects her growth, bone development, ligaments, spine, heart and various other internal organs and has left her in a wheelchair.

Aisha's life has been one long, ongoing medical treatment, but she bears her condition lightly.

"She is such a happy and cheerful girl," says mum Asma "We are so proud of her."

Today, the pupils at Manor High School in Oadby will be wearing shoes of their choice to raise awareness of the MPS Society and help their friend, Aisha.

This is the Seedat family's story; about the child they longed for whose life has been blighted by a disease that even the experts know little about.

Mum (Asma): "We both wanted children but it took five years for us to conceive with Aisha. It seemed like an eternity, but now we know why – she was a gift from God to us. He had to make sure we were ready. I was so... wary. All the time, we prayed and hoped for the best. Unfortunately, it wasn't to be."

Dad (Sharif): "Aisha was born with a protruding sternum. I was so thrilled at the birth, so pleased to see her – It was such a mix of emotions. I could see something wasn't right. I saw the look on the faces of the nurses and that affirmed it. They looked horrified. No-one needed to say anything to me."

Asma: "Aisha was the first girl to be born in our extended family for many years. We were so happy – and yet... we could see she was ill. I felt weak but I wanted to be positive. But, inside, I was contorted with grief."

Sharif: "We came home and we didn't know what to feel. We finally had our daughter... but we could see she was ill. We just didn't know how ill she was."

Asma: "I remember when I was giving Aisha a bath (she was just nine days old) and she stopped breathing. We rushed her into A&E and they didn't know why she stopped breathing. They told me I must have had the bath water too cold… But I hadn't. I knew that wasn't right."

Finally, after a very long time, the hospital put the Seedat family in touch with Dr Nichani at the Leicester Royal Infirmary. He ordered an extensive list of tests. These confirmed that little Aisha had an MPS disorder and later confirmed it was Morquio Syndrome, an inherited condition with a long list of serious symptoms including deformities of the bones, joints and ligaments and problems with internal organs also affecting the respiratory system.

Asma: “It’s a Metabolic disorder. We didn’t know it, we were both devastated.”

A few years later, Asma became pregnant again. She was delighted, yet terrified.

Asma: “I think I went into a kind of depression. I knew there was a strong possibility our unborn child could also have Morquio”

Saffiya was born at the Leicester Royal Infirmary in January 2001. They did a series of tests after she was born. Saffiya did not have Morquio syndrome.

Asma: “We were very relieved.”

As Aisha was developing meanwhile we found her type of Morquio was quite severe.

Asma: “Aisha could walk. She could dance. But gradually, until last year, she lost the ability to do all of these things. We knew it can happen (but not with our Aisha). It doesn’t make it any easier to take when it does. I have videos of her dancing and skipping. I can’t watch them today. I can’t even look at the photos. They’re too upsetting. But she can talk. Oh my, can she talk! I never expected so many questions. Her wit and intelligence amaze me constantly. Her reaction to her disability has always made me burn with pride.”

Sharif: “When she was five, she had a major operation to secure her spinal cord. For the next nine months, she had to wear a halo traction, one of those things that keep your head still but look like they’re from a horror movie. There were four screws that held the halo contraption to her skull. I had to tighten the screws of that traction once a week. It was always a frightful process.”

Asma: “She is only 92cms tall. The Morquio restricts her growth, but her internal organs are still the same size. That causes all sorts of problems for her.”

Sharif: “The prognosis is unknown. There’s a new treatment undergoing Clinical Trials – ERT, enzyme replacement therapy – which provides the enzyme which Aisha’s body can’t give her. It won’t cure her. But it will alleviate some of her symptoms. It sounds brilliant, but they’ve told us she’s not ready for that yet – This is due to some forthcoming neurosurgical operations that she needs again.”

Asma: “We don’t think too much about the future. We go to Morquio conferences and we know one sufferer who’s 42. So you just don’t know do you?”

“We don’t talk about it. Maybe we should. But we treasure every day we have with Aisha. She’s a gift from God.”

Sharif: “Even though we have been living with this for 12 years, I don’t think we have accepted her condition. We make room for it. We deal with it. But we haven’t really accepted it.”

Asma: “Aisha keeps us going. She’s so positive. She goes to Manor High, a mainstream school in Oadby and also attends Ashfield School (for children with physical difficulties) in Leicester on a weekly basis. She’s very bubbly, chirpy, full of life. She’s 12 now and I can see her changing as she moves into adolescence. She’s a beautiful girl and we love her dearly. I just wish she didn’t have to go through all she has to go through.”

This article appears courtesy of the Leicester Mercury. **www.thisisleicestershire.co.uk**

Raising Awareness

Birmingham Children’s Hospital
Since 2007 we have hosted a number of LSD study days at Birmingham Children’s Hospital and other major hospitals within the West Midlands to raise awareness of MPS and other LSD disorders. However in 2009 we held a National LSD Study Day when we invited a representative from each of the NCG centres to give a presentation in order to share experiences across the country. NCG centres were invited to nominate local delegates to raise awareness nationally. This study day was universally very well received and led to the organisation of a further event which was held in the centre of Birmingham during May this year.

Again speakers were invited from each NCG centre in order to provide nursing and medical representation from across the service as a whole. The timing for the event was selected to coincide with the National MPS Awareness Day on Friday 14th May. The event was very well-attended by over 70 delegates who arrived from across the country including from Kent, Devon and Newcastle on Tyne. The feedback received has been very positive and it is planned to hold a similar event in 2011. The date of Friday 13th. May has been selected to coincide once again with MPS National Awareness Day next year.

Catherine Stewart, (nee Little), Clinical Nurse Specialist, Birmingham Children’s Hospital

Newcastle Clinic

22 April 2010

My first Newcastle clinic took place on 22nd April. It was a week after the volcanic eruption and I was travelling on a train to meet with our MPS families.

It was the first clinic in a new hospital building but I managed to find the clinic rooms with Dr Rylance and Dr Wraith. The families found us too. Daniel and his mum were excited to be coming to our event in Camelot. Laura was smiling a lot and talking about her college. Luke was the last one to come and he was interested in anything but the movie 'Robots' which was played in the waiting area.

The clinic went well and soon I was back on a train to Manchester to attend the BMT Clinic the next day.

Thank you to Dr Rylance and Dr Wraith, the team of nurses in outpatients for their support and to the families who made me feel so welcome. **Jolanta Turz**

Manchester BMT Clinic

23 April 2010

The whole week was very sunny and I was happy to discover that Manchester can be sunny too from time to time!

It was a mixed age clinic so the youngest child was 4 and the oldest 11 years old but the age range didn't mattered. It looked as if the competition for the day was to build the highest tower from the big lego bricks. There were children who did it on their own, some worked together (good team work Cody and Melissa!) and some with parents, who were really into it. When the tower was up, the next most exciting thing was to push it, kick it or hit it down with lots of giggles. The only thing I can say is 'Let's keep it going!'

I would like to thank everybody who helped with tiding up! **Jolanta Turz**

Manchester BMT Clinic

30 April 2010

After a week I was back in Manchester. The age gap at this BMT clinic was even bigger.

The younger one was Sonny, who was smiling all the time. Then there were: Demi-Leigh demonstrating Irish dances in her ballet shoes, Steven attempting to destroy every lego tower he built, Harvey playing with his Thomas engine, Holly playing in a play house and Kiera who couldn't wait to go camping over the weekend.

The oldest one was Emma and she had some exciting news. Emma just passed the theory test for her driving licence and she will start the driving lessons soon. We wish you good luck with the driving test!

Dear Manchester Team, thank you for another two successful clinics! **Jolanta Turz**

Great Ormond Street Clinic

3 June 2010

On one of the hottest days of the year Jolanta and I braved the rush hour traffic and travelled to GOSH for the MPS III clinic. As always it was a hectic clinic and Dr Vellodi, Niamh, Victoria, Sonia and Michelle were all kept very busy. The waiting room was a bustle with parents and children and as the clinic came to an end, the toys and books packed away, it was hard to believe we were in the same place.

On behalf of the Society, Jolanta and I would like to thank Dr Vellodi and the team for another successful clinic.

For me this clinic was particularly special as it was my last one. I would like to thank everyone at GOSH who has made me feel so welcome over the last few years, I shall miss you all very much. **Linda Warner**

Here are a selection of photos from the clinics featured on these pages.

Sibling Week 2010

From 12th – 16th April 2010 two of our MPS staff members, Sophie and Jolanta headed off to the PGL Camp at Windmill Hill in East Sussex… not for a quiet girl's holiday but for a jam-packed week of fun activities for the MPS Siblings Week funded by BBC Children in Need.

This week of structured fun was arranged for children who have brothers and sisters with an MPS or a related disease that are 9-17 years of age. These breaks away from home enable these siblings to have fun with other youngsters in similar circumstances, meet new friends and have new experiences.

As well as various activities at the PGL camp ranging from BMXing, raft building, Zip wires and ludicrous amounts of fun on a giant swing and trapeze, the group also spent a day in France where everyone was taken on a tour of the chocolate factory before going into Boulogne to see the centre of the town and for a bite to eat.

Everyone left PGL camp for the Euro tunnel at 8am in order to catch their Eurostar Train… the one and only aspect of the trip that the children mutually disliked!

"I really enjoyed visiting PGL and France. I thought it was really fun. My favourite activity was the giant swing. It was nice being with people my own age and I made some new friends. I just didn't like having to get up so early!" – Becci

After this early start the group arrived in France at 12.30 and headed to the chocolate factory ready for their tour to commence at 13:00.

"This week I have really enjoyed myself and made lots of new friends. I really enjoyed my trip to France and the chocolate factory was pure heaven! Getting up that early in the morning is definitely not my favourite thing as I am not an early morning person!" – Mollie

After the chocolate factory experience everyone headed to the centre of Boulogne where they had a tour of the town, and after a quick bite to eat the group then made their way back to PGL Camp.

We were delighted to hear back from the children that attended this sibling week. Here are a few of their comments which sum up the week:

"I really loved this week because we did the Zip Wire, BMXing and had a disco!" – Mia

"I loved the week at PGL. It was jam-packed with exciting things to do. My favourite activity was the Zip Wire because it felt like you were flying. BMX was great too though! I would definitely come again if I had the chance." – Charli

"It was fab! This week at PGL has been so much fun because I have made new BFF's (Best Friends Forever). My favourite activity was BMXing because at the beginning I was rubbish but now I'm really good at it!" – Freya

"I enjoyed PGL. The activities were good but the best thing was seeing Emma Stuart and Emma Hooper again. We both enjoyed PGL. We had lots of giggles and really enjoyed the giant swing, the trapeze and many others. We loved seeing everyone again. We want to say thanks to everyone who organised it. It was FAB!" - Emma & Emma

"I really enjoyed the week at PGL and I made lots of new friends. My favourite activity was the big swing along with the Zip Wire. I hope I can go again next year." – Amy

The MPS Society would like to thank the support that was provided by PGL Staff and the experienced MPS Volunteers who helped to make this week such a success.

All Ireland
MPS and Fabry Conference

Between 14 - 16th May 2010 the MPS Society held their first All Ireland MPS and Fabry Conference at the Hilton Templepatrick.

There were two separate conferences taking place, one focusing purely on Mucopolysaccharide diseases and the other focusing solely on Fabry disease.

These conferences offered individuals, parents, partners, carers and professionals the opportunity to hear state of the art talks on clinical management for those affected by MPS and Fabry. It also gave everyone the opportunity to hear other members' personal experiences.

Sophie Thomas, Senior Advocacy Support Officer for the MPS Society says:

"Our aim in organising these conferences is to offer all who attend the opportunity to meet and talk informally with professionals and to share their own experiences with others whilst learning about new treatments and any advancement in research. The childcare that we provide to coincide with these events not only allows the adults to make the most of their time at the conference but allows children and vulnerable adults with MPS and related diseases and their siblings the opportunity to liaise with others in similar circumstances. An important aspect for both the children and adults alike"

The childcare programme was packed with entertainment and events that kicked off on the Friday evening with Captain Franko providing family entertainment after dinner. His balloon modeling and fun filled magic show were enjoyed by the whole family.

Saturday consisted of a fun filled day at Belfast Zoo where Jenny the elephant celebrated her 50th Birthday in style with a super sized birthday cake to mark the occasion. The children also saw a variety of animals such as giraffes, elephants, bears, monkeys, kangaroos, lions, penguins and many, many more.

Upon their return the children attended a grand balloon release, but not before our colleague Gina Page sprung into action when she was informed that we had to receive the all clear from the aviation authorities due to the hotel's location on the flight path!

The Gala Dinner took place on Saturday evening where attendees from both the MPS and Fabry conference amalgamated for a sit down dinner which gave everyone the opportunity to relax and take on the information from the day.

We all hope that those who attended this event had far less stress during their journeys home in comparison to our colleagues Gina, Sophie Thomas and Chief Executive Christine Lavery and various professionals that attended the conference.

With thanks to that persistent volcanic ash cloud there were no planes, just trains, automobiles and the odd ferry to rely on. Our MPS colleagues were either stranded or in transit for up to ten hours longer than expected. Even with the unexpected dramas that unfolded the All Ireland MPS and Fabry Conference was a success.

The MPS Society would like to thank the professionals who contributed their valuable time, as well as the volunteers for the care that they provided to the children and vulnerable adults over the duration of this conference.

Family Weekend at Camelot

Over the May Bank Holiday Weekend the MPS Society held their Family Weekend at Camelot.

On the Saturday afternoon families began to arrive at the Park Hall Hotel, with various stories of their eventful journeys through the ever predictable, ever problematic bank holiday traffic!

After settling into their rooms everyone met in the Park Suite for a buffet dinner where the Children's Entertainer wowed both the children and adults alike. But it was later that evening when the party atmosphere really kicked in resulting in a lengthy conga line and a seamless rendition of the YMCA!

After a hearty breakfast at Brookes Restaurant on the Sunday morning everyone set off to the Camelot Theme Park where the young, and the young at heart, were able to enjoy all the rides on offer. It was a busy day for all, but the fun didn't stop there.

After a short breather it was time to commence with the activities planned for the night ahead.

The children attended their own Gala Tea where they were supported by volunteers and entertained by one of the resident magicians.

As well as having the opportunity to enter a colouring competition to meet King Arthur, the children were also given the opportunity to design a Christmas card for entry into the MPS Society's very own draw. There were plenty of other arts and craft projects on offer as well as a DVD to keep them entertained and amused.

Whilst the children enjoyed their Gala Tea, the adults attended the AGM and Gala Dinner which was held in the Medieval Banqueting Hall. Not to be outshone by the children, the adults were entertained by jugglers and even witnessed a medieval fight scene, which was interrupted by the fire alarm and a subsequent evacuation from the hotel due to an electrical fault in the leisure complex…it was no hoax!

All adults and children were evacuated safely and after what seemed like an eternity standing in the chilly hotel car park, we were finally allowed back in.

The children continued with their Gala Tea and the adults Gala Dinner resumed, ironically with fire eaters on hand to entertain!

A little while later the volunteers brought the children to the forecourt outside the Medieval Banqueting Hall to be reunited with their parents and to meet one of Camelot's knights and his trusty steed.

Even after all of this the party continued with a family disco and pass the parcel, which saw the children enjoying the fun bit of unwrapping the layers and eating the sweets and the parents having to do the forfeits… the chicken impersonators were second to none and we do believe that we have found a few X Factor contenders!

The winners of the Camelot colouring competition were announced later in the evening. Huge Congratulations went out to Kaiden Lyon aged 3 years and Sophie Thompson aged 7 years. Both were given the opportunity to meet King Arthur personally the following day.

The following day everyone had the opportunity to have another day at Camelot Theme Park before making their way home. A marvellous time was had by all.

Once again the MPS Society would like to thank the invaluable volunteers that provided childcare throughout the weekend.

MPS ANNUAL GENERAL MEETING 2010
The Annual General Meeting of the Society for Mucopolysaccharide Diseases took place at the Park Hall Hotel, Chorley, Lancashire, at 7pm on Sunday 30 May 2010. 8 Trustees and another 58 members were present at the AGM.

The Chairman, Barry Wilson, opened the meeting and welcomed those present. The Chairman stated that apologies had been received from Judy Holroyd, Tim Summerton, Faith Parrott and Peter Conlin.

The minutes of the Annual General Meeting held on Sunday 28 June 2009 were published in the Summer/ Autumn 2009 edition of the MPS Magazine, and were distributed in advance to those members present at the meeting. The minutes were accepted as true and accurate.

The Chairman presented the Trustees' Report. This is published in the MPS Annual Report and Accounts for the year ending 31 October 2009.

The Treasurer, Judith Evans, presented the Statement of Accounts for the financial year ending 31 October 2009, the details of which are also to be found in the Society's latest annual report. It was proposed and seconded that the auditors McLintocks, Chartered Accountants, Chester, be appointed the Society's auditors for the financial year ending 31 October 2010.

Trustee nominations took place. As there were four nominations and three places vacant on the Management Committee, members present were asked to vote. The votes were verified and announced during the Gala Dinner and Judith Evans, Bryan Winchester and Wilma Robins were elected.

The Chairman thanked everyone for attending and the meeting closed at 7.20pm.

Barry Wilson, Chairman of Trustees
On behalf of the Management Committee
30 May 2010

We are grateful for a contribution from the Hilton in the Community Foundation, The Clover Trust and The Riverside Charitable Trust to the costs of this Family Weekend.

Living with Fabry

Ian continues his series of articles written for the MPS magazine sharing his experiences of living with Fabry

This month I have found it hard to write but I will do my best. I think as people with Fabry enhanced bodies we have a double edged sword. Yes, we suffer the difficulties of living with the condition and many of us are at different stages and also we have different experiences with the condition. But one thing we all have in common is the fact that we DO NOT ACCEPT THE WORD NO.

Maybe this in some ways makes us stronger. As we are used to finding things a little tough, we can deal with being knocked sideways and our inner strength helps us to keep getting up. It just takes us a bit longer as we get older.

Three weeks ago a friend of mine (Tim) visited. I have known Tim for close to 20 years and he has a passion for travel just as I do. So, when he received a marketing email from Keycap we were straight on to the website to see what we could get. He had to go but my fire was lit. I continued to look and eventually I was on the phone and securing a week in France for £139. I was happy but the next thing was to convince Sharron (Tim's wife) that it was a good idea to go, and for Tim to drive.

All parties agreed and, come Wednesday morning, away to Dover we went. We were heading to a place called Guines, between Calais and Boulogne. It was a place I had been to before but also I know the area quite well down to Paris having worked there with coaches and trucks. This time I wasn't driving but new challenges were ahead.

This was to be my first holiday after my stroke. In fact, it was the first real experience of travel since the big day but Ann, my wife, needed a break as much as me, if not more. By mid afternoon we arrived at our accommodation and the courier said the weather was to be fantastic and it was.

As I said I know the area quite well so there were things I wanted to show every one. Boulogne, for instance, was a big staging port for Napoleon to invade the UK. It was also the place where the v1 and v2 rockets were flown in World War II and it is also an area that saw severe fighting in World War I. In fact at Etaples they have one of the biggest cemeteries in France with the dead of those British soldiers who died in that area with 12,000 graves.

Near there is Le Touquet-Paris-Plage which has beautiful beaches were we spent some time, after which we went into town where the main street was closed to traffic. This town is where a lot of people who live in Paris have their second homes and spend the weekends in Le Touquet so it was full of boutiques and pretty people. Guess what though, I spent most of the time looking at peoples' waists and knees as for the first time I was in my wheelchair.

To those who have always used to them or have used them for a long time I take my hat off to you. I feel the wheelchair will be a permanent aspect of my life from now on. I can get about short distances under my own power but my legs have a habit of giving way so I struggle to go to the shops (what a shame) but Ann has found it has benefits when I do go as she can park me up and go to look at what she wants! So using the wheel chair is something that is going to take time to get used to, more so mentally than anything else.

Just a small note before I finish. I was at MPS House yesterday for a Fabry Focus Group (see next page for more information) along with four other Fabry patients, four of whom I knew and one was new to me. We had a great time and were looked after very well. However it really is sad that we were able to see by talking that there were differences in the way we are treated in different parts of the UK.

So how do we hope to change things if we do not make our voice heard? It is true that we all have things to do and going to Amersham is a long way, well it certainly is from North Wales but believe me when I say it is worth it.
Ian Hedgecock

EUROPEAN MEDICINES AGENCY GIVES NEW TEMPORARY TREATMENT RECOMMENDATIONS FABRAZYME.
Supply shortages for Fabrazyme to last longer than expected

The European Medicines Agency's Committee for Medicinal Products for Human Use (CHMP) has been informed by Genzyme that the supply shortages for Fabrazyme are expected to continue at least until the end of September 2010, because of a new manufacturing problem.

For Fabrazyme, the Committee decided to revise the recommendations made on 25 September 2009, since, based on information supplied by Genzyme, at least 12% of patients on the reduced Fabrazyme dose regimen have already experienced a worsening of their disease. For such patients, the CHMP has recommended that physicians should either consider restarting the original treatment with the full dose of Fabrazyme or switching to an alternative treatment, such as Replagal.

Given the new information, the temporary treatment recommendations for Fabrazyme are as follows:

- Children and adolescents (under 18 years) should receive Fabrazyme according to the standard dose (1.00mg/kg) and frquency of one infusion every two weeks.
- Adult patients already treated / stabilised may receive Fabrazyme with an adjusted dose of 0.3mg/kg as a maintenance dose every two weeks.
- All patients, especially those with adjusted dose regimens, should be under close clinical surveilannce. A full medical examination, including all relevant clinical parameters, should be performed every two months. It is of the utmost importance to monitor the plasma GL-3 or urinary GL-3 levels, as for the moment the GL-3 level is the most sensitive parameter.
- For patients on the reduced dose who demonstrate a deterioration of the disease, physicians should consider restarting the original treatment with the full dose of Fabrazyme or switching to an alternative treatment, such as Replagal.

These recommendations are temporary and do not change the currently approved product information for Fabrazyme.

The full European Medicines Agency (EMA) press release can be found on www.ema.europa.eu. **Christine Lavery**

Fabry Focus Group

The MPS Society is proud to be assisting Miss. Alison Whann in her research study entitled 'Exploring the Needs and Experiences of Individuals Diagnosed with Fabry Disease – A Comparative Study.'

Alison Whann is a student at Cardiff University studying for a Masters in Genetic Counselling and as part of her Masters degree will be exploring the needs and experiences of individuals diagnosed with Fabry Disease. Her aim is to ascertain which services are routinely accessed by individuals with Fabry Disease; whether or not individuals feel adequately informed about their condition; and whether or not the information that they been provided with has aided them in key life decision making.

As the title indicates, the research conducted will be in the form of a comparative study. The aim of this is to compare the needs of experiences of individuals with Fabry Disease from different parts of the UK to highlight differences in service provision and levels of client satisfaction.

Alison says:
"Fabry disease is a condition that I have had a longstanding interest in; so when asked to complete a research project as part of my Masters course I knew instantly that Fabry Disease would be the focus of my study.

When I began researching the condition I noticed that much of the literature on Fabry disease is about diagnosis, enzyme treatment and other medical advances; and that little research had been carried out in relation to the experiences of individuals diagnosed with the condition. I therefore chose to focus my study on the needs and experiences of individuals diagnosed with Fabry disease; and to add an extra dimension to the project I felt that it would be interesting to determine if the experiences of patients vary depending on which part of the UK they live in.

At the end of my project I hope to paint a clearer picture of what it is like for individuals diagnosed with Fabry disease. I also hope that in speaking to individuals diagnosed with the condition I will be able to identify any unmet needs that could be addressed with the overall aim of improving patient experience"

Part of the study involved a Focus Group that took place at MPS House on Saturday 12th June 2010. This gave all who attended the opportunity to have a face to face discussion on the care that they have received in the UK.

A second Focus Group is planned to take place in Ireland later on this summer which we wish Alison the very best of luck with. The MPS Society will continue to support her for the duration of this study. **Kirsty Wyatt**

Fabry disease in children:
emerging data from the Fabry Registry

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

Hopkin RJ, Bissler J, Banikazemi M et al. Characterization of Fabry disease in 352 pediatric patients in the Fabry Registry. Pediatr Res 2008; 64: 550-5.

Fabry disease has not been well-studied in children, partly due to its rarity and the difficulty in making an early diagnosis. However, the gene defect that is ultimately responsible for Fabry disease symptoms is present at birth, and the problems associated with this disorder often start at an young age. Moreover, the diagnosis of Fabry disease is often delayed, possibly because symptoms and complications are not specific to this disorder and so are often mistaken for similar problems caused by more common diseases [1]. The Fabry Registry – an ongoing global observational registry for monitoring people with this condition – allows large groups of people, from around the World, with Fabry disease to be monitored (www.fabryregistry.com) [2]. Here, we provide information on children enrolled in the Registry [3]. The data included in analyses are before their treatment with enzyme replacement therapy (ERT), and so represent the natural, untreated course of Fabry disease in children. This information is important to understand the presentation, progression and severity of the disease, and to examine how well it is diagnosed and managed, in order to improve the lives of children with Fabry disease.

This study includes 352 children under 18 years of age at enrollment in the Registry, of whom 194 (55%) were male and 158 (45%) were female. Children were diagnosed with Fabry disease at a relatively young age of 9 years for both boys and girls. At enrollment, more boys (77%) than girls (51%) had symptoms, and boys tended to have had symptoms earlier (from a median age of 6 years compared with 9 years in girls).

Symptoms reported by children at the time of enrollment in the Registry are shown in Figure 1. This shows that common symptoms are more frequent and occur at a younger age in boys than in girls.

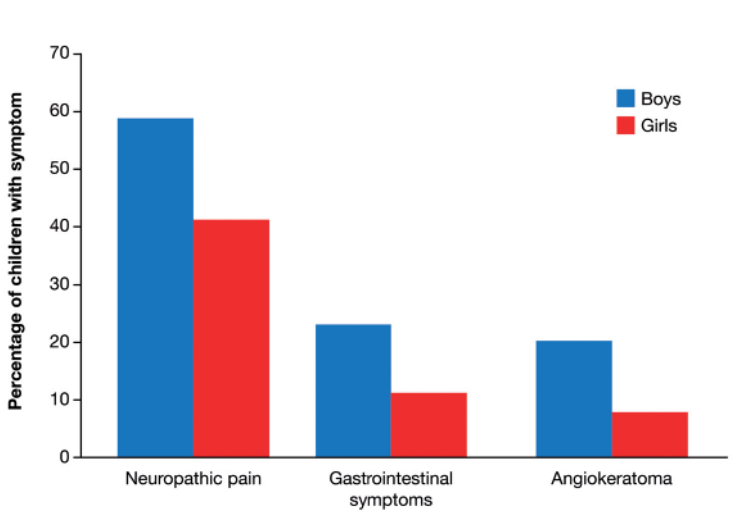
The most common symptom in children is a type of pain called neuropathic pain (which can be long-term or experienced in short, severe bursts, often affecting the hands and feet from where it can spread to other parts of the body). This pain is reported more often in boys (59%) than girls (41%) and at a younger age (median of 7 years vs 9 years, respectively)

More boys than girls had stomach pain and diarrhoea (gastrointestinal symptoms), but when later clinical assessments were included, a similar proportion of boys and girls had these symptoms. Gastrointestinal symptoms also happened at a younger age in boys than in girls (median of 5 years vs 10 years of age, respectively).

Angiokeratomas (small dark red- or purple-coloured raised spots on the skin) also affected more boys than girls (20% vs 8%), and at an earlier age in boys (median of 7 years vs 10 years of age).

Figure 1. The most common Fabry disease symptoms in children on enrollment in the Fabry Registry.

Figure 1.



Other, more serious manifestations of Fabry disease affecting the kidneys or the heart are far rarer at enrollment: two children had heart problems and one had poor kidney function. However, the follow-up clinical assessments detected more of these serious effects. For example:

- 32 children had problems with their heart valves (21 boys and 11 girls)
- 9 (7 boys and 2 girls) had problems with their heartbeat rhythm (an arrhythmia)
- 3 (2 boys and 1 girl) had an enlargement of the left pumping chamber of the heart (termed left ventricular hypertrophy or LVH).

Moreover, these effects were often reported at a young age (cardiac arrhythmias were reported in patients as young as 12 years). Thus an early diagnosis of heart problems, including the use of tests such as electrocardiograms (ECGs) and echocardiography, are essential for children with Fabry disease. Likewise, kidney function was severely affected in 3 of 144 patients who were tested, with 13 others showing some sign of potential kidney problems as indicated by protein being present in the urine. Therefore, the monitoring of potential kidney problems by using simple tests to monitor protein levels in urine may be useful in children with Fabry disease.

Children's growth and development were also monitored, and boys' height and weight were generally below what is normally expected in the general population. However, girls were generally of normal height but slightly heavier than population average values. These results may reflect differences in the way Fabry disease manifests in males than in females, although further research is required.

A questionnaire was used to measure quality of life, and these results compared with scores for the general U.S. population. Average scores for boys aged 14–18 years were generally lower (poorer quality of life) than normal in 7 of 8 questionnaire categories, whilst scores for girls aged 14–18 years were also lower than normal, but only in 2 of 8 categories. Moreover, a pain questionnaire showed that 53% of boys and 15% of girls had experienced severe or moderate pain during the previous day, but despite this most had never received any painkillers. Furthermore, only 62% of boys and 13% of girls in the Registry had ever had ERT, and of those who were symptomatic, 53% and 83% of boys and girls were not receiving ERT at the time of enrollment in the Registry.

In summary, children show early signs and symptoms of Fabry disease, with neuropathic pain and digestion problems representing an early and common burden in this group. Traditionally, as it is sex-linked, Fabry disease was thought to affect only males [4]. However, these data support the growing realisation that the disease affects girls as well as boys, though boys are more likely to have earlier and more severe symptoms than girls. Serious problems involving the heart and the kidneys become apparent in a proportion of children. Doctors need to be aware of this, to monitor for these effects, and be prepared to treat them as early as possible. Doctors should also be aware that neuropathic pain is very common at an early age in children with Fabry disease. Pain management in children with Fabry disease is poor, and should be improved. Moreover, further research on the effects of early intervention with enzyme replacement therapy is needed to determine whether this prevents or delays serious complications of Fabry disease.

References

1. Schiffmann R. Fabry disease. Pharmacol Ther 2009;122(1):65-77.
2. Eng CM, Fletcher J, Wilcox WR et al. Fabry disease: baseline medical characteristics of a cohort of 1765 males and females in the Fabry Registry. J Inherit Metab Dis 2007;30(2):184–92.
3. Hopkin RJ, Bissler J, Banikazemi M et al. Characterization of Fabry disease in 352 pediatric patients in the Fabry Registry. Pediatr Res 2008;64(5):550-5.
4. Desnick RJ, Ioannou IA, Eng CM. α-galactosidase A deficiency: Fabry disease. The Online Metabolic and Molecular Bases of Metabolic Disease 2001; Chapter 150: 3733–3774.

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

CLINICAL TRIAL UPDATE

BIOMARIN REPORTS ENCOURAGING PRELIMINARY DATA ON PHASE I/II ERT CLINICAL TRIAL FOR MPS IVA
Preliminary clinical data from the first 24 weeks of the study involving 12 weeks of enzyme at 0.1mg/kg and 12 weeks 1.0 mg/kg has been evaluated, and BioMarin plan to announce top-line results for the full 36 week study after completion of dosing at 2.0mg/kg in the second quarter of 2010.

Key Observations in the first 24 weeks of the study are:
- Keratan Sulfate (KS) levels fell within a few weeks of the start of therapy
- Improvements in 6 minute walk distance and 3 minute stair climb at 24 weeks are consistent with those observed with clinical studies for MPS I, MPS II and MPS VI.
- The frequency and severity of infusion reactions appear comparable to those observed with Naglazyme and Aldurazyme

Hank Fuchs, Chief Medical Officer at BioMarin stated *"Although still early, we are encouraged by these initial signals of efficacy of the GALNS enzyme replacement therapy for Morquio Disease. We feel encouraged by the reduction in Keratan Sulfate and improvements in walk distance and stair climb. Based on these results, we feel more confident about endurance as a primary endpoint for a Phase III trial and that a Phase III trial can be conducted as expeditiously as previous trials of enzyme replacement therapy. We plan to work closely with the FDA and other health authorities to finalise a Phase III protocol after completion of the current study and have increased confidence that we will initiate a Phase III registration - enabling programme by the fourth quarter of 2010 or the first quarter of 2011".*

RECRUITING SOON IN THE UNITED KINGDOM
Phase I / II Clinical trial 'Intrathecal Enzyme Replacement Therapy for MPSIIIA

Phase I /II Clinical trial of Enzyme Replacement Therapy for Alpha Mannosidosis

If you are interested in knowing more about these two clinical trials please do contact Christine Lavery **c.lavery@mpssociety.co.uk**
or Sophie Thomas **s.thomas@mpssociety.co.uk**

DNA BLOOD SPOT BANKS
Some of you may have read the Sunday Times article of 23 May 2010 regarding hospitals that have quietly created banks of DNA blood taken from millions of newborn babies without the informed consent of their parents. Freedom of Information requests to hospitals around Britain have established that the blood samples taken in heel-prick tests to screen for serious conditions, have been privately stored by parts of the NHS since 1984. According to guidance obtained by the Sunday Times, the DNA can be looked at by police, coroners and some medical researchers and the samples are identifiable by the name of the individual.

It is suggested that mothers of newborns are given a leaflet that academic experts say fails to make it clear the distinction between consent for vital clinical tests to safeguard a baby's health annd for the use of the baby's DNA in medical research and police enquiries. Do you believe this amounts to informed consent?

Government guidance says the bloodspots should be kept for at least five years, but it would appear hospital practice varies widely.

Central Manchester University Hospitals Trust has according to the Sunday Times, 1 million samples in storage dating from 1984 and a further 250,000 further samples are stored in the hospital's laboratory. Apparently they plan to store them indefinitely.

Cambridge University Hospitals Trust retain samples, according to the Sunday Times, for 18 years. Great Ormond Street hospital began storing samples in 1990 and preserves them for at least 20 years at a rate of 120,000 babies a year. Again according to the Sunday Times it has confirmed that it has occasionally handed samples to coroners but not police.

There are clearly differing views on this matter but as Newborn Screening becomes increasingly likely for at least some Lysosomal Storage Diseases your views are important to us.
- Do you think that there should be informed written consent for blood taken in the heel prick test of every baby in the United Kingdom?
- How long do you think the DNA from the heel prick test should be stored?
- Do you think DNA from the blood of a baby under going a heel prick test should be available to researchers without explicit consent?
- How do you feel about the possibility of the DNA taken from a baby's heel prick test being accessed by coroners or police?

If you have a view on this matter please email us at **mps@mpssociety.co.uk**

New LSD National Specialist Commissioning Service

The University Hospital Birmingham has been given Ministerial approval to be designated as a Lysosomal Storage Disease (LSD) Centre for adults as of 1 April 2010.

Dr Tarek Hiwot, the lead clinician for this new LSD service wrote: *"Dear Christine, I just wanted to thank you personally for your help in establishing an adult LSD service in Birmingham. Without your support this wouldn't have happened. I admire your dedication and willingness to help. The MPS Society is doing a terriffic job for our patients. You have done your bit now and now we have to deliver a world class LSD service. I promise we will do our best. Thank you again. Tarek"*

Editor's Note: We have invited Tarek and his team to write an article for the next MPS Magazine giving information about the service and the people who will be delivering it.

INTERNATIONAL SYMPOSIUM ON MUCOPOLYSACCHARIDE DISEASES
Adelaide, Australia, 23 - 27 June 2010

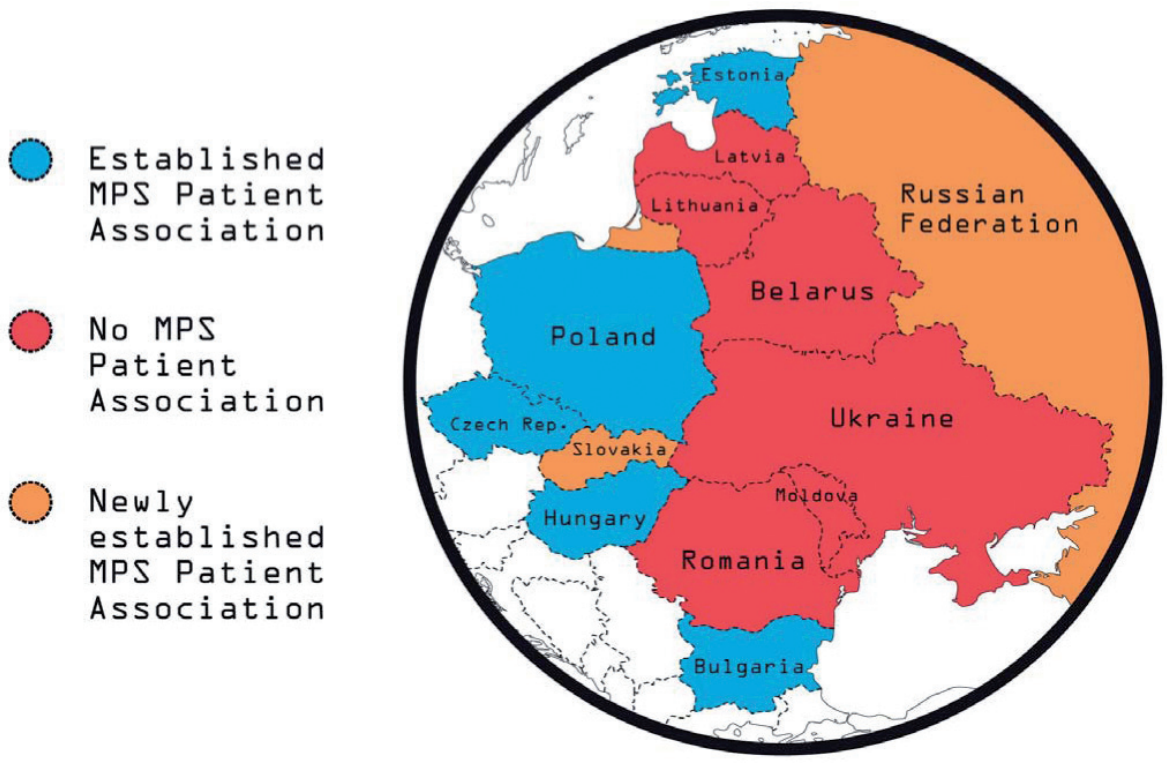
It has been an immense priviledge to accompany our six young adult ambassadors to the International Symposium on Mucopolysaccharide Diseases held in Adelaide at the end of June. Our six MPS ambassadors, Faye Longley (MPS IVA); Jibreel Arshad (MPS IVA); Lois Pack (MPS IH/S); Megan Rennoldson (Mannosidosis); Joanna Wilson-Smale and Roshini Nonis (sister of MPS III sufferer) along with two volunteer carers, Rosie Allen and David Murphy, and MPS staff team, Linda Warner, Jolanta Turz and I, all travelled out together on 19 June. We changed planes in Singapore before arriving in Adelaide on 21 June.

Apart from attending relevant conference sessions our young ambassadors enjoyed some local culture and had the opportunity to meet lots of children and adults with MPS and their families. Apart from saying what a huge success the Conference was I am not going to say more at this time as one of the roles of being an MPS ambassador at this conference was to be a roving reporter and write up their varying experiences for the MPS Magazine.

Whilst Faye, Jibreel, Lois, Megan, Joanne and Roshani write up their experiences for the next MPS magazine I would like to say a huge thank you to the Shauna Gosling Trust for funding our young MPS ambassadors to make this trip of a lifetime.I would also like to thank Genzyme, Shire, Biomarin and Healthcare at Home for supporting Linda, Jolanta, Chairman Barry Wilson, Trustee Sue Peach and I to participate in this Conference. **Christine Lavery**

MPS II CODEBREAKER'S MEETING
25 - 27 May 2010

This meeting held in Vienna, Austria, hosted by Shire HGT and chaired by Dr Jo Muenzer, Consultant Paediatrician at Chapel Hill, North Carolina in the USA offered a unique opportunity for clinicians from Eastern Europe and the Baltic States to learn in detail the biology, clinical manifestations and treatment for MPS II, Hunter disease. I was invited to speak on the role of the Patient Organisation. In the three workshops there was an opportunity to discuss in more detail the emerging MPS Society's for the Russian Federation and Slovakia and acknowledge in particular the excellent work of the Polish MPS Society. **Christine Lavery**



ALLIANCE OF BRAZILIAN MPS SOCIETIES (Alianca Brasil De Mucopolissacaridose)

The 'Alianca Brasil de MPS' (ABRMPS) has been created by the coming together of most of the local or state MPS Societies that exist in Brazil and comprises of all the national territory. It currently represents over 90% of the MPS populastion in Brazil. The Alliance was created in response to the pressing need to e mpower state MPS Societies and in response to an absence of a strong and legitimate Brazillian association to represent the MPS patients and their families.

It is very important to point out that the association called Sociedade Brasileira MPS (Brazilian MPS Society) that has participated in International MPS Network Meetings is not the legitimate representative of Brazilian patients, their families or their interests. Despite holding the name 'Sociedade Brasileira MPS' this Society is not constituted by the local / state associations and the state MPS associations have never been able to see the legal terms of 'Sociedade Brasileira MPS' constitution. Sociedade Brasileira MPS promotes itself as a 'virtual society' despite the website not having been in existence for over a year. Despite these negative facts Sociedade Brasileira MPS was also invited to be part of the Brazilian MPS Alliance as the Alliance's goal is to demographically unite all state MPS Societies.

The Alliance of Brazilian MPS Societies intends to work seriously, honestly, transparently and uncompromisingly to uphold the quality of life of all those affected by MPS. Our board of directors is nationally well known, not only within the MPS community but also amongst Ministers of the Supreme Court, Public Ministry, Ministry of Health, Deputies and the Senate. All the physicians involved in caring for MPS patients in Brazil and the other Latin American Associations are also aware and trust our work on behalf of the MPS patients and their families.

We hope to be accepted as the legitimate representative of Brazil in the International MPS Network at the meeting in Adelaide, Australia.

Regina Prospero

Alianca Brasil de MPS Representative

Expert Meeting on Sanfilippo Disease, MPS III

27 - 28 August 2010
Northampton Hilton

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

Archie's great grandmother helps the MPS Society gain £100,000

It was late Summer 2003, the Rudham family, parents Jake and Geraldine, their children, Emily (born 1998) and Archie (born 2000), were living with us whilst they renovated their house. Geraldine came into the garden with an anxious look on her face after visiting the doctor with Archie. We were worried about Archie, after Emily, then 5 years old, realised that he couldn't stretch his fingers properly. Archie had been referred to a specialist paediatric doctor by his GP. Whatever was wrong was uncommon, even rare, since the local doctor had never seen a case of this condition.

Searching the internet gave Jake the information he needed after reading the letters 'MPS' in the doctor's notes. Archie's illness was indeed rare, and at that time, treatment had not been fully tested, so we were shocked, upset and desperately in need of reassurance and hope. Not long after this diagnosis, the Rudhams moved into their own home. During the following weeks and months, they were reassured to find that an organisation existed to help anyone who needed information about MPS illnesses. They found out more about Archie's illness with the help of the MPS Society. This organisation proved to be a continual source of support and guidance especially in the early years of Archie's diagnosis of MPS II, Hunter's.

We told all relatives and friends, repeatedly explaining the nature of the illness since no-one had heard of it. But how were we to tell great-grandma, who was so fond of her grandchildren and now her two great-grandchildren? We decided to break the news gradually. Living 35 miles away, she didn't see Archie very often, and we first told her about Archie's inability to open his fingers. She accepted his boisterous behaviour and made allowances for his condition, without fully realising the implications of the illness.

Archie Rudham was born in 2000 and was diagnosed in 2004 with MPS II Hunter's, an illness passed down the female line via the x- chromosome. Tests on the family showed that Archie's great grandmother, Kathleen Bennett, carried Archie's condition or was herself a carrier from earlier generations. Kathleen knew that Archie was seriously ill but never knew that the condition had passed through her. When Kathleen died in 2009 a few weeks short of her 97th birthday following a short illness, she left a considerable estate which was to be passed on to her daughters and grandchildren.

Can you help us?
The MPS Society is looking at a new initiative to fund research into issues related to MPS and related diseases. We would very much like to see whether there is any possibility of working mutually with the Brains Brewery. If anybody has any connections to the Brewery we would be very pleased to hear from you. **mps@mpssociety.co.uk**

Kathleen (pictured above with Archie) and her husband, Ernest, who died in 2005, lived rather modestly but were successful in creating a useful estate, the bulk of which passed to Kathleen on Ernest's death.

After Kathleen died and the estate was valued, the family decided that it would be right and that Kathleen would have wished it, to make a donation to the MPS Society. This was particularly efficient from a tax point of view using a Deed of Variation agreed by the beneficiaries affected by the donation.

A Deed of Variation was drawn up and signed by the relevant members of the family, some of whom were foregoing their rights to part of their inheritance and this was approved by the Inland Revenue. This is a particularly efficient operation from the tax point of view because the amount of the inheritance foregone by the benefactors of the Will is increased by two thirds when the tax saving is added to the donation. By this means a total donation of £100,000 including the saved tax was given to the MPS Society on 19 January 2010 from the Estate of Kathleen Bennett. The amount is free from any encumbrance and the Society can use it for whatever purpose it chooses.

Anyone who feels that a family estate can make an efficient donation can obtain further information and advice from the MPS Society, any Probate Service or from a solicitor or accountant. This can be done at any time between the person passing away and the application for Probate. Anyone writing a Will can choose what they want to happen to their Estate. Making a bequest to the MPS Society can be particularly efficient. **Marian Henshall**

If you would be interested in leaving a Gift in Your Will, please contact the MPS Society for a fact sheet on this subject. **fundraising@mpssociety.co.uk**

Leave a Gift in your Will and support the MPS Society in your lifetime and beyond

wear your jeans to work

help children like Tom

Get everyone at work to wear their jeans for genes on Friday 1st October. You'll raise money for equipment, vital support and research for children with genetic disorders across the UK.

We're changing the world for children with genetic disorders. All you have to do is wear your jeans.

JEANS FOR GENES™

Sign-up for your FREE fundraising pack today at:

www.jeansforgenesday.com

or call us on 0800 980 4800