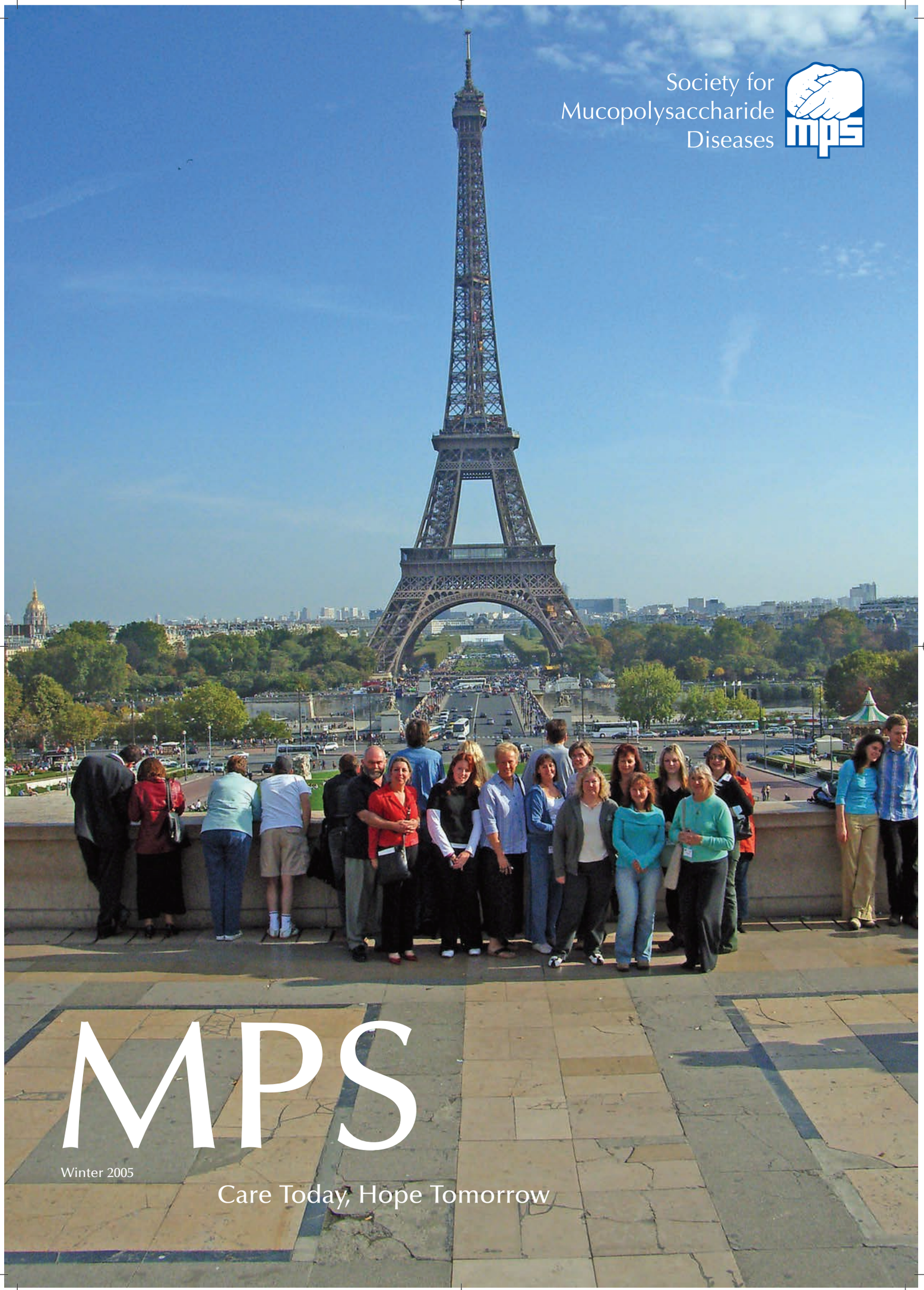

Society for Mucopolysaccharide Diseases

MPS

Winter 2005

Care Today, Hope Tomorrow

Mucopolysaccharide and Related Diseases are individually rare; cumulatively affecting 1:25,000 live births. One baby born every eight days will be 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.

What is the Society for Mucopolysaccharide Diseases?

The Society for Mucopolysaccharide Diseases (the MPS Society) is a voluntary support group, founded in 1982, which represents from throughout the UK over 1200 children and adults suffering from MPS and Related Diseases, their families, carers and professionals. It is a registered charity entirely supported by voluntary donations and fundraising and is managed by the members themselves.

What are the aims of the MPS Society?

To act as a support network for those affected by MPS and Related Diseases

To bring about more public awareness of MPS and Related Diseases

To promote and support research into MPS and Related Diseases

How does the Society achieve these aims?

Advocacy Support
Provides help to individuals and families with disability benefits, housing and home adaptations, special educational needs, respite care, specialist equipment and palliative care plans

Telephone Helpline
Includes out of hours listening service

MPS Befriending Network
Puts individuals suffering from MPS and their families in touch with each other

Support to Individuals with MPS
Empowers individuals to gain independent living skills, healthcare support, further education, mobility and accessing their local community

Regional Clinics, Information Days & Conferences
Facilitates eleven regional MPS clinics throughout the UK and information days and conferences in Scotland and Northern Ireland

National & International Conferences
Holds annual conferences and offers individuals and families the opportunity to learn from professionals and each other

Sibling Workshops
Organises specialist activities for siblings who live with or have lived with a brother or sister suffering from an MPS or Related Disease

Information Resources
Publishes specialist disease booklets and other resources

Quarterly Magazine
Imparts information on disease management, research and members' news

Bereavement Support
Supports individual families bereaved through MPS and the opportunity to plant a tree in the Childhood Wood

Research & Treatment
Funds research that may lead to therapy and treatment for MPS and Related Diseases as well as furthering clinical management for affected children and adults

Cover photograph:
Sightseeing during the Paris Fabry Conference

CARE TODAY HOPE, TOMORROW

MPS Magazine

MPS Society

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Newsletter Deadlines

Spring	1 Mar 2006
Summer	1 Jun 2006
Autumn	1 Sep 2006
Winter	1 Dec 2006

Become a Friend of MPS

Subscriptions may be taken out from the UK or overseas by contacting the MPS Society's Office. The articles in this magazine do not necessarily reflect the opinions of the MPS Society or its Management Committee. The MPS Society reserves the right to edit content as necessary. Products advertised in this magazine are not necessarily endorsed by the Society.

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CHIEF EXECUTIVE'S REPORT

Winter 2005

For so many in the MPS Society, the Christmas season will have been a rollercoaster of feelings. Indeed the last twelve months have been like that. The year started in the wake of the devastation of the Tsunami when we waited anxiously to learn the fate of the families of three Morquio children who live on the beach in Chennai, India. We gave thanks that, although they had lost everything, they were alive and some MPS members have helped financially towards the rebuilding of their homes.

We are grateful that MPS staff were safely in the office on 7 July, the day of the London bombings and that those we work with at Great Ormond Street Hospital and on the tube at the time were saved.

Further afield, there was the enormous earthquake in the Kashmir region of Pakistan and India. The suffering of so many goes on as the winter snows settle in. Our thoughts at this time are with members of the Society who have lost relatives in this tragedy.

On 28 November the retrial of Andrew Wragg for the murder of his son, Jacob, who suffered from Hunter disease started at Lewes Crown Court. Over two weeks, Sophie and I sat and heard in detail the evidence of this case. It took the jury of eight women and four men five hours to deliver a verdict of not guilty to murder and guilty of manslaughter on grounds of diminished responsibility. It would be an understatement not to say this was a unique, complex and tragic case that only those who sat through the two trials can truly appreciate.

So much has been written and said by the press that to be fair to all our members we have published in this magazine the full verbatim statement given by Mrs Justice Ann Rafferty when passing sentence on Andrew Wragg. The Society's press statement is also published. You will also see the Society does not condone the actions of Andrew Wragg but fully respects the unanimous verdict of the jury and the judgement made by Ann Rafferty. We acknowledge that our members will have diverse views on the outcome of this case. If you are affected in any way or feel that you want to talk, please do contact us.

Finally on behalf of the MPS staff team and Trustees, we wish you all a peaceful New Year.

Christine Lavery, Chief Executive

On a positive note for the New Year, after many months of running backwards and forwards to oversee builders, surveyors, solicitors, telephone engineers, sign writers and an endless string of telephone calls, emails and letters, the Society has moved! Our new contact details are as follows:

MPS House, Repton Place, White Lion Road, Amersham, Buckinghamshire, HP7 9LP

Tel: 0845 389 9901, Fax: 0845 389 9902

Email: mps@mpssociety.co.uk, www.mpssociety.co.uk

News from the MANAGEMENT COMMITTEE

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Management Committee Meeting held 26 November 2005.

Personnel

The Trustees were pleased to be advised that the staff structure agreed some months ago is now in place and all posts filled. The Chief Executive (CE) reminded trustees that Clare Cogan would be starting her maternity leave in December and that Sophie Denham will be Acting Senior Advocacy Officer.

Fundraising

The Finance Officer (FO) informed trustees that fundraising receipts from members for the past quarter achieved budget predictions. In some ways this was attributed to the ever-expanding MPS Magazine and Fundraising newsletter. The FO also reported ideas for the development of the website and Society's information resources.

Alton Towers

Applications for Alton Towers have been coming in thick and fast. In a separate mailing the Society informed members that they could reserve a room before Christmas for a deposit of £50 and make final payment in the New Year.

Childhood Wood

Bob Devine, Barry Wilson and Wilma Robins have visited the Childhood Wood (CHW) recently and reported that the new path fits neatly into the landscape. The CE advised Trustees that the plaques in respect of all trees planted prior to the year 2001 have been removed. Members are able to receive their plaque (if it was removed from the CHW) upon request. The 'memory board' will be in place in the Spring of 2006. It was agreed that Wilma Robins will proofread all Welsh text in respect of the board.

International Symposium

The CE and Sophie Denham have met with the organisers of the International Symposium in Venice and will return to carry out a risk assessment in respect of the venue and our members' safety in the New Year. Trustees considered how to support members wishing to go to the International Symposium and agreed that the Society will meet the costs of the registration of the conference. This will allow families to arrange their own travel and accommodation and, if they wish to do so, extend this into their own holiday.

Governance

The CE spoke of the papers tabled and the new guidelines for this year's end accounts. The Trustees were asked to complete a skills audit at the meeting or send it back before Christmas. It was also agreed that the skills audit form should be redesigned to allow Trustees to qualify their skills.

Strategic Plan

All Trustees were in receipt of a draft 5 year strategic plan and full discussion took place particularly around the Society's mission statement. It was agreed that the Trustees would consider this plan and report back by 10 January 2006. At the same time the first year objectives were agreed with the suggestion that a public relations section should be added.

Jeans for Genes

Trustees and staff gave thanks to Chris Holroyd in his absence for all his assistance with Jeans for Genes over the last few months. The Trustees agreed the Articles of Association and Memorandum at the meeting and are considering the four other documents further. Wilma Robins gave an overview of the last Jeans for Genes carol service meeting attended by her and it was acknowledged that the MPS Society had sold a majority of the tickets.

Policies

The Trustees reviewed and agreed with amendments the Maternity Policy and Reserves Policy. All other policies scheduled to be reviewed at that meeting were deferred until the New Year due to time constraints.

International Symposium on Mucopolysaccharide and Related Diseases
29 June - 2 July 2006, Venice Lido, Italy

In order to enable as many MPS families as possible to attend, the Society will fund the conference registration for up to 20 UK members of the MPS Society. The cost of travel and accommodation will be the responsibility of the individual families. Childcare will be available during the conference and those members registering through the MPS Society may request experienced English-speaking childcare volunteers.

Whilst the childcare programme is not yet finalised, it is expected that those not in the crèche will spend time on a private beach. The Society will be carrying out a full childcare risk assessment in the New Year and will report back to members who have booked.

For more information please visit **www.congress2006mps.it** or contact the MPS Society on 0845 389 9901.

Verbatim Sentencing Statement from Mrs Justice Ann Rafferty

I deal with you for the taking of your son Jacob's life at a moment when your responsibility for what you did was diminished as a consequence of an acute stress disorder and adjustment disorder.

You are no threat to the public. I treat you as of good character.

Killings range from the actions of the sadist to those of a man who as a mercy kills a loved one in response to a plea for release from a terminal illness. Although Jacob was unable to make such a plea to you I accept that your genuinely held belief was that what you did would bring to an early end a life afflicted and drawing inexorably to its close without intervention.

Jacob was vulnerable by virtue of his disability and age, and you held as to him a position of trust. Mitigating factors are your belief that what you did was an act of mercy, that you reacted to stress you found insupportable, and that you have from the moment you telephoned the police admitted what you did. I give you maximum and generous credit for that honesty as I remind myself of the evidence called by the Crown that Jacob's airways were so obstructed that without your admission a death certificate would have been signed.

Your then wife, late that night, removed your younger son from his bed and drove via a late opening shop to a layby where she stopped the car. Only after you had telephoned her with the news that Jacob was dead did she drive on to her mother's flat and without any prior arrangement leave George with her.

One would have to be quite remarkably naïve to accept that this dedicated and experienced mother behaved in that way solely so as to enjoy an evening of prolonged intimacy with you. I have no doubt she was complicit. Had I concluded otherwise I should have formed a harsher view of you. I accept that you would not have taken Jacob's life had you for a moment thought that she disagreed with what you were to do.

In my judgement what you have done and said since you killed Jacob imported to begin with a desire to shield and protect her, as did your instruction to her to quit the house before you arrived, so that as far as you could, you guarded against incriminating her. All who listened to the evidence must have wondered at the remorseless strain Mary bore lovingly and bravely during the ten years she dedicated to Jacob – as you concede you did not so consistently do. I shall sentence you loyal to what you have always said, that you did not do it for her or for yourself but for him.

You served your country in a successful army career, which you gave up out of love for your family. In my view it was when you quit that life, which demanded structure and which recognised valour, that your path to this court began. With hindsight you and Jacob and Mary and George would have been better served by the experienced support and discipline of your regiment had you stayed. You would almost certainly by now have been commissioned into a service career of distinction.

No matter your motive, the end of Jacob's life was not in your gift. I am well aware that no sentence the court passes can be measured against the loss of him. Nor should it be.

Mr Sayers QC told the jury that you took your son's life as he stood at the gates of his last dreadful journey. No words could better have caught the mixed tragedies with which I have to grapple.

For all the reasons I have set out your case seems to me exceptional. That being so, I consider that there is nothing to be gained by taking from you your liberty. Sending out as I sentence you the resounding message that this was not a mercy killing but a deed done by a man suffering from diminished responsibility, the sentence of the court is of two years imprisonment suspended for two years.

ANDREW WRAGG TRIAL

Press Statement
on the verdict of the Andrew Wragg trial on behalf of
The Society for Mucopolysaccharide Diseases

We have heard over the last two weeks many statements about Jacob and how Hunter Disease affected him. Hunter Disease is a complex condition that tragically results in affected children at the severe end of the spectrum losing all skills gained and dying in childhood.

There is no doubt that Jacob, like a majority of those suffering from MPS, required constant attention and care. Children like Jacob are doubly incontinent, have poor mobility, difficulties in communicating and considerable medical problems. MPS diseases are tragic and complex. Parents and carers face many issues that are emotionally, as well as physically challenging.

However, this does not mean that it was right for Jacob's life to be taken in this way. Jacob had his own unique quality of life that included going to school, enjoying outings with his family and seeing his friends at Naomi House Children's Hospice.

It was abundantly clear in the evidence given that the family felt that their local authority had failed to provide appropriate care and services to them, which ultimately added significantly to the stresses already inherent in caring for a child like Jacob. If there was to be one positive outcome from the tragedy of Jacob it would be that local authorities throughout the land urgently review their provision of services to families caring for children with degenerative, life-limiting conditions, ensuring that resources are appropriate, adequate and timely. There are over 500 MPS families like Jacob's in the UK, many not receiving an adequate level of resources to meet their needs.

12 December 2005

Thank you to all the families who have expressed their personal views.
We will endeavour to share these with you in the next Magazine.

Announcements

Introducing... Neisha Hall


Nikki McAuliffe

I'm the baby of the team – not in age to my dismay though! I joined the team as Advocacy Support Officer at the beginning of August 2005 and I must say I am enjoying every aspect of the job. It's a pleasure to talk to those families I have been fortunate to have contact with so far – if you're feeling left out though please don't hesitate to ring to say hello! My background has pretty much been in the care industry. Prior to starting at the MPS Office I worked in the office of a large domiciliary agency and before that I managed a large nursing agency. Both on occasions very much hands on.

My one great love in life is to travel and I have been fortunate to have holidayed in some beautiful countries, and one very special time for me was living with relatives and travelling throughout Australia – many years ago now I hasten to add!

I hope to speak to and meet many more families and if I can be of any help you'll usually catch me on the office number, although I do ask you to be gentle with me as I'm still learning!

Dr Ken Nischal, consultant opthalmologist at Great Ormond Street Children's Hospital, London who spoke at the 2005 MPS Society weekend conference in Northampton, has received an Honourable Mention for his clinical poster at the World Cornea Congress held in Washington, USA in March 2005. Mr Nischal was also invited to be Visiting Professor at the Kellogg Eye Institute, University of Michigan, USA at the same time.

Dear MPS Society

We have just returned from a month in Canada and we are delighted to tell you that our daughter Kate has a beautiful baby boy whom they have named Benjamin. Coincidentally he was born on Kate's birthday so we have reason for a double celebration!

Sue and Vic Lowry

Editor's Note: Sue and Vic's other daughter, Sarah, suffered from MPS VI.

I'm at High School now!

This is my first high school portrait. I started high school on 21 August 2005. I was very nervous as I knew that I would lose a lot of my good friends from primary school. However, I have managed to make new friends and I am still in contact with my best friend, David.

I have learning support and they help me get around school so I am not late for any of my classes. I can go to some of my classes myself now. I want to be like everyone else so I try to do things myself. I enjoy art and woodwork and I like my science teacher. I have to watch the Bunsen burners as I don't want to singe my eyebrows! I'm starting to venture up town for dinner too. There is a MacDonald's nearby too, which is great. I still need my taxis up and back in the morning and the evening as I couldn't walk that far as my feet would be sore.

I'm coping well on Fabrazyme and keeping very well. My nurse, Margaret Russell, is leaving to get a new job so I will miss her. I'm looking forward to someone new so good luck to Margaret.

Love **Colin Hickman**, aged 13, Fabry sufferer

New Members

Mrs Scott has recently been in contact with the Society. Caroline has a diagnosis of Fabry Disease. The family live in the South East.

Glenys Berry has recently been in contact with the Society. Glenys has a diagnosis of Fabry Disease and lives in East Yorkshire with her family.

Claire Harris has recently been in contact with the Society and has a diagnosis of Fabry Disease. Claire lives with her family in the South East.

Lisa Kirby has recently been in contact with the Society and has a diagnosis of Fabry Disease. Lisa lives with her family in the South East.

Edward and Lucy Brock have recently been in contact with the Society. Their daughter Hannah has a diagnosis of Maroteaux-Lamy Disease. Hannah is two years old and the family live in the North West.

Ms Fair has recently been in contact with the Society. Her son James has a diagnosis of Hunter Disease. James is 8 years old. The family live in the North East.

Mrs Senior has recently been in contact with the Society. Her son, Rajiv, has a diagnosis of Sanfilippo Disease. Rajiv is 7 years old. The family live in East Anglia.

Nicola Cooper has been in contact with the Society. Her daughter Hannah was diagnosed with Hurler Disease aged 18 months. In August she had a bone marrow transplant.

Deaths

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

Ben Wilton who suffered from Hunter Disease and who died on 29 July 2005 aged 10 years.

Wing Kim Yeung who suffered from Maroteaux Lamy Disease and who died on 31 August 2005 aged 25 years. For more information visit http://www.livejournal.com/users/wingsblog

Kerry Jones who suffered from Sanfilippo Disease and who died on 8 October 2005 aged 39 years.

Katie Smithers who suffered from Hurler Scheie Disease and who died on 17 October 2005 aged 21 years.

Megan Fasey who suffered from Hurler Disease and who died on 9 November 2005 aged 11 years.

Oliver Hall who suffered from Sanfilippo Disease and who passed away peacefully at home on 8 December 2005 aged 10 years. He will be sadly missed.

Syed Suqlain Abbas Shah who suffered from Morquio Disease and who died on 3 December 2005 aged 20 years.

Courtney Taylor who suffered from Hurler Disease and who died on 7 December 2005 aged 8 years.

The Society would like to apologise to Mr and Mrs Allen for an error in the Autumn 2005 MPS Magazine. Their son, Daniel, was 16 years old, not 15 years old, when he sadly passed away on 5 July 2005.

Congratulations to Claire Beirne on the recent birth of her twin boys, Matthew and Adam.

On behalf of all the staff at the MPS Society, we would just like to wish her all the best with her two new arrivals!

This is a picture of Ben Wilton. His parents hope that it can show other families how happy Ben was all the time, despite having Hunter Disease.

MEMBERS' NEWS

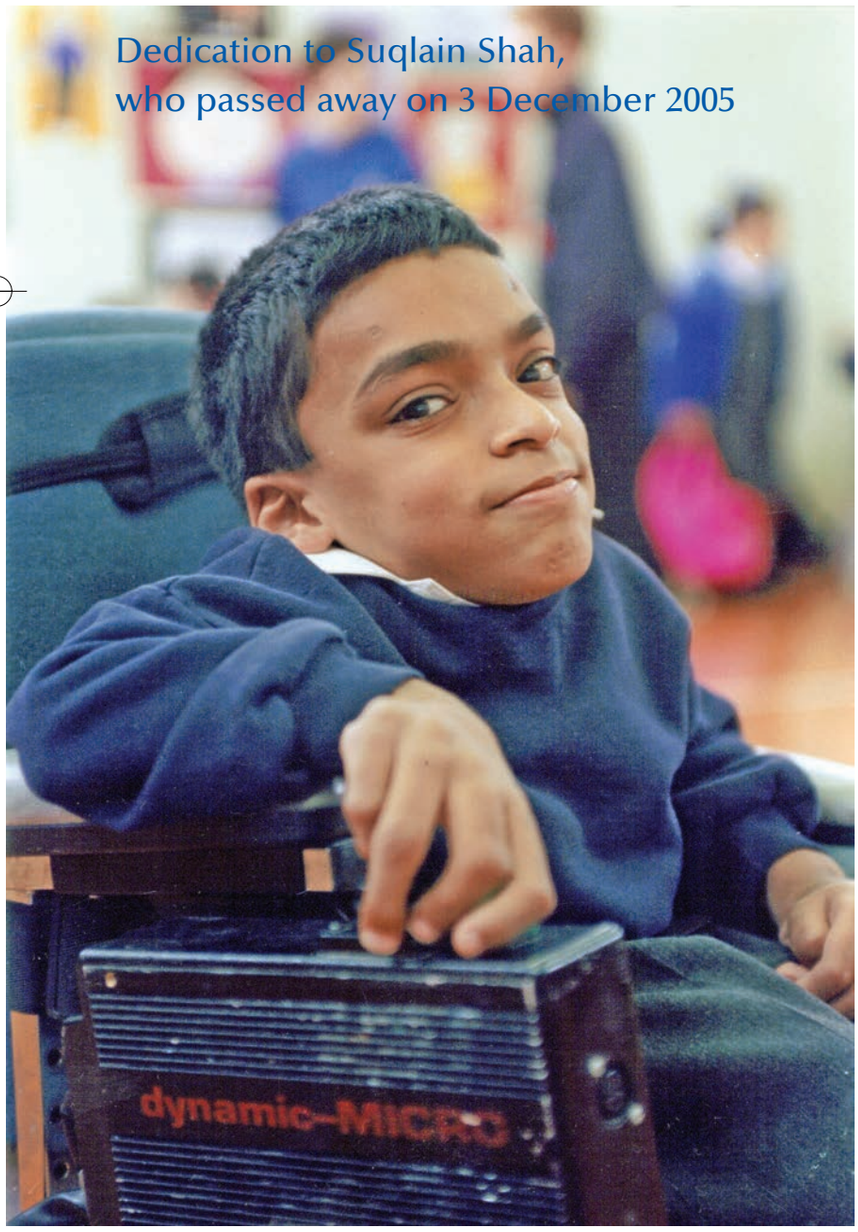
Dedication to Courtney Taylor, from her brother Billy

There are no words in which I could describe Courtney that would truly do her justice.

Anyone who knew her would know how she instantly won you over with her huge personality and insatiable lust for life. Courtney wanted to try everything once and loved every second while trying it. Despite however restricted she was she took life and got everything she could out of it.

It is very rare you come across anybody so charismatic and so full of light and happy and thoughts. To myself, my mother and Ian, who she loved so dearly, Courtney was our little silver lining through all the bad times. Now our silver lining is gone I don't know what we will do.

All I know is that Courtney was a tribute to herself. I've never been prouder of anyone and I will never stop loving my little sister. She touched all of our hearts and changed our lives. She will always be a part of us.



Dedication to Suqlain Shah, who passed away on 3 December 2005

"To Allah we belong, and to Him is our return."

The road, the journey hard,
but Suqlain you always were a star.
An inspiration to one and all.
He never complained not even once,
just carried on; he waged it all.

A lovely son, a treasure to have
an eternal brother, we'll never forget.
Not once a burden never at all.
An honour to have you Suqlain.

The loss too huge to fill.
The hurt too strong to forget.
The need in the family,
no one can ever fill.

You are at peace now my son.
My faith in this belief is strong.
You were an angel, a star sent down
to shine in this world, and in our lives.
Your light will be eternal
in our hearts and in our minds,
forever till we die.

Allah saw you getting tired,
when a cure was not to be.
So he wrapped His arms around you
And whispered, "come to me."
You didn't deserve what you went through,
So Allah gave you rest.

May Allah bestow all the blessings in heaven
you never had on earth,
run free, run strong;
play with the angels
and sleep with the stars.

Rest in peace
our Beloved Syed Suqlain Abbas Shah.
Dedicated by Auntie Shaista and Cousin Maria.

I wanted to take this opportunity to say a 'temporary goodbye'. As a few of you already know, I am due to have a baby in January 2006 unless he or she decides to make an early appearance and become our unique Christmas present! In my absence, Sophie Denham will be taking over my role as Senior Advocacy Officer and the rest of the advocacy team, Linda, Neisha and Nikki will be, as ever, on hand to provide any support or information which you may find helpful. I hope to be back in the middle of next year to combine my much cherished role with the responsibilities of parenthood!

Clare Cogan
Senior Advocacy Officer

The Society is recognised for its excellence in volunteer management and practice

On the 20 October 2005, Clare and Sophie attended an awards ceremony run by Voluntary Action Group, Chiltern and South Bucks. We had been nominated by Helen Patterson, one of our long-standing volunteers for the work and support we have done with our volunteers. There were a number of local charities and voluntary groups who had been nominated for this award and their work recognised. The Society came joint runner up and Clare represented the Society by collecting the award.

It was lovely to be a part of this and for the work of the Society to be recognised. Our thanks go to all our volunteers who ,without your continued support and dedication, we would not be able to provide the high level of service that we do to our members at our events. We would like to thank Helen Patterson in particular for all her hard work and for nominating us for this award. Thank you.

Caught in the Act!

The Society, along with many hundreds of other charities, entered a photographic exhibition called 'Caught in the Act' which showed various images of volunteering.These went on show in Amersham Market Hall. The aim was to capitalise on the huge interest generated by Year of the Volunteer 2005. Hopefully this will lead to volunteers in the Chiltern and South Bucks area to lend a hand in their communities. The exhibition showed that everyone can make a contribution to their community. Volunteers featured ranged from 14 to 92 years of age. It clearly showed the enjoyment and satisfaction that volunteers gain from working with others. Volunteer centres are located all over the country and help people who wish to volunteer to find the right opportunity to match their skills, location and time available. If you want to find out more look on the national volunteering website **www.do-it.org.uk** which is easy to use and lists local vacancies in your area.

Taieyyib's 18th Birthday celebration

On receiving the invite to Legoland Windsor; we thought it would be a good place to celebrate our sons Taieyyib's 18th birthday, as he was actually turning 18 on this day. Taieyyib has Sanfilippo disease and we knew he would just love going to Legoland. We went as a family and Taieyyib's sister Sarah came down from the North to help celebrate his special day. We had great fun at Legoland, Taieyyib especially liked the the boat ride around the park, which he went on twice with his sister Sarah.

Towards the end of the day we were asked to meet all together at a certain point. When we arrived the MPS Society had a birthday cake and drinks to celebrate Taieyyib's birthday. It was nice to share his special day with other families who attended the event.

After an exhausting day we returned home for more celebrations with family and friends who were unable to join us at Legoland.

Hannah (MPS III) and Great Gran Robinson

Four year old Hannah Shannon (MPS III-A) and her Great Gran Robinson don't get together as often as they'd like because of the Irish Sea between them. But here they are having fun when the family was in England in the summer for the MPS conference. Great Gran Robinson has been busy in the last couple of months selling MPS raffle tickets, and won't let anyone leave the house without buying one. She's single-handedly sold ten books!

Do you have any stories, photos or interesting news for the MPS Magazine? If so, email us at newsletter@mpssociety.co.uk

Rachael by Lauren Kermode (Rachael's sister)

A sister is a best friend
A sister is supposed to drive you round the bend
A sister is caring and loving
A sister is supposed to be fighting and shoving
My sister shares lots of smiles
Rachael's smiles shine for miles
Rachael may have Sanfilippo C
But that doesn't matter to me
She loves to laugh and giggle
At parties she loves to wiggle
My life is so great
Rachael's a great mate
We have a laugh
We love the bath
Rachael and me look like our dad
We have a song that says he is bad
Thank you Rachael for being my friend
You don't drive me round the bend

Date for your diaries!

Northern Ireland
Thursday 4 May 2006 (MPS Clinic)
Friday 5 May 2006 (MPS Conference)

Scotland
Friday 16 June 2006 (MPS Conference)

'Our Rachey' by John Kermode, Rachael's Dad

Born September 1983, Rachael Louise Kermode

Our first child was due at the end of October. It was a Sunday evening and we were watching TV when my wife Hilary said she was having contractions. I remember saying that the baby is probably just having a practice. But, just a few hours later in a maternity ward in Chester, baby Rachael was fighting for her life. The cord was around her neck and her breathing was getting worse. Staff decided that an emergency caesarean was the only option and 30 minutes later a large nurse was giving me a bear hug whilst walking me to the special care baby unit. There I saw Rachel. She was a tiny little girl with tubes and electrodes all over her body. She opened one eye and I knew then what she was thinking - 'I know I am a little early, and I am a little bit poorly at the moment, but I think you will understand.'

We had a beautiful blonde-haired, brown-eyed baby who was just short of 5lbs. Whoever said there are no dress rehearsals in life was telling the truth. Rachael was now showing signs that she was somewhat slow in her development and her behaviour was somewhat different, but to us she was perfect. Playschool were having problems and this left us feeling that everybody was against our little girl. Professionals at the time told us we had a very naughty daughter and Rachael was given her own child psychologist who taught us to take time out with Rachael when her behaviour was at it worst. In fact, this turned out to be a complete waste of time as Rachael was not having any of this.

Social workers finally realised that she may have special needs. They told us something that we already knew - that Rachael was special. Years spent in special needs schools seemed to help but it was when we met Dr Ed Wraith at Manchester Children's Hospital that our whole world changed. We were told that our daughter had Sanfilippo Disease Type C and that Rachael's life would be limited.

Whilst travelling home in the car both myself and my wife were crying. Things would never be the same again. Nine years on, we finally had a diagnosis and that Rachael had a terminal illness. Like so many families we too have many stories of mass destruction in our home. Rachael's energy did not waver during years of lack of sleep and her need for continuous support in everything she did.

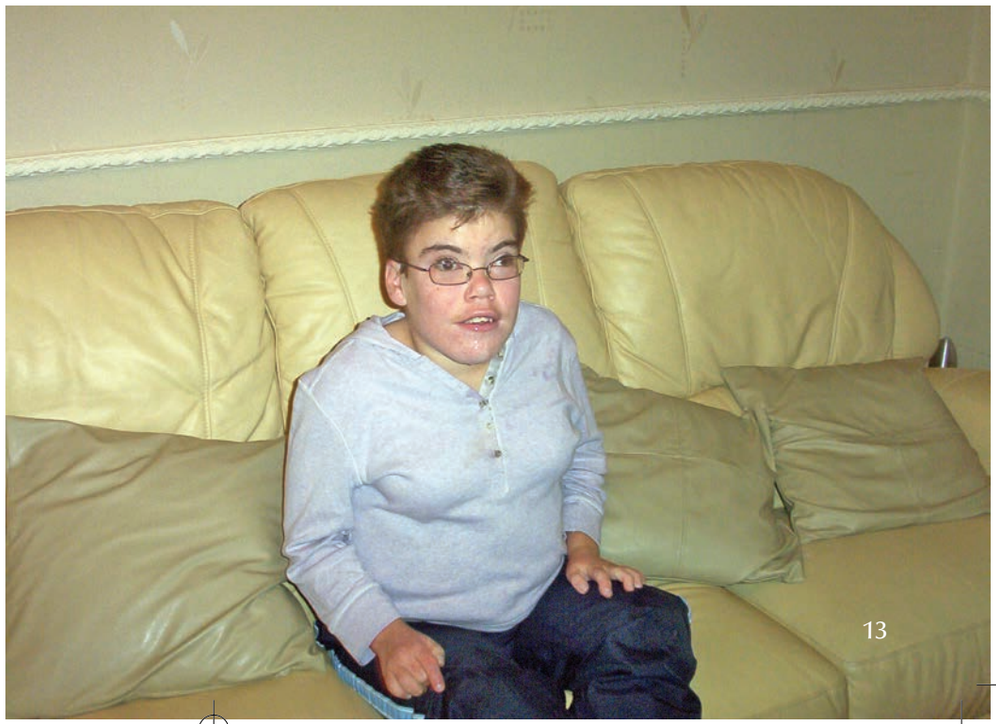
For a period of time Rachael lost use of her legs. She developed Perthes disease which meant her legs dislocated periodically and she would need to spend time in a wheelchair. We felt the longer she spent in the chair her condition would worsen. We all had different ideas about this and continued to walk her as much as we could. Given time this had a benefit and for some years Rachael no longer needed her chair.

Just as we thought things were going well Rachael developed an ulcer which burst whilst on holiday. We called the local doctor to be told she had a virus. We knew this was untrue so we rushed in the car to see Dr Wraith at Manchester. He told us that Rachael had internal bleeding, she was very ill and that it was touch and go for some time, but she pulled through.

Thinking that this was the end to this particular chapter Rachael decided it was time to start having convulsions just to keep us on our toes! This was a distressing time as for once we felt that we could not help her. However, as the months went by her fits did stop and she has not had one since.

There are so many stories I could tell you all about Rachael. As is often the case with Sanfilippo children, these stories are both amusing and at times upsetting, but Rachael's story continues. Celebrating her 18th birthday was in itself a land mark, but a year ago Rachael was 21 and this was a champagne party in itself.

As a family we continue to do as we have always done. Rachael's spirit and bravery continues to be our leader.



CHILDHOOD WOOD PLANTING

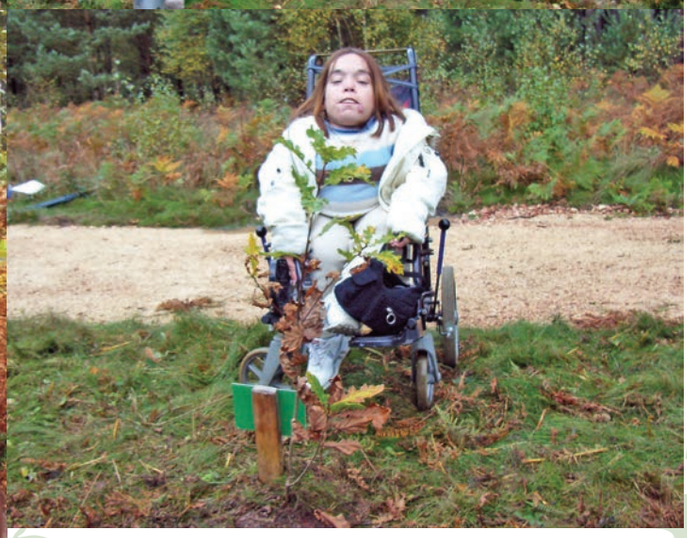
On Friday 21 October six families attended our annual tree planting at the Childhood Wood, Nottingham to plant trees in memory of those who had lost their lives to an MPS or related disease.

The day started off very wet and windy and we all hoped for a break in the clouds when it came to the tree planting. While we all gathered at the Clumber Park Hotel for the buffet lunch, the weather did just that and we had blue skies and sunshine. Barry Wilson, the Society's Chairman, gave a welcome speech to all the families who were participating in the day and invited them all to follow him in convoy to the wood.

We were met at the information board by Paddy Tipping MP, who welcomed everyone to the wood and shared with us all his reasons why the wood is so special to him. He then read out the names of the children and adults who were being remembered at the wood today. Wilma Robins, one of our trustees then read the poem 'Remember' before the families each collected their oak saplings and planted them together with their individual plaques.

Barry Wilson and Paddy Tipping MP planted the trees for those families who were unable to attend the wood and Byron Wibberley, Forest Ranger and his team were on hand to help with the planting if needed.

It was a lovely autumnal day and the wood looked beautiful with its new paths and growing trees, which really do look like they are reaching out and linking hands.



Photos clockwise from top right: The family of Christopher Rees, Larraine Mullen - sister of Samantha, the family of Evie Mason, the family of Ashleigh Tomes

Plaques have been retrieved from the Childhood Wood for dates up to 2001. Due to the growth of the trees, a few plaques could not be found. However, a large number were and these have been cleaned and the stakes removed. If you would like to have your plaque, please phone Maureen Cummins on 0845 389 9901. We will need you to send a self-addressed 20 x 27cm (8" x 11" approx) 'Jiffy' bag with 50p in stamps on it. We will have another look for any remaining plaques in January when the foliage has died back.

Bone Marrow Clinic by Cheryl Pitt

On 7 December the bone marrow transplant clinic for the under-5s was held at the Willink Unit RMCH, and on 9 December the clinic for the over 5's was held. As usual it was a delight to see the little ones thriving after their transplants and to see the slightly older children progressing so well. It was also a pleasure to meet some new faces, who hopefully will become members of the Society and will continue to develop relationships with other families and with the Society. It was also a good opportunity for Neisha to meet families for the first time. During both the mornings the waiting room at the Willink could have been mistaken for a kindergarten, as the younger children happily played with one other and with the toys. There also seemed to be a competition going on, to see who could eat the most crisps and biscuits at the same time! The clinics themselves ran like clockwork, and the idea to split the clinic over two days appears to be working well. So it just remains for Neisha and I to say a big thank you to Jean Mercer and Dot for organising the clinics and arranging refreshments. Thanks also to Gill Moss and the Consultants: Dr Ed Wraith, Mr Tim Meadows, Mr Brad Williamson, and Dr Rob Wynn for their excellent work and a successful clinic.

Photos clockwise from top right: Rachel Rothwell, Steven O'Reilly, Oliver Gosling, Leighton Barker, Charlie Escalonilla, Melissa McKie (all MPS I)

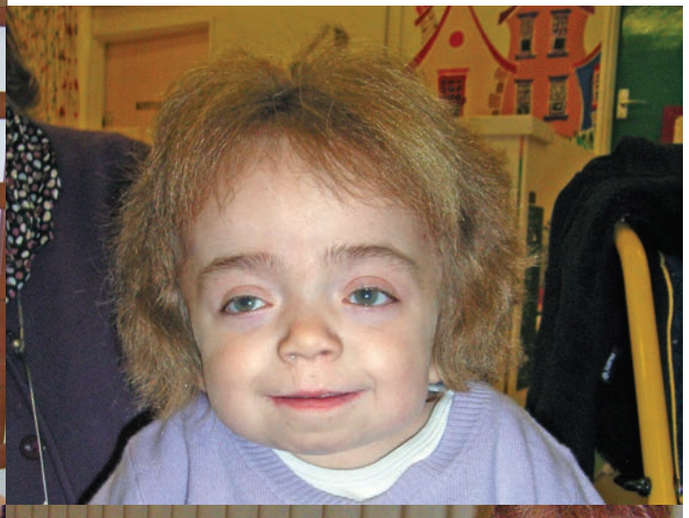
BRISTOL MPS CLINIC

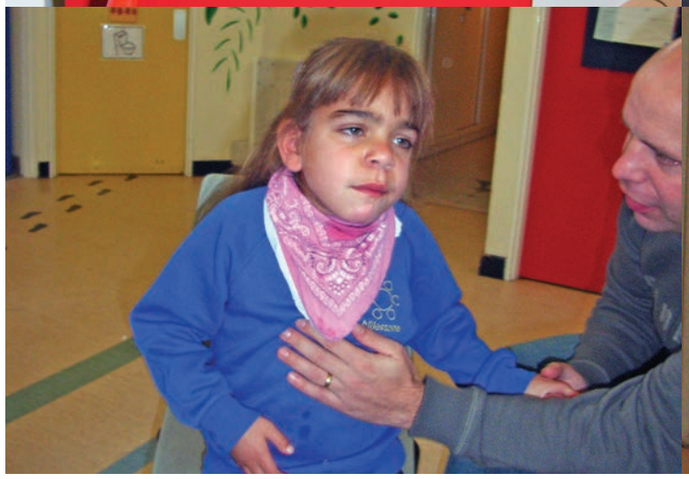
by Nikki McAuliffe

Firstly, I'd like to start off by saying a big thank you to Dr Wraith, his secretary Christine Caveny, Dr Jardine and his marvellous secretary Deirdre Freke for yet another well co-ordinated clinic.

Sophie and I arrived at the Frenchay Hospital shortly before the first patient was due to arrive at 9am. Unfortunately, the local traffic situation and an absconded cow (!) managed to put all the appointments back by half an hour until lunchtime when we finally managed to re-assemble some kind of order!

All in all, this was another successful clinic and I was honoured to have met such lovely people. Hopefully I shall get to meet some of you again soon.





Photos clockwise from top right: Lauren Graver (MPS I), Lewis Quant (MPS III), Jasmine Heap (MPS III), Archie Eaton (MPS IV), Georgia Lewis (MPS III)

BIRMINGHAM & NORTHERN IRELAND MPS CLINICS

by Neisha Hall

I have been asked to write an article on the clinics I was fortunate to attend with Sophie. My first experience was going to be the Birmingham Clinic on 26 October and after an early morning start, we arrived at the Victoria School, Northfield, South Birmingham and Dr Chris Hendriksz, Dr Anupam Chakrapani, Joy Hardy and Louise Simmons made us very welcome. It was lovely to meet the families who attended, and the clinic ran to time. I hope all who did attend benefited from the clinic, and a special word of thanks to Dr Chakrapani and Dr Chris Hendriksz for their ongoing support, and for Joy who went to get our lunch - funded by Dr Chakrapani, and for Louise who kept us all entertained throughout the day.

My second experience was an even earlier start, but this time involved a flight to Northern Ireland on 17 November. Again Sophie had to put up with me and after a short flight we touched down to a rather chilly Belfast. The clinic ran very smoothly, and I would like to say a special thanks to Dr Ed Wraith and Dr Fiona Stewart for their ongoing support and time, without which we would be lost, although Dr Wraith did have a rather long journey home as his flight got diverted and he then had to endure a long coach ride to Manchester - I do hope this hasn't put you off future Northern Ireland clinics! It was a pleasure to meet the families and I hope you all benefited from your appointments.

Photos clockwise from top right: Kyle Shields (MPS III), Luke Morrison (ML II), Hannah Shannon (MPS III), Dean and Aaron Doherty (MPS III)

Legoland – by Tim Summerton

Our Sunday started early as usual but with the added excitement, for William at least, of a trip to Legoland!

Before I begin on the day's events, let me first introduce myself. I am Tim, married to Sally and we have William age 9 and Sophie age 8 who has Sanfilippo type A and for the day's event was in her wheelchair.

The weather was a bit grey as we left home but it improved as we drove towards Windsor and by the time we got there, the sun was out but it was chilly. It was going to be a lovely day.

We met up with the other families and Sophie and Neisha at the entrance where we had an opportunity to talk, collect our tickets and the invaluable exit pass which is not what it sounds like! We took the hill train down to Mini Land but before we got there Will dragged us away to the more exciting Knights Kingdom and the Dragon roller coaster. I lost the argument (I don't think it was ever mine to win!) and took Will on the roller coaster while Sally stayed with Sophie and chatted to another family. The ride was great but I can say with certainty that Sophie would not have enjoyed it.

Next, it was off to the Maze and a race between Sophie (with me pushing) and William to reach the centre first. William won, but I blame the steps at the end!

It was then time for Sophie to enjoy the rides. We all had a gentle sail around Fairy Tale Land which Sophie thoroughly enjoyed. To keep up the nautical theme, we went to the boating school where we had great fun especially William driving Sally at speed! For both of these activities we were able to use the exit pass which gave us a fast track to the front of the queue.

We had lunch watching the log flume and then William dragged me on it, we both got rather wet! We spent the afternoon trying a further six rides and still no rain. We met up with the other families for a birthday celebration of cake and orange juice and it began to rain!

We took that as our cue to leave having had a wonderful day out. We were only sorry we didn't have time to visit all the lego models.

Thank you to all involved, especially Sophie and Neisha.

FAMILY DAY TRIP TO LEGOLAND WINDSOR

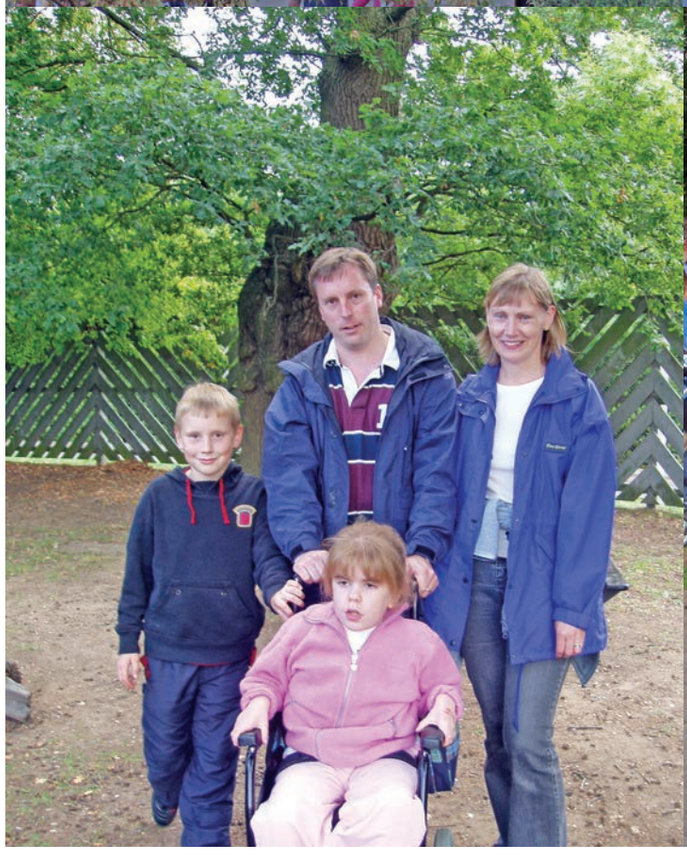
by Neisha Hall

Sophie and myself would like to thank all the families who attended the family day trip to Legoland on 25 September. We do hope the early Sunday morning start was well worth it!

Everybody seemed to enjoy themselves, the weather was especially kind to us – only one light shower of rain in the afternoon. It was a special day for one young man, Taieyyib celebrated his 18th Birthday, and it was lovely to be able to wish him a very Happy Birthday as we all met at 3pm for Birthday Cake and copious quantities of orange juice.

Although Sophie and myself weren't brave enough to go on any of the rides, feedback from families was positive and the Exit Passes issued were a real bonus as this enabled families to go on the rides without queuing.

We do hope everyone enjoyed the day and would like to thank everyone who attended for making it such a special day. Here are a selection of photos from the day.



Leeds Family Day

Aamina Hanif (aged 9) writes about the party

On 27 November 2005 there was an MPS Party at the Craiglands Hotel in Leeds. Me, my sister, my Mum and my Uncle went. On the way there my sister Aisha (who suffers from Sanfilippo Disease) fell asleep but when we arrived there she was wide awake.

When we went in there were a few families already there. We sat down to have our lunch. Neisha from the MPS Society joined us at our table. I had chips and beans to eat that were delicious, then I had ice cream. Neisha was very nice. As I was having my lunch I saw Truffles the Clown. I went up to him to introduce myself.

Truffles started his show and it was BRILLIANT! My sister doesn't laugh very often but the funny clown made her laugh so much, she was jumping up and down in her wheelchair. Truffles was that funny. I was laughing too. I was chosen to be the clown's assistant. He asked me to pick a counter out of a plastic bag. I picked out a blue counter so I had a blue cracker and this is what happened. He asked me an easy question but he did say it was hard. "What are Santa's helpers called?" I answered "Elves). I pulled the blue cracker and I won a watch (a toy one of course). There were another two girls there that got watches as well because they answered their questions right. After that Truffles carried on with his show.

Near the end of the show Truffles started to blow up some balloons and started to shape them into animals. We had to guess what animals he was making then he gave us the balloon to keep. My sister got a reindeer balloon with a shiny red nose on it.

After about an hour Santa Claus came. Everyone got a present, except for the adults. Santa pulled out a present from his sack that had a name on it. A few children did not come so somebody in their family got their present from Santa to give to them. I was very excited to see what Santa had given me but I waited until I got home. I really enjoyed everything about the party. It is the only party that has had the funniest clown. I really enjoyed myself.

Thank you to Neisha and everyone else for making it such a wonderful day. And special thanks to my sister for behaving so well!

On the 27 November 2005, we held the Leeds family day Christmas Party at the Craiglands Hotel, Leeds. Neisha and I travelled up the night before and braced ourselves for snow. Fortunately for us and everyone else we only got wind and rain.

The staff at the Craiglands were superb and had decorated the room magnificently with a tree, crackers and some noisy blowers to which Ben proceeded to entertain us with. After a feast from the carvery and some extremely sinful desserts, Truffles the clown was ready to take the floor with magic and fun for all. We even had a visit from fairy bear who sang us her special song. To calm everyone down Truffles the clown finished off with some balloon modelling while we waited for our surprise guest to arrive (I have to say at this point I was beginning to get a little nervous as a few children were ill and needed to get home and there was not a clattering of hooves to be heard!). However, there was no need to panic as Father Christmas arrived on cue, bearing gifts for all the good boys and girls, even those who thought they were too old to receive a gift from Santa.

After all the gifts had been given out and Santa's sack was empty, he wished everyone a very Happy Christmas and returned back to the North Pole to prepare for Christmas Eve and the families returned home to await his arrival.

By Sophie Denham

Scottish Christmas Party

By Nikki McAuliffe and Neisha Hall

Saturday 10th December saw myself and Neisha up in Scotland attending the MPS Scottish Christmas party at the Almond Valley Heritage Centre.

It was a good turnout and fortunately the weather wasn't too bad when we arrived. The day started in the soft play area where the children seemed to be enjoying numerous rounds of ball-throwing at one another, which gave the parents a chance to mingle and chat amongst themselves.

At lunchtime, everyone gathered in the party room where a party food style buffet had been laid out with balloons, hats and blowers for the children and a tray of sandwiches with much needed tea and coffee for the adults!

Once everybody had eaten, the children had a special visitor in the form of a gift-bearing Father Christmas. Each of the children received a gift and had a chat and their picture taken with him before heading off outside for a trailer ride around the park.

The day ended in the adventure play area giving the children (and some of the adults!) a chance to burn off some steam before the journey home.

A great day was had by all, including Neisha and I and I would just like to end this article by saying how lovely it was to meet the children and their parents again, and to thank the staff at Almond Valley for being so accommodating once again.

Here are a selection of photos from the day.

Starlight Wishes deminstrate 10th birthday!

To highlight Jeans for Genes' 10th Birthday, Starlight Children's Foundation was delighted to grant the wishes of ten very special children suffering from genetic disorders.

LIFE IN THE FAST LANE

Oliver Moody, who was referred to Starlight by MPS, was the first child to have his wish granted. Oliver is ten years old and he suffers from MPS VI, Maroteaux-Lamy Disease. He is car mad and he got a taste of life in the fast lane when Starlight granted his wish to visit the Ferrari factory in Maranello, Italy.

Oliver and his family thoroughly enjoyed their guided factory tour. They were fascinated by the robot-free production line and learnt how the road cars are put together by hand. They watched people stitching the interiors and saw how just one man puts the whole engine together. The factory staff are not used to seeing children in the factory so Oliver, his brother and sister got lots of attention! At the end of their tour they were presented with a 'factory passport' so they are welcome to visit again in the future.

Oliver and his family spent the rest of their time in Italy visiting the Ferrari museum, where they had the opportunity to get up close to the Formula One cars, and watching the high-performance cars being put through their paces on the Ferrari test track. Oliver said, "It was brilliant. It was like being in a fantasy. It was like a dream. We got to see one of the newest Ferraris, the F430, and I got to see the Enzo. It is my favourite Ferrari. It was the best day of my life."

Oliver's Mum added, "He loves cars and knows everything about them. He was thrilled when he was told he would be going to the factory. They made a real fuss of him and he had a huge grin on his face all day. It was lovely to see him so happy."

I am writing to say how much I enjoyed my trip to the Ferrari factory in Maranello, Italy.

We left home on Monday evening to drive down to Stanstead in London. It was 9pm when we arrived at the Radisson Hotel. Our room was a special family room with lots of gadgets! In the morning we had a lovely leisurely breakfast before strolling down to the airport.

We flew to Bologna them made our way to Marinello. We stayed at the Planet Hotel (right across the road from the Ferrari factory). Our room was right at the top and even had an upper level! We could see right over the whole factory. On Wednesday morning we walked over to meet Danielle in the factory reception and she arranged a guide to take us around for the morning to see how these amazing cars are made.

My favourite part of the visit was seeing the production line, with no robots and everything built by hand. It was fantastic because we turned a corner to see two lines of Ferraris facing towards us. Later on my family and I were all given a Factory passport which allows us to visit again at any time. After our tour around the factory we went to look around the Ferrari museum, there we saw how much the cars have changed throughout the years. And on Thursday we returned home and came back down to earth with a ride in Mum's Fiat Panda!

Finally, thank you to everyone involved in making my wish come true!

Oliver Moody

Photo of Oliver courtesy of Starlight

McFly me to the moon!

Roma, who is twelve years old, also had her wish granted in honour of Jeans for Genes' 10th Birthday. Roma suffers from MPS IV, and as a massive fan of McFly, she wished to meet the band when they were in Northern Ireland. Starlight worked its magic to make the night very special.

Roma's mum said, "We had a really super night. We got to meet all the boys in person; they shook our hands and spoke to Roma personally. We got our Starlight t-shirt autographed and Roma's CDs as well. Even though we had questions prepared, we felt a bit overwhelmed by everything and couldn't think of what to say! I must say that Roma really enjoyed the night - we all did - and Roma smiled the whole way through the evening. So a big thank you for this, it was a night to remember."

Roma tells us about her wish come true...

McFly are one of my most favourite bands and that's saying something as I listen to a lot of music! I really like their new album. When I was told about going to meet McFly I was very nervous, what if my family didn't know the songs well enough? What if Mum said she had all their albums (after all, they have only had two albums so far)! This was all very possible, so I took them on a crash-course into McFly teaching them their songs, their names, everything!

When we got down to the Odyssey Arena in Belfast we had dinner in this lovely restaurant that sold Spanish food and then on to the box office to find out where to meet McFly. I was very, very excited. I mean it's not every day you meet your favourite pop stars.

When we got to actually meeting them I was excited and nervous so I yelled at my Mum to get out my CD to get it signed. It was really funny because the girls in front could not keep their hands off McFly so security had to be called in to get them.

When I got my turn to meet McFly I was very nervous about it. They had scored massive number one hits and released the single for Comic Relief in 2005, but they were very down-to-earth people. Just like the kind of guys you would bump into down at the local supermarket. They are very polite and know their manners!

They were a bit mad, very excited about their concert. They introduced themselves and shook hands with us all. They had no make up on and were quite ordinary. They asked me if my little brothers were annoying but I said they were not. Otherwise, the boys would have nudged me from behind very sharply!

The concert was absolutely fantastic, with many of their old songs that they played. Unfortunately, the concert ended too soon. The stage set was brilliant and the lights were superb.

I just want to say thanks to everyone for making it all possible. It was a night we will always remember and it was truly fantastic!

STARLIGHT
Brightening the lives of seriously and terminally ill children

Starlight Children's Foundation brightens the lives of seriously and terminally ill children by granting their wishes and providing entertainment in hospitals throughout the United Kingdom. All Starlight's activities are aimed at distracting children from the pain, fear and isolation they can often feel as a result of their illness and at strengthening family bonds during difficult times. Last year alone Starlight helped over 400,000 children.

If you know a child who you think would benefit from having their wish granted, please visit **www.starlight.org.uk** or ring Starlight on **020 7262 2881** for more information.

Daniel Cooke

Hello everybody. My name is Daniel Cooke and I am 10 years old. I have Hurler Disease. I'm doing very well. If I need to know anything about my disease, my Mum, Sally, tells me. I don't always understand what she is talking about but we just get on with it. I have a younger brother Ben who is 7 years old and a sister called Kelly who is 14 years. I mustn't forget my dad Karl but I won't tell you his age!

I have always wanted to be in the MPS Magazine but have never had a reason before. So, now I have. I was a page boy at my Mum and Dad's wedding blessing, with my brother Ben on Saturday 24 September 2005.

It was a very long day and leading up to it I was very worried that I wouldn't be able to wear a suit like Ben and my Dad. My Mum always has trouble finding me clothes which fit properly because I have a large tummy and am a little overweight. I can never wear a pair of jeans as I need a large waist, then the legs are too long as I'm only short. It's not fair.

So, one Sunday morning, me and Mum and Dad went to Matalan to see if we could find a suit, but it was a waste of time. I tried on a jacket and it hung off the arms. It was too long and the jacket was like a skirt. I have got a curved spine (scoliosis) which means I lean over more on one side and some tops look funny and my chest pokes out too.

So, the funny thing was I looked in the mirror and we all started laughing. I looked really funny and the people who were walking by were laughing too – not at me, but with me because I was hysterical. I nearly wet myself. So, Mum said it was no good. We would have to go to a proper tailors to get a suit.

One day after school we all went to Mr Lennard, the tailor. He was really nice to me. He measured my legs. What a surprise – one leg was longer than the other, and the same with my arms.

It does upset me a bit because we all wanted to wear a top hat and tails but we couldn't. Mum put a penguin jacket on me and it hung down my arms again and the tail was like a train hanging on the floor. We all cracked up! It wasn't fair. We all ended up having to wear normal suits which we were all disappointed about because we couldn't get one to fit me right.

I was measured up a few weeks later. I went for our suit fitting and Mum put mine on and Dad put Ben's on. I looked in the mirror and Mum was nearly crying. She was happy and told me I looked handsome and looked like a prince. She said I did her proud.

The big day had arrived. We were all rushing around but my Mum was very calm. This made a change as normally she's stressing and running around like a headless chicken. My Nan got me dressed. When we were all ready we had to have photos taken. We had to stand this way, sit that way, smile, say 'Cheese!'. I was glad when we were finished.

Then the surprise came – a big white stretch limo came to pick us up. It was wicked. There was music inside and a TV. We had leather seats and we had a little champagne. It was very cool. I loved it! But then there was problem. The limo had trouble getting through the gates near the chuch. We kept reversing in, then coming back again. We tried again and five attempts later we were in. We all cheered.

I was so happy seeing my Mum and Dad happy and being here because when my Mum and Dad got married in 1997 I don't remember much because I was only 2 and a half years old and I hadn't long had a bone marrow transplant.

When we got into the church I found my seat right up at the front with Ben and my uncle Brian and Uncle Aaron. I had to sit between them so I couldn't get up to mischief. As if! I had to laugh when my Mum and Dad got their words wrong. Then there was a little boy running around. Normally it would have been Ben running around but he was very good, for a change.

When the service had finished it was the boring bit with all the photos. My back ached, my feet hurt with my new shoes and all I wanted to do was sit down for a little while. Unfortunately, there was nowhere to sit. I let everyone know that I'd had enough, so the photographer picked me up and sat me on one of the gravestones. I ws quite high up and I was very impressed as I was the only one who had a seat. Everyone else had to stand.

Everyone told me how smart I looked in my suit and they all wanted photos. It was getting boring so I may have pulled a few funny faces in some of the photos. Ben was also being a bit awkward and began sitting with his back to the camera.

A few weeks later we got the photos back. They were lovely and the day was so beautiful and so was my Mum. Here are some of the photos. As you can see I looked very smart and handsome, even if I do say so myself!

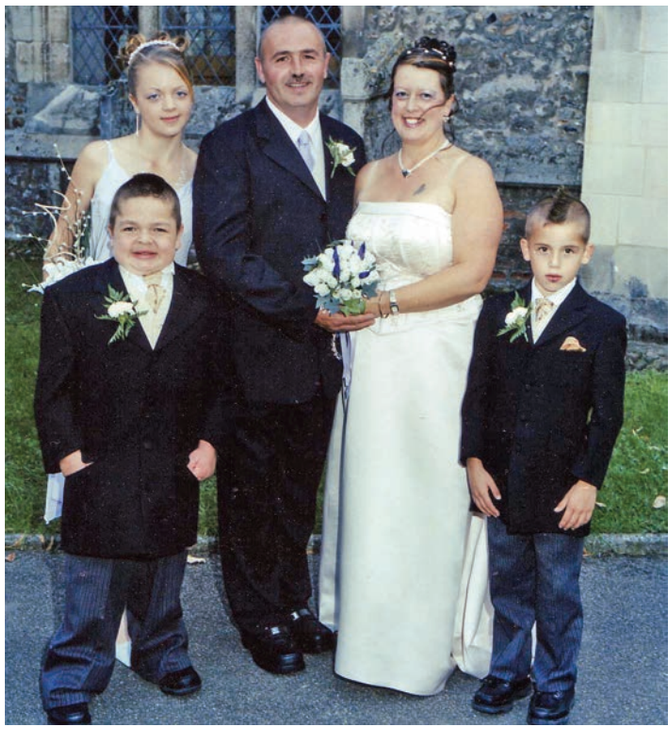


Photo courtesy of **ADK Photography** 01992 573091

International Workshop on Lysosomal Storage Disorders

Athens, 4 - 5 November 2005

On 3 November 2005, Christine and I flew to Athens to attend the International Workshop run by Shire (formally TKT). We arrived in Athens early in the evening and met various professionals from around the world for dinner before retiring to the bar to soak up the magnificent view of the Acropolis by night. The workshop began on the Friday, where various professionals gave presentations and up to date information on Fabry disease in females, exploring specifically whether outcomes can be predicted.

We were then given an update on Hunter disease looking at the natural history as well as receiving the results of the ERT clinical trials. These results were presented by Dr J Muenzer from the United States. Outcomes from the trial are positive and it is hoped that licensing will be approved by the end of next year. The day ended with us attending a dinner near the Acropolis.

Saturday was a very full programme with the workshop starting at 8.30am. The workshop was opened by its Chairs, Dr A Mehta and Dr U Widmer and solely looked at Fabry disease covering various topics such as clinical monitoring of ERT in Fabry disease; efficacy and safety including presentations on skin, eyes, kidneys and the heart. After lunch the focus was on children and the concept of early treatment and prevention in metabolic disorders.

After the afternoon break, Christine and I took the opportunity to meet with other MPS patient organisations present at the meeting. This included representatives from Italy, Spain, Norway, Germany and Sweden. These meetings are invaluable to all Societies, as they give people the chance to share information and ideas, as well as finding out how other countries are supporting their members, including the positives and negatives, the struggles and victories, especially with access to treatments. After this meeting it was a quick change for the gala dinner and again we were taken out for the evening and wined and dined. Before retiring, Christine and I headed for the bar to take one final look at the Acropolis by night.

The following day, we were flying home but before this we were afforded the opportunity of seeing some more of the sights of Athens including - yes, you've guessed it - the Acropolis by day!

After an exhausting weekend, we made our way to the airport where we flew back to England full of new found knowledge and understanding.

4th INTERNATIONAL FABRY CONFERENCE

by Nikki McAuliffe

Paris 2005

Firstly, I'd like to start by saying how lovely it was to finally be able to put names to faces after weeks of planning and organising in the build up to the event!

I think I'm safe in saying that the event went very well on the whole although, there was a bit of a panic before we even managed to leave the country what with the threat of delayed and even cancelled flights and, at the other end, the missing luggage saga!

Drama aside, much of Thursday was taken up with travelling and settling into the hotel, which I have to say was very impressive. Friday began with a brief welcome speech by the organisers and then went straight into a highly informative oratory from Dr Hughes about our understanding of Fabry so far and Enzyme Replacement Therapy. The following talks centred on the disease in Males, Females and Children which was interesting.

Friday afternoon was focussed on an international question and answer session. This allowed delegates to submit, and hopefully receive answers to questions that interested them.

One large dinner, a multitude of puddings and an early night later…suddenly it's 7.30 on a Saturday morning! Dr Sally Davis took the floor first to start the day with an impressive and thoroughly detailed speech about the genetics involved in Fabry. I think it's fair to say here that she set the precedence for the day as well as giving delegates the chance to get to grips with the fundamentals of Fabry. Afterwards came the updates from various national Fabry support groups allowing attendees to acknowledge the work of other patient organisations.

There was a choice in the afternoon between a meeting for young patients and national group meetings. We felt that it was best to form separate groups within the national meeting as it was a big group and people could then choose a group relevant to issues they wanted to discuss. On the whole, this was successful and many interesting points were drawn from it.

Afterwards, we were all taken on a whistle-stop tour of Paris' most famous sights. Our guide was very entertaining and although it may have been more enjoyable to have left the coach a bit more, it was a nice way to spend the latter part of the afternoon.

Saturday night's entertainment took the form of a gala dinner away from the hotel. The interior of the restaurant was very extravagant and I admit, on first sight I felt somewhat underdressed for the occasion!

Overall, I felt that this was a very successful patients meeting and I hope that those who came got as much out of it as I did! Once again, it was a pleasure to meet all the U.K attendees and I look forward to seeing you all again soon.

Here are a selection of photos from the Conference...

INTERNATIONAL NEWS

I was delighted to receive the invitation to attend and speak at the 4th International Fabry Patient Meeting in Paris in October. Having first persuaded my family that they could do without me, I proceeded to pack my bag and make my way to Paris. The meeting provided me with the opportunity to explain the role of Genetic Counselling for families with Fabry and emphasise the difficulties that may arise with confidentiality and consent surrounding information about an inherited disorder.

More important to me was what I learnt from the families present at the meeting. The difficulties that family members faced in obtaining a correct diagnosis as well as appropriate medical care was a salutary lesson. I valued the openness and the honesty of the patients keen to discuss their experiences. The realistic expectations of families in relation to therapy and their enthusiasm for ongoing research were very impressive.

One cannot visit a beautiful city like Paris without fond memories including the experiences shared with the many new friends I made during the meeting.

By **Sally Davies**, Deputy Director of Hospital Practice, Uni. of Wales College of Medicine

"Just a quick note to thank you all for making the Paris trip so fantastic, especially Sophie and Nikki. I had a wonderful time."
Love Sue Jenkins.

"Just a little note but a big thank you for all the hard work behind the scenes for the Fabry Anderson Conference in Paris. Once again we found it extremely beneficial and have made many new contacts with families abroad. The children's programme had good feedback from Glennon, Lyndon and Laura but they wished it had been on the first morning as it broke the ice a little too late in meeting new youngsters. Hope you all got home safely."
Juanita, Phil and Laura Davenport.

The Children's Hospice Association in Germany

Elizabeth Volk, Founder Member of the Children's Hospice Association in Germany, gives a personal account of the Association's development. This is the second part of her article, the first of which appeared in the Autumn 2005 Magazine.

A lot of hospice work is about subtle touches to people's lives. Many families take us up on our offer of balloons which are distributed and sent up at a child's funeral. It can be an uplifting moment to see these colorful greetings on their way to the heavens. Another of the quieter things our Society does, is that ultimately, when all is seemingly said and done and the years subsequent to the death of a child tally up, our Society still remembers the birthday of each of its children, if the family so wishes. We send out cards handmade by an artist who designs them especially for our Society, and more precisely, for our children. Many people relate to us their joy at receiving a birthday card in honor of their child, when other people have fallen silent, a card which expresses our gratitude that this particular child was a guest to this earth, and remains unforgotten in our hearts.

A saddening experience has been to see how many marriages have not made it over the course of the years. I know, a marriage is work in any case, but from our own experience we know that grief is a tremendously personal thing and that it does not necessarily bring you closer together. And of course, the whole problem often begins years earlier. How often does a mother at a children's hospice excuse her husband's absence there with, "I'm sorry he won't be coming along to stay with us. He just isn't ready to deal with all of this yet." And then I think, well, hats off lady, who asked you if you were up to this monumental task?

Finally, finally in these recent years in Germany the subject of palliative care is being addressed more and more with respect to pain control for children. Our personal experience tells us that there is nothing, really nothing, worse than seeing your child in pain. I sincerely hope the days are gone forever when a doctor says "Your child does not feel pain like we do. Or: If aspirin doesn't help, it isn't pain. Or: You'll just have to live with it."

Our own odyssey to a palliative ward only came about following months of seemingly endless misery. But once there, doctors were able to get our daughter pain-free in less than an hour, as they came up with a cocktail for various types of pain. I will be grateful for this for the rest of my life. Not only was our daughter, and later on, our son, made comfortable, we were able to stand being around them again. A child who is pain-free is certainly easier to care for at home. People around the child are much more relaxed and happy as well. In the meantime, the staff at children's hospice has become very alert to the possibility of pain.

The children's hospice we benefited from, Haus Balthasar, is very fortunate to have a doctor who is qualified in pain control for children. An anesthesiologist from a local hospital additionally trained in palliative care, he comes to visit the children at the house as needed. We feel very fortunate that this man treated our children so well. Pain control ultimately allowed them to die with such dignity.

Elizabeth Volk stands to the right of Christine Lavery. This photo was taken at the International Working Party on MPS Diseases, Frambu, Norway and the article is taken from a speech given by Elizabeth Volk.

Probably the most sensitive point for me in the years subsequent to the deaths of my own children has been meeting and listening to the younger parents in our Society. I recall only too well that in those earlier days I was always looking for someone to talk to about various matters, I myself never really wanted to be told what to do, the motto being something like: talk to me, explain things to me, but don't talk me into anything I don't want. I sincerely believe that in times of crisis, it is up to those surrounding the parents to explain the pros and cons of every decision in far-reaching detail, but after doing so, then it is strictly up to those parents to make the decision themselves, with us standing by them. It is not up to us to bully them and talk them into something. Only then is it possible for the family to say when it is all over, "Yes, it was good that way."

Whenever we were confronted with a tough decision, (and yes, it happened more than once, and yes, it is a terrible thing to constantly have to make decisions of life and death matters and speak for your own child,) nevertheless, it helped to always make sure we knew exactly why we decided the way we did at a certain time. Doing this, we never had to second guess our motives later on. Knowing that we acted as best we could at the given point in time really helps. It prevents the regrets and the "if onlys..." Hendrik and Emily have reached a fine and warm place in our hearts and in our memories only because we have no regrets about what could have been.

Advice, however well meant, can often be taken by sensitive and hurting parents as criticism. If they feel bullied into a decision they themselves have not reached and understood fully, then these are things that can be a problem later on. Of course, we were often unsure about decisions we made at the time, but the main point here is we made these decisions ourselves and we knew we were going to have to live with the consequences. Afterwards, the initial insecurities about those decisions just didn't matter any more because we knew why. I never doubt that people involved with hospice work mean well, but I do hope that we all continue to bear in mind the will of the family who must live with all the consequences of the decisions made. Even if it means swallowing your own need to cry out, "No, you can't possibly want to do it like this." It's something worth remembering, and I think it's about one of the most difficult aspects of people in the business of accompanying the other families. I myself have been guilty of it, all in the name of meaning well, despite knowing the whole time how hard it was for me to listen to other people's well-meant advice. Subordinating our own personal feelings in order to best serve others is a foundation stone of a well-functioning hospice and an aspect which demands constant vigilance in the education of volunteers.

This respect for the dignity of the family in reaching their own decisions goes hand-in-hand with the key word "flexibility" of the volunteers, caregivers and any of the rest of us who accompany the families on their way. This is a tall order to fill, at times bordering on virtually superhuman efforts on the part of the person on the outside, I know. But the families themselves living under such pressure should not be expected to be totally flexible in their encounters with children's hospice society and the services we offer. And often, the points where we can really help are when flexibility and understanding can be shown and we can meet the needs of a family in an unexpected emergency. I know these were the situations I was most grateful to get help in.

Our rapport with the children's hospice house was good enough that, even when our children weren't staying there, if a dilemma arose at home, which it did once or twice in the night, I felt like I knew where I could call and someone would be awake and willing to think the situation through with me. More than once calling emergency only to end up home again with no real change in the situation could be averted. You just have to do it, be willing to share your lives, and you may come out on top in the end.

There are many things about our lives within the Children's Hospice Society that I find gratifying today. Our Society publishes an annual magazine called "The Chance" and in so many ways, the opportunity to think things over has been just that. I am grateful that I learned so many things. From having to face fears and our innermost feelings head on, life took on a new reality, new depths charged with meaning. I came to find solace in symbols which fit our situation - the tree of life in our logo, the butterfly in its transitions from cocoon to a beautiful form of life, the hands of our children - beginning with the hands of Simon Lavery on the MPS logo, extending through the hands of all the children who have visited Haus Balthasar there on the wall, to the helping hands of the other children in the classes Hendrik and Emily attended at their school, the solidarity and love extended through the hands of my four children, and finally the right hands reaching heavenwards carved onto the gravestones of our children.

There was an empowerment which came with the knowledge of the inevitability of death, that you can prepare a few things beforehand if you want, if you don't want to be surprised by matters at the end, that you can express special wishes and have them granted, that you don't have to go careening into a situation, even if it feels true you are never really wholly prepared for it when the time comes. You can consider the examples of others who have gone before you and weigh the experience for your own situation. It was a tremendous relief to find that other people shared the same inner conflicts between emotion and reason, of being torn between tough decisions, and just being torn in every which direction. It helped get things into a perspective. Life could once again achieve a focus. And as I mentioned before, we learned that it is possible to stand with other families when their children die, and emerge from it stronger, less fearful ourselves, through their examples.

What I would wish our families is such a support beneath and behind them as they continue to write their life stories. What I would wish for our groups, be it MPS, children's hospice, or any other self-help group concerned with our children is that we function as a firm support for these families, that we are solid but unobtrusive, not the focus of the limelight but certainly there to do our job. Above all, I wish that we will succeed in keeping the families on their pedestal, treating them with all the dignity and respect they deserve in their difficult situations. ■

'AN EVENTFUL YEAR' Fer Pidden

2004 and 2005 has been a very turbulent and eventful year for us. Natalie, our 24 year old Sanfillippo daughter was in hospital at the beginning of 2004 with pneumonia. She has recovered remarkably.

My mother-in-law had a minor stroke and was in hospital for a few weeks during summer while her partner had to go to Dorothy House. We clocked up some miles doing the round trips every day. They both came home after a lot of home help was put into place. In September my mother-in-law's partner passed away after a long battle with prostate and bone cancer.

On December 17, Natalie was admitted to hospital again, this time with severe pancreatitus as a very rare side effect of Epilim. We were told to expect the worst. We spent Christmas by her bedside after she had made a miraculous recovery the day before. Natalie came back home on 12 January and had to be nursed back to her normal self after she had developed the most horrific sores on her bottom during her stay in hospital, after I left her alone for a few hours to go home to get refreshed. Rest assured, the hospital staff and all present in the ward knew and heard about it when I found her in bed, catheter detached, swimming in urine and faeces! Still, poor Natalie had to endure a lot of unnecessary pain.

While all this was going on in hospital in December, I had communication from the MPS Society to say that a family from Turkey had contacted them. I tried to e-mail them, but the address was wrong. They rang me, I was in hospital. My son tried to get the number, but missed a digit. Strangely though, the code number was for the city I come from and my mother still lives in. Anyway, during January we made contact and spoke on the phone.

I was going to visit my mother in Izmir, Turkey, at the end of March and beginning of April. I went and found them and spent a day with them. A lot of things have snowballed since then. I went to Oslo to the MPS Societies Conference and started to find contacts and families to form an MPS Society in Turkey. We also celebrated Natalie's 25th birthday on 1 October, which I think is a great landmark to reach.

This is a very long winded introduction to my next piece of writing, but I wanted to put things into context. I hope during the course of time, to write about my endeavours about doing something in Turkey regarding MPS families there. Here is a translation of the article this Turkish mother has written...

Emre's Story

Hello, I am Nalan Cetin, the mother of Emre who has Hunter Disease. I am going to tell you our story.

Emre was born on the 31 July 1994. His height and weight were normal. He had difficulties sucking from the first day he was born. I noticed afterwards that his upper palate was deeper then normal. I was only able to nurse him for a month. Later on he became a big baby (perhaps from the feed I was giving him). When he was 4-5 months old I used to get tired carrying him.

When he was 6 years old, the paediatrician told us that his hips were narrow and in the future he might have dislocated hips. For three months, his legs were put into orthopaedic straps, pulling them wide apart to either side. At eight months old he was still not able to sit up, swinging himself backwards, preferring to lie on his back. At 1 year old he was not able to walk, fearful of stepping down on his feet. At a year and a half he was trying to hold on to things as he tried to walk. He was not able to say more than mama, papa, and pointed at things he wanted to have.

One day when he had a high temperature we took him to a doctor. He referred him to a brain surgeon because of the largeness of his head. After the examination and the MRI scan, we were told that Emre had hydrocephalus and that

he had to have an operation immediately. We were devastated. We did not expect this. (How were we to know that this was the beginning of everything!). We agreed to have the operation and he had the shunt put in. I will never forget the day when we took him back a week later to have his stitches out. He was running along the hospital corridors as if he was demented. We were not able to stop him after that, It was as if he was making up for lost time, he was running everywhere.

Two months after the operation, he had developed hydrocele in his left testes and hernia. They told us this was the side effect of the shunt. He had another operation for hydrocele. They did not want to touch the hernia since it was near the shunt tube for the hydrocele and there was risk of infection. A month after the operation, his left testes began to swell up again. His tummy was extended. They did not want to operate again thinking that it might be a recurring problem. He waddled around with his testes down to his knees for about two years. This was affecting his quality of life adversely, but there did not seem to be a cure for him.

When we discovered that he had hearing problems, he was put on a long course of antibiotic treatment. When this did not solve the problem, T grommets were put in. Then his tonsils and adenoids were taken out. The audiology department wanted to keep an eye on him.

Emre was 3 and a half while all this was happening. Tests were very difficult to conduct. Even with medication, he would not go to sleep, and tests could not be conducted when he started to move. In the end he was referred to the genetics department. There we were told that his palate and teeth formation was different, he had sensorial deafness and they were suspecting he had MPS.

I will never forget that day. They showed us a photo of an MPS boy. It could have been my son's photo. We were in shock. At the same time I was seven months pregnant. During my pregnancy the only thing they tested for was hydrocephaly and I was told I was carrying a healthy baby daughter. Now they were mentioning an unheard of disease. Emre & I were referred to the genetics department.

There they sent Emre's urine and blood for tests to England. The doctor told us that it could be Hurler or Hunter; names we heard for the first time in our lives. Things were looking very bad and how was it going to end? I was told that it could be better if it was Hunter because of my pregnancy since it did not affect girls.

These were the worst 15 days of my life. My son was ill; my unborn daughter could be affected as well. What was this MPS? Why was anybody not explaining anything? The result after 15 days were Hunter Syndrome.

After this point, everything changed for me. First I could not believe. Then, I rebelled. I asked "Why my son"? (I could never find the answer). It took me some time to think rationally. After that I started to research the disease. My daughter was saved but alas, I was losing my son.

I learnt about the disease from the information I found on the Canadian MPS site. Unfortunately, there was nothing in Turkish on the internet. As we learned more about the disease, we realised that all of Emre's behaviour and problems were due to Hunter's.

Emre was hyper active. He would not sleep at night. He was nicknamed "Atomic Ant" in the school for learning difficulties that he attended. Even though he had swollen testes, he was still trying to run. In the end, when he was 6 and a half, his testes became hardened. He had to have an emergency operation. An operation that would have lasted 1-2 hours, took 5 hours and he had a job to come out of anaesthetics. During this operation, in order to support the membrane of the stomach, they placed a mesh inside the abdomen. Two months after the operation, the left testes started to swell again, but the doctors did not want to operate again until the last minute.

All of this would not deter Emre. He was full of joy. He liked visitors. He would make everyone laugh with his mischievous acts. 7 and half years old he stopped talking. 8 and half years old he lost his speed. He was not running anymore. He never managed to eat on his own. He would walk at home, but would not do so outside. He would get ill very quickly, run a temperature and have a blocked nose. The spasms and twitching he has been having for years increased.

10 and half years old, he started having seizures. He would choke in his sleep and start breathing with our help. Later he would go rigid and purple, and would relax and come out of it trembling and groaning. Doctors told us that because it was not originating from the brain, it was sleep apnoea.

He is now 11 years old and is not able to walk or sit up. He cannot eat solids. He chokes while he is drinking. He sleeps in the foetal position and bites his hands. I know that it is very difficult to live with these children, and all the stages families go through after they learn about the diagnosis and prognosis.

I congratulate all the MPS Societies who stand by the families and their children with all their problems. I heartily wish that my country will have one as well. ■

Ornella

The most marvellous treasure to fight for

Karen, Gad and Ornella (MPS III)

This has been a very special day and one which we will never forget but rather always remember as the “black Thursday”, 28th July 2005. One day before driving for our Summer holiday. One day before being with Ornella again, our six month old sweet daughter, our first baby. Ornella was already there with Gad’s parents, expecting us to come as soon as possible, with many dreams and many “first times” to look forward to: the first time to go to the beach, the first time to breathe the seaside air, the first time to listen to the gulls. We had rented a snug and cosy house and were really eager to share this first summer together. This day, I was having lunch with my Mum, when my mobile rang. It was raining so much this day. Gad, my husband, told me “Professor X has just called. He wants to see us by the end of the day.”

A couple of weeks before, at the 5th month visit, our paediatrician had decided to perform a number of analyses. Through a clinical exam, he noticed for the fourth time that Ornella had a slightly enlarged liver. This might be very normal for a newborn, he said, but he was anxious anyway. Experience and intuition were already telling him that Ornella might have something wrong but we did not know it. So we went through the analyses with some anxiety though not too worried at this point.

The first outcomes in July were very good. So much so that little by little, our initial fears turned into hope and confidence. On 28 July, even though we were still expecting the results of urine analysis, we were very confident that everything was going to be alright. Urine samples would just bring us confirmation that Ornella’s health was at its best, or so we thought. We did not have to worry about it! The only thing we really had to keep in mind was that we were going to spend the most wonderful summer of our lives! And that is exactly what we did have in mind this very special day.

Looking back now, we were actually going to enter the most frightening time we ever lived. What has followed Gad’s phone call resembles a long black tunnel. Gad picked me up. Suddenly, we had become intensely anxious, nervous, and a fear had overwhelmed us. It was raining again. Our appointment at the hospital was at 5.00 pm. We had a good couple of hours to spend, during which time we just kept on talking to kill the developing pain. After this interminable afternoon, we entered the hospital, and after a fairly long time, were invited to proceed to a simple and sad looking office. Professor X was sitting there, with empty indifferent eyes. He made a brief and rather obscure introduction. Then, he went to the main issue, and struck us with a litany of scientific issues concerning Ornella: her blood, skeleton, retina, and eventually, urine samples. The conclusion of all this, as far as we could understand, due to our own limited knowledge, was overwhelming. Ornella had just been diagnosed with MPS Type III A, otherwise known as Sanfilippo disease.

This disease occurs with a prevalence of 1 to 50,000 births, the Doctor said. Clinically the disorder is progressive and the diagnosis is usually made at 2-4 years of age when the children show upper airway obstruction, progressive deceleration of mental capabilities, hyperactivity, loss of speech… As the disease progresses the children become increasingly disabled, both mentally and physically.

The majority of affected children die in their second decade at the best. With Ornella, the diagnosis was set up with an utmost precocity, but nevertheless, no cure was to be expected at this stage and for the next coming years. There is nothing to say. For us, and for all the families that have to face such diseases, it is like being expelled directly to hell, with no good reason for that.

The following day, Gad and I drove to Normandy, making innumerable stops to wipe back the tears so that we could see. The moment when the door opened at our parents' was the hardest of our lives: Ornella was here, so much present, welcoming her parents with an adorable surprise and smile and a glance full of hope and innocence. I will never forget this very moment. Gad and I were crushed. But seeing Ornella's hope in life gave us the strength.

For seven years, my beloved husband and I have been sharing numerous and intense moments of love, happiness and intimacy. Now, together with our families, we have to share the unbearable fear and pain to be the impotent witnesses of Ornella's decline.

To win against the Sanfilippo disease, we decided to build up a deep knowledge of the disorder mechanism, stimulate the research dedicated to MPS III, unify our experiences, strengths and know-how, and make the best out of them. That is why we created the Alliance Sanfilippo in France.

The Alliance Sanfilippo is a patients' organisation, non-profit, non governmental and non political. The headquarters are based in France. The Alliance Sanfilippo though has a vocation to act without distinction of geographical area. It has a single objective: to defeat the Sanfilippo disorder. To do so, the Alliance Sanfilippo commits itself to unify and help the patients and their families, raise funds to sustain the research and undertake any action that has a chance to facilitate the fight against the disease. ■

Ornella, Karen & Gad

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The Ear in Fabry Disease

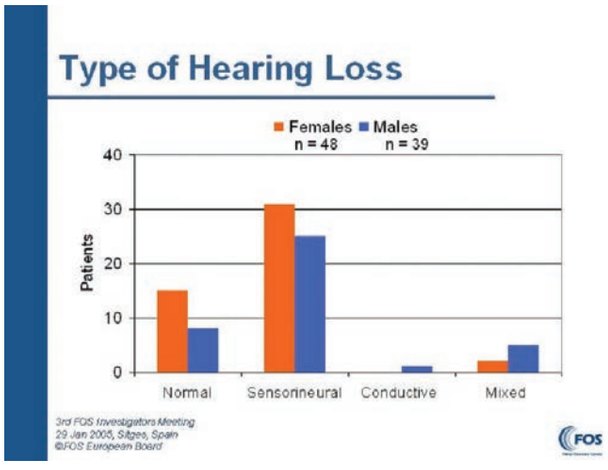
Derralynn Hughes and Atul Mehta
Lysosomal Storage Disorders Unit, Royal Free Hospital, London

Clinical Symptoms

Symptoms relating to ear pathology are common in Anderson Fabry disease and include hearing loss which may be sudden or progressive, vertigo (dizziness) and tinnitus (a sensation of ringing in the ears).

Historically the prevalence and nature of hearing loss has been unclear and to some extent under-recognised compared with more well known symptoms. In 2001 a survey of hearing symptoms performed by Dr Kay Macdermot and team found that 41% of men reported hearing loss and 38% tinnitus. 78% of men had abnormal hearing tests. This data was reproduced by French workers who also described a high incidence of sudden hearing loss, and later by a team in our unit who looked at patterns of hearing loss in patients prior to their commencing enzyme replacement therapy.

In this study Hajioff et al found that 80% of patients had abnormal hearing on one or both sides. The most common pattern suggested damage to the nerves which carry sounds from the ear to the brain however 12% patients had audiograms which indicated that the hearing loss was due to mechanical problems with sound conduction due to fluid in the middle ear space. Hearing has also been investigated in the Fabry Outcome Survey (FOS), a large European database of signs and symptoms in Fabry Disease. Most patients had nerve deafness but some patients were here also found to have conductive hearing loss or a mixed pattern of nerve deafness and conductive loss.



As with many symptoms of Fabry disease, males describe hearing problems at a younger age than female patients. By the age of 50 years about 70% men compared with 20% women describe hearing impairment. Sudden hearing loss is also more common in men and in total has been reported in about 5% patients in FOS compared to 1% of the general population. When asked about other symptoms about 30% patients describe vertigo and tinnitus which are also manifestations of nerve damage in the inner ear.

Pathophysiology

The ear is a complex and delicate sensory organ consisting of nerves, hair cells, blood vessels, bones and vibrating membranes all of which are vital for sound to be conveyed accurately and efficiently from the outside world to the brain where it is recognised and interpreted. Fluid-filled structures known as the semi-circular canals detect change in three dimensional position and are therefore important for balance.

There are many points at which these processes may be disrupted but the exact nature of the disruption in Fabry disease is not well understood. Deficiency of alpha galactosidase A in Fabry Disease results in accumulation of ceramide trihexoside (CTH, also known as GB3 and GL3) in many body cells including those in the kidney, heart, brain and eye. This results in characteristic structural and functional changes in these organs.

Damage to nerve cells, such as those in the ear, can be due to the direct effect of CTH storage or to damage to their blood supply due to storage in blood vessel cells. Nerve damage is most commonly suggested in association with the nerves which carry touch and pain sensations resulting in neuropathy and acroparasthesia. It is not known if the mechanisms of damage in the ear are the same as those occurring in peripheral nerves. Damage to the hair cells which detect sound vibrations in the inner ear is known to result from excess noise, toxic medications, infection, trauma aging and some genetic conditions. There may also be damage due to CTH accumulation. When these cells are lost in humans they are not replaced and

permanent hearing loss results. Finally impairment of hearing & balance might also occur if there is damage to an area of the brain involved in processing of sound or balance signals.

There have been few studies directly exploring the pathology of hearing loss in Fabry Disease. In 1989 Schachern and colleagues examined the changes found in the ears of two patients with Fabry Disease. They found CTH accumulation in the vessels and the nerve cells of the middle and inner ears. There was also hair cell loss, purulent fluid in the middle ears and evidence of thickening and fibrosis of membranes. Other clues come from features which have been found in association with hearing loss such as tortuous retinal vessels, high blood pressure, kidney disease, increased age and male gender. Statistically significant correlations have been found between hearing loss and renal and cerebrovascular, but not heart, disease.

The pure tone audiogram

The degree to which a patient suffers hearing impairment may be accurately quantified by means of a pure tone audiogram. This detects the loudness threshold at which sound can be heard at a number of different frequencies or pitches. Some conditions have characteristic patterns of pure tone audiogram where hearing is lost at a particular frequency. It has generally been thought that hearing loss in Fabry Disease is most prominent at the high pitched frequencies. However more recently it has become apparent that hearing loss occurs at all frequencies compared to age matched health control individuals. When this hearing loss is classified according to the World Health Organisation definitions only 22% patients would formally be said to have a significant level of hearing impairment however 77% have impairment across one or more frequencies.

Response to ERT

For any patient with hearing impairment related to Fabry disease it is of course important to know if there is an expectation of improvement with enzyme replacement therapy. Pure tone audiograms were collected as an extension of the original trial of enzyme replacement therapy conducted at the Royal Free Hospital. Audiology was performed every six months in 23 men and 2 women receiving enzyme for upto 3 ½ years: 8 males received placebo rather than enzyme for the first six months. After an initial decline, which was probably due to underlying progressive deafness, small improvements were subsequently noted. Since this initial study a positive effect of enzyme replacement therapy has been noted in a larger number of patients in the Fabry Outcome Study. This was of greatest magnitude in patients with mild to moderate hearing loss and appeared to occur at each frequency tested. Patients with severe hearing loss responded less well indicating that advanced damage may not respond so well to reduction in CTH storage.

Conclusions

Hearing loss is a common and disturbing complication of Fabry Disease occurring in up to 80% of patients. Males experience earlier and more severe hearing loss which is mainly sensori-neural and occurs at all frequencies. Progression of untreated disease occurs more rapidly than in the non-Fabry population and sudden hearing loss is five times more common than in the non-Fabry population. Small improvements in absolute threshold of hearing have been observed with enzyme replacement therapy and are best in ears with mild to moderate hearing loss. However despite progress in describing the natural history of hearing loss in Fabry disease further work is required to properly understand the mechanisms of disease and the full potential for improvement with enzyme replacement therapy.

Further Reading

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DVD: MPS and Anaesthesia

A Challenging Relationship

Patients with MPS will often require surgical interventions as part of their ongoing care. Anaesthesia is a risk factor in these patients due to a number of disease characteristics.

Genzyme have produced a new resource for anaesthetists, clinicians and paediatricians caring for patients with MPS which describes those challenges and explains how to maximise safety for the patient around the time of anaesthesia.

If you would like a copy, free of charge, to pass on to your local clinicians or anaesthetist please send a first class stamped addressed A4 envelope to the MPS Society with your request.

Great News!

On 27 October 2005 the government announced that means testing for the Disabled Facilities Grant (DFG) was to be abolished in England from December 2005.

This decision means that families with a disabled child will no longer be means tested and will be entitled to apply for the maximum grant, which currently stands at £25,000.

DFG's can meet the costs of improvements to the home, which include such things as extensions and adaptations.

If you would like further information on this or support with housing and adaptations please do not hesitate to contact a member of the Advocacy Support Team.

0845 389 9901

Understanding Home Improvement Agencies

Home Improvement Agencies (HIA's) were originally created by the government as locally based not-for-profit organisations that support home owners and private sector tenants who are usually disabled, elderly or on a low income to improve, adapt, maintain or repair their homes to fit their needs. Since their conception, HIA's have expanded and developed their services to incorporate the care health and housing sectors, as well as professionals such as Occupational Therapists, Grant officers and Social Services.

They are usually advertised as a cost-effective and people-centred service that helps individuals to stay in their homes and live as independently as possible. HIA's are often overseen by local authorities, housing associations, charities and independent management committees and can be known by other names such as 'care and repair' or 'staying put' schemes.

The primary purpose of these organisations is to help vulnerable individuals to identify problems or potential problems within their homes and to suggest how best these problems may be resolved in addition to reviewing all housing options and providing legal, welfare and financial advice.

Funding for adaptations and improvements is usually sourced through a Disabled Facilities Grant awarded from a local housing authority. However, alternative funds can be accessed by raising loans against the equity of the property and seeking advice from an Independent Financial Advisor.

Additionally, HIA's should offer guidance on the type and duration of the work required and assurance that any work undertaken will be completed to budget and on time.

A reputable HIA should ensure that specified building work receives competitive estimates sought from vetted contractors and that work is overseen by the agencies technical staff to ensure that the end product meets the client's needs and expectations.

However, there are a few critical issues pertaining to the use of Home Improvement Agencies that need to be raised in order for potential users to build a more lucid and unbiased opinion and understanding of the service. HIA's are marketed on the belief that their not-for-profit claim means that they make no money whatsoever from any work executed by themselves. Unfortunately, this isn't always the case, therefore it is imperative that, if you are seeking housing aid through a HIA, you read all the supporting evidence and documentation. HIA's have been known to charge additional payments and include financial clauses within their paperwork that will inevitably be taken out of the grant money that you have been awarded.

Once again, it is very important that before committing yourself to any HIA scheme, make sure that you have fully read and understood everything that you are putting your name to, and if you are unsure about any of the details you must ask.

Update to Changes to Education in Scotland By Clare Cogan

This article follows on from one written in the Spring 2005 magazine, detailing the changes to the system, which is currently in place for children who have additional needs at school. Currently, most children who have special educational needs have a Record of Needs in place that outlines their situation and resulting support needs.

It is now confirmed by the Scottish Executive that The Education (Additional Support for Learning) Act 2004 will become law on 14 November 2005. This means that from this date, children who have additional learning needs will no longer be assessed for a Record of Needs but a Co-ordinated Support Plan. How this new assessment process will work is outlined in the Additional Support for Learning Act - Code of Practice. This has been sent to schools, local authorities, health agencies and key voluntary organisations. Copies of this can be obtained by calling the Scottish Executive on 0131 244 7139 or downloading it from the publications section of the Scottish executives website, www.scotland.gov.uk.

Safeguarding Record of Needs provision

For children who already have an existing Record of Needs, there are safeguards in place to ensure that this is not removed following the new Act and the implementation of Co-ordinated Support Plans. From the date the Act commences, Education Authorities must ensure that the provision being made in school, as detailed in the Record of Needs prior to the Act remains the same and is not reduced. They then have two years to reassess the child to ascertain whether they meet the criteria for a Co-ordinated Support Plan, which will replace the child's Record of Needs eventually. Children will meet the criteria if they have enduring, complex or multiple barriers to learning and require support from a number of different agencies, including health and education. If the education authority decided that the child is not eligible for a Co-ordinated Support Plan then parents will have the right to appeal to the Additional Support Needs Tribunal.

As ever, the Advocacy Team is happy to support families who are concerned how this reassessment will impact on their child's support in school and to provide additional information, where available on the Act and its implications. Please do not hesitate to contact the MPS Office if further support or information is required.

If you would like to contact the Society's advocacy team telephone 0845 389 9901 or email advocacy@mpssociety.co.uk

"Telling Stories, Understanding Real Life Genetics" - Helping educate nurses about genetic conditions

A new website is being created to help nurses, health visitors and midwives learn about genetics and help them understand the effect genetic conditions have on people's lives.

The project called 'Telling Stories: understanding real-life genetics' will collect around eighty stories from individuals, families and health professionals who are affected with different genetic conditions, or have a particular story to tell about their experience in caring for people with a genetic condition. The stories will be collected as video clips, sound recordings and written accounts of peoples' experiences.

Running the project is a team from the University of Glamorgan, University of Plymouth, the Wales Gene Park and the Genetic Interest Group. Project leader, Professor Maggie Kirk said: "This website will help the nursing professions to understand more about how living with a genetic condition can affect a person and their family's life. Being able to understand a patient's viewpoint will help a nurse to care for their patient."

We are now looking for people to participate in the project and tell their story. Are you affected by a genetic condition, or are you a family member or health professional? If you would be willing to tell us your story, we would be happy to hear from you. For further information, please contact us at the details below:

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A QUALITATIVE INVESTIGATION OF FATHERS' EXPERIENCES OF LOOKING AFTER A CHILD WITH A LIFE-LIMITING ILLNESS, IN PROCESS AND IN RETROSPECT: EXECUTIVE SUMMARY

Jane Ware, July 2005

1. Background to the study

1.1 Introduction

This study is concerned with the exploration of fathers' experiences of having a child with a life-limiting illness.

Life-limiting illnesses (LLIs) are those for which there is no reasonable hope of cure and from which children will die (Sutherland et al., 1994). Recent advances, both enabling earlier diagnosis of illness and in medical techniques, have resulted in children surviving longer and a growing population of children with life-limiting illnesses (Association for Children with Life-Threatening or Terminal Conditions and their Families (ACT), 2001).

Reporting of epidemiological studies by ACT reveal that in a health district of 250,000 people with a child population of 50,000, in one year between 60-85 children are likely to have a life-limiting illness, about half of whom will need active palliative care at any one time (ACT, 2003). The annual mortality rate from LLI is 1 per 10,000 children aged 1-17 years (Goldman, 1998).

Life-limiting illness has only recently separated from chronic illness to become a focus of inquiry in its own right, thus the research to date is extremely limited; furthermore, studies often have failed to distinguish clearly the type of illness (chronic illness, life-threatening illness or life-limiting illness) being investigated.

A review of the literature revealed that research has largely focussed on stress and coping models, with the aim of trying to identify predictive variables (both child and parent related) for parental maladjustment and to identify coping strategies used by parents, especially those which might mitigate against the stress of having a sick child. Thus, often central to the quantitative research remains a focus upon psychological pathology through the measurement of maladjustment and coping. The limited qualitative studies available to date aim to focus upon the process, sequence and meaning of the experience of parenting a child with a life-limiting illness.

Mothers have received far greater attention than fathers although comparisons of coping between parents have also been examined. Studies have failed to address how fathers experience having a child with a LLI. Outcome variables seem to have been chosen prior to exploring how men themselves actually think about their experience. Thus we seldom learn of the 'common and unique nature of men's experiences' (Chesler & Parry, 2001 p.363).

The overall picture to emerge from the wider research literature is one of fathers showing less outward distress than mothers, fathers tending to focus on helping their spouse to cope with the crisis, and hiding their concerns about their child in order to help provide emotional support to their partner. They also strive to maintain a feeling of control through their work outside the family.

Significant gaps in the literature remain. For example, quantitative studies in particular have failed to distinguish between short-lived episodes of stress and long-term stress resulting from living with a child with a LLI. The impact of different illnesses and their different trajectories and the resultant service implications have yet to be addressed.

My clinical work with families with children with life-limiting illnesses, intervening both directly with families and consulting with and supervising other professionals providing services to these families, has also highlighted a difference in levels of engagement with services between fathers and mothers. In my experience mothers are far more likely to request psychological input for themselves and other family members (particularly their partner) than fathers. Having frequently heard comments, from mothers and from professionals, that men are perceived to need help for emotional difficulties related to their child's illness, but that they either do not identify the need themselves or would not find it acceptable to request or utilise such help, I began to question whether this was actually the case. I wanted to step back and understand what the experience of having a child with a life-limiting illness is like for fathers, and its impact upon them and how they perceive it has affected their family relationships prior to engaging with the question of whether services need to be made more available to them and if so how this might be achieved.

1.2 Purpose of the research

This study aims to give voice to fathers' of children with a life-limiting illness and to explore their stories in detail. It was hoped that the method of enquiry would facilitate the teasing out of the many and complex emotional, social, political and cultural influences impacting on fathers.

1.3 Research objectives

The study aimed to elicit an understanding of fathers':
1. Subjective experience of parenting a child with a life-limiting illness.
2. Their sense of the impact this has had on them.
3. To gain their views about how well service provision has met their emotional and psychological needs.

2. Method

2.1 Sample

Eight fathers were interviewed; two of their children had died and seven were still living. No restrictions were placed on the time since diagnosis or the actual diagnosis. The men were recruited mainly through voluntary organisations.

2.2 Data collection

A semi-structured interview schedule was designed and piloted. The in-depth interviews were tape-recorded and they lasted between 60-120 minutes.

2.3 Data analysis

The interviews were transcribed and analysed using a qualitative approach, Interpretative Phenomenological

Analysis (IPA) (Smith, et al., 1999). Each transcript was examined in detail; a list of emerging themes was noted and from this clusters of themes or super-ordinate concepts were developed for each interview - these themes were continually cross-referenced with the transcript to ensure they were grounded in the data. Finally a list of themes and their sub-themes for all the interviews was developed.

3. Findings

Four super-ordinate themes emerged from the data and the main findings from each are presented.

Main theme 1: Diagnosis turns the world upside down

All the participants could vividly recall the events leading to a diagnosis and being given the actual diagnosis, regardless of how long ago it happened.

Three had had to fight to get professionals to listen to them and two had sought private appointments, as the NHS had been so slow to provide an assessment.

A sense of alienation and abandonment was keenly felt by most of the men. Also a sense of their life having been tainted permanently, and their naivety and ability to really trust anything anymore had been lost forever. Three participants identified the rareness of their child's illness as increasing their sense of alienation. Few professionals had the specialist knowledge to help them and few families in similar situations were available to offer advice or support. Despite this, many felt it had later changed them for the better, leading to personal growth and a change in what, and how, they valued in life.

Six of the participants identified problems with professionals and service providers. Inadequate follow up after diagnosis, insufficient support and poor communication between professionals and across services exacerbated their sense of a lack of containment.

Main theme 2: Living with what has happened

This main theme emerged from the participants' descriptions of their attempts to come to terms with the diagnosis, to accommodate to it over time, and their assessment of how well or otherwise they have coped with this unwished for change in their lives.

All the participants talked about how they sought out further information about their child's condition soon after being given the diagnosis. In some cases they talked about needing to do so partly because professionals had given them insufficient information, but for all of them it seemed to be a useful coping strategy, a way of feeling they were taking charge.

All of the fathers spoke of, or implied, a heightened feeling of love towards their ill child, wanting to make the most of their chance to spend time with them and enjoy their relationship; thus, despite being acutely aware that the relationship would end prematurely, none of the participants seemed to have responded to this by emotionally distancing themselves from their child at any time.

The participants talked about their relationships with their child's mother or partner and in all cases this relationship seemed to have been intensified, in some cases strengthened but in others fractured, as a result of their child's illness.

The participants who had other children commented briefly on the impact of the LLI on the sibling(s). They all expressed a desire to be as open as possible with the other children about their sibling's condition and were aware of the emotional distress this caused the children.

The participants identified relatively few coping strategies that they had successfully utilised. The most frequently mentioned ones included: the importance of having a close, confiding relationship with their partner (although that was not necessarily their child's mother); the importance for them of talking and sharing with others outside of the family; support groups for their child's particular condition. It is interesting, and perhaps surprising, to note that relatively few of the men voiced feelings of their masculinity being undermined as a result of fathering a child with a LLI.

Main theme 3: Men are different

No question in the interview schedule directly raised the issue of gender differences, yet interestingly all of the participants firmly identified such differences and wanted to discuss them. All the participants identified a gender difference in how they respond emotionally and how this affects their ability to cope.

They all perceived men as not allowing themselves to explore their emotional responses in the same way as women do. They considered this to be particularly strong in their relationships with other men.

Five of them explicitly commented on men having to have a public face of being strong and coping, even though privately they might be in considerable turmoil. The perceived need to mask their true feelings from others, and indeed from themselves, was generally thought to be an unhelpful mechanism.

In contrast, they perceived women as being able, and sanctioned if not actively encouraged, to more readily express their intimate and inner thoughts. The participants implied, and often stated, an envy of women's ability to be able to more openly acknowledge and express their feelings.

They perceived women as generally coping better than men because they have more effective ways of handling their emotions and are able to access more varied coping mechanisms; women have more opportunities, including wider social networks, than men to gain support.

The participants identified that in order to change men's attitudes and to encourage them to show their emotions more readily a major shift in attitude at a societal level would be required.

Men believe that services are geared to women: appointments are arranged during their working hours and getting time off work is often difficult; professionals often do not readily include fathers in the consultation or treatment process, especially if they are separated from the child's mother. Men may have specific or different needs from mothers and service provision does not acknowledge or provide for such differences.

Most of the participants, whilst acknowledging that some fathers might continue to absent themselves from the process, felt that there might be major gains to be made

by shifting the current focus of service provision from mothers to both parents.

Main theme 4: The wish to make it easier for others
The participants wanted to tell their stories, at least in part, so that they might be able to contribute to changing and improving how future fathers might cope with having a child with a LLI. They hoped that some of the restraints and problems which they had encountered, particularly in relation to emotional and coping aspects and service provision could be identified and resolved.

The participants identified ways in which service provision could be improved. Some indicated a lack of trust in services and most highlighted bureaucracy and staff changes as problematic. Appointments, particularly health ones, are held at inflexible times, and usually involve long periods of seemingly unnecessary waiting. Staff resources often seem inadequate to deal with the needs of the patients and their families. Participants commented on being deluged with appointments and feeling that poor communication between services led to duplication and a lack of clarity and responsibility.

They advocated the introduction of a key worker system; they believed one individual with whom they could communicate about all aspects of their child's care and who would then liaise with all the other agencies and professionals would significantly improve service provision and continuity of care. They felt men should be given greater opportunities, independently of the child's mother, at the time of and following the diagnosis, to discuss their child's illness in detail.

Service providers and professionals needed to understand that men often find it harder than women to acknowledge that they are struggling emotionally, that they have learnt effective defensive and protective ways of brushing off suggestions that they might need help and this results in men's distress often being ignored or overlooked and men being unable to access help.

Service providers should take these differences in men's needs into account, think more subtly about relevant and appropriate provision for men, and find more effective ways of engaging fathers. This includes the recommendation of more flexible types of provision, particularly counselling related, which men could opt in to as and when they felt able to. The importance of support remaining available on an open access basis was frequently highlighted.

4. Conclusions

All the fathers were profoundly affected and in various ways by their child's life-limiting illness. The interview data revealed that from the initial diagnosis onwards the fathers struggled to adapt to their changed world and circumstances. They lived with ongoing distress and stress, the intensity of which varied over time and their child's condition, but never completely disappeared. The participants adopted a mainly gendered account as a way of explaining their experiences and coping strategies. They felt strongly that men's stories and needs have been largely unheard and unacknowledged and that services and clinicians need to change in order to address men's emotional and psychological needs. They identified specific ways in which these service issues might be addressed.

5. Recommendations arising from the study

i) Professionals working with families of children with life-limiting illnesses need to pay specific attention to the emotional and psychological needs of fathers, with a view to providing a range of interventions and approaches aimed at addressing these needs.

ii) Services should consider the early introduction of a key worker system as a means of improving the integration and coordination of provision for families of children with life-limiting illnesses.

iii) Further research exploring men's perceptions of both service and therapeutic interventions, and the complexities of providing relevant and appropriate provision with which men will engage, is required.

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Acknowledgements

I am particularly grateful to the men who took part in this research. I very much appreciated their interest in, and support for, this study and I hope I have done justice to their wish to ease the suffering of other fathers. My thanks to Lizzie Chambers at the Association for Children with Life-Threatening or Terminal Conditions and their Families for her support in the early stages of the project. Finally my sincere thanks to the staff of illness support groups, hospices and other services without whose help I could not have completed this research.

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Psychosocial Research Update:

Present Progress and Future Plans Cheryl Pitt

The research project Psychosocial Outcomes of BMT for MPS IH, which many of the families with children affected by MPS IH who have undergone bone marrow transplant took part in, is now coming to an end. The data have been analysed and the results are currently being written up. When this is complete, several papers will be written for publication in academic journals. However, in addition to this, booklets, information sheets, and newsletters will be written so that the information gleaned from the study can be disseminated to parents, schools, and health care providers at the local level. Once again I would like to thank all the parents and children/young people who took part in the study for their time and effort, without which the research could not have taken place.

As of April 2006 I will be embarking on a five-year research programme, which will continue to focus on the psychosocial aspects of living with MPS and related diseases. Over the next five years a further three conditions will be studied - MPS IV-A Morquio disease, Fabry disease, and MPS III Sanfilippo disease. In particular, the research will explore:

The Psychosocial Adjustment of Individuals Affected by MPS IV-A Morquio Disease

Due to the rarity of this condition, literature regarding the psychological adjustment of patients is limited. However, due to the physical manifestations of the condition, namely short disproportionate stature and skeletal dysplasia, paired with normal intelligence, there is a potential for emotional difficulties, which can have a major impact on a person's functioning within society. In order to maximise healthy psychosocial development it would be useful to explore the emotional experiences of individuals affected by Morquio disease from childhood into adulthood. This will enable us to ensure that appropriate psychological support is provided.

Are The Psychological Sequelae of Fabry Disease Relieved by ERT?

Very little research has explored the psychological distress that is associated with Fabry disease. However, the little research that has been carried out clearly illustrates how psychiatric disorders, most commonly depression, complicate the disease. Enzyme Replacement Therapy has shown promising effectiveness in treating Fabry disease, and has widespread therapeutic benefits. However, the


Cheryl Pitt

extent to which the psychological sequelae of Fabry disease are relieved by ERT is not clear. This study will not only bring us closer to determining the frequency and aetiology of psychological complications in patients with Fabry disease, it will also help to demonstrate whether treatment has a direct or indirect effect on symptoms of psychological distress.

Living with Sanfilippo Disease: A Sibling's Story

Quite a lot of research has explored the experiences of children who are siblings of a child with disabilities, including learning disability. A lot of research has also focused on children's experience of living with a terminally ill sibling, most notably cancer. Such research highlights psychological and behavioural problems, however it also illustrates how some children have positive experiences and see benefits in terms of psychological adjustment and learning. To date, siblings' experience of living with Sanfilippo has not been studied. Since this disorder encompasses physical and learning disability, and challenging behaviour, while also being a progressive and terminal illness, the experience of the sibling certainly warrants investigation, so that appropriate support can be given.

From April 2006 families who are affected by these conditions, and meet the criteria for the studies, will be sent further information about the projects and will be invited to participate. I very much look forward to meeting and getting to know you, and will be in touch very soon.

Shire files Elaprase ™ (idursulfase) with the FDA for the treatment of Hunter syndrome

Philadelphia, PA, US, and Basingstoke, UK – November 24, 2005 -Shire plc (LSE: SHP, NASDAQ: SHPGY, TSX: SHQ) announced today that it has submitted a Biologics License Application (BLA) with the U.S. Food and Drug Administration (FDA) for idursulfase under the tradename ElapraseTM, formerly referred to as I2S. If approved, ELAPRASE would be the first human enzyme replacement therapy for the treatment of Hunter syndrome, also known as Mucopolysaccharidosis II (MPS II). Idursulfase has previously received Fast Track designation from FDA, and Shire has requested Priority Review of this submission, which would result in a six-month review. Submission to the European Medicines Agency (EMEA) is anticipated before the end of 2005 and typically takes 12 months for review and approval.

"Our filing with the FDA is a milestone for Shire and our team, who have been steadfast in the research and development of this much needed treatment," said Dr. David D. Pendergast, executive vice president and general manager of Shire Human Genetic Therapies, the Shire specialty unit focused specifically on genetic diseases. "We are now a significant step closer to helping patients and their families living with Hunter syndrome, and I am pleased that this application has been submitted on schedule and I look forward to approval and subsequent launch in 2006."

The BLA contains data results of the pivotal AIM (Assessment of I2S in MPS II) study, which studied 96 patients over 52 weeks, and is the largest study ever conducted for a MPS disorder. The primary efficacy outcome assessments were distance walked during six minutes (6-minute walk test, or 6MWT) as a measure of endurance, and percent predicted Forced Vital Capacity (FVC) as a measure of pulmonary function. The primary endpoint combined these two components into a composite score. Patients who received 0.5mg/kg of ELAPRASE on a weekly basis showed a statistically significant difference (p=0.0049) in the primary efficacy endpoint, compared to patients receiving placebo. Additional data demonstrated improvements in key secondary endpoints.

Treatment with ELAPRASE was generally well tolerated by patients in the trial. The most common adverse events observed were associated with the clinical manifestations of Hunter syndrome. Of the adverse events considered possibly related to ELAPRASE, infusion related reactions were the most common and were generally mild. There were two patient deaths during the study, both of which were considered to be unrelated to treatment with ELAPRASE. No patient withdrew from the trial due to an adverse event considered related to ELAPRASE, and to date all patients have agreed to continue in the extension study.

The BLA also contains data from a randomized, placebo controlled, dose escalation Phase I/II clinical study. Safety data is derived from the two controlled trials, as well as from two extension trials in which some patients have been treated for up to 42 months. Ultimately, patients will be transitioned from the extension studies to the Hunter Outcome Survey (HOS) Registry, which will allow long term follow up of both safety and clinical outcomes.

For further information please contact: Investor Relations Cléa Rosenfeld (Rest of the World) +44 1256 894 160 Brian Piper (North America) +1 484 595 8252 Media Jessica Mann (Rest of the World) +44 1256 894 280 Matthew Cabrey (North America) +1 484 595 8248

Shire is committed to helping patients and families with Hunter syndrome.

Further information about Hunter syndrome is available at http://www.hunterpatients.com.

Amicus Therapeutics is a clinical stage bio-pharmaceutical company developing small molecule, orally active drugs to treat a wide range of human genetic diseases. Amicus develops pharmacological chaperones - oral therapies that bind to the affected proteins, restoring shape, proper trafficking, and biological activity.

Amicus' innovative therapies apply to conditions in which crucial proteins are defective as a result of improper folding. Instead of trying to replace these complex proteins, Amicus' paradigm-shifting approach uses pharmacological chaperones to selectively bind and "rescue" the misfolded target protein to restore its proper conformation, trafficking, and biological activity, which in turn restores the function of the affected cells. This unique technology represents the next-generation approach to the management of human genetic diseases, and offers the potential to dramatically improve treatment options for patients.

For more information regarding the clinical study opportunity currently available in the UK for men with Fabry Disease please visit the MPS Society's website: **www.mpssociety.co.uk**

Amicus Therapeutics, Inc.
6 Cedar Brook Drive, Cranbury, NJ 08512 • 609-662-2000
www.amicustherapeutics.com

Shire files idursulfase with EMEA for treatment of Hunter syndrome

Basingstoke, UK and Philadelphia, PA, US,– December 1, 2005 -- Shire plc (LSE: SHP, NASDAQ: SHPGY, TSX: SHQ) announced today that it has submitted a Marketing Authorization Application (MAA) to the European Medicines Agency (EMEA) for idursulfase for the treatment of Hunter syndrome. Review of a MAA by EMEA typically takes 12 months. If approved, this would be the first human enzyme replacement therapy for the treatment of Hunter syndrome, also known as Mucopolysaccharidosis II (MPS II).

“Following last week’s filing in the United States, our MAA filing with the EMEA is another key milestone in bringing to market a treatment for patients and families around the world living with Hunter syndrome,” said Dr. David D. Pendergast, executive vice president and general manager of Shire Human Genetic Therapies, the Shire specialty unit focused specifically on genetic diseases. “I look forward to approval and subsequent launch in Europe in late 2006 or early 2007.”

As previously announced, Shire filed idursulfase under the tradename ELAPRASETM with the U.S. Food and Drug Administration (FDA) on November 23, 2005 under a Fast Track designation and has requested Priority Review of that submission, which would result in a six-month FDA review.

For further information please contact: Investor Relations Cléa Rosenfeld (Rest of the World) +44 1256 894 160 Brian Piper (North America) +1 484 595 8252 Media Jessica Mann (Rest of the World) +44 1256 894 280 Matthew Cabrey (North America) +1 484 595 8248

Hunter syndrome, also known as Mucopolysaccharidosis II (MPS II), is a rare, life threatening, genetic disorder with no available treatment. Individuals with Hunter syndrome lack the enzyme iduronate-2-sulfatase, which is essential in the continuous process of replacing and breaking down glycosaminoglycans (GAG). As a result, GAG remains stored in cells in the body causing progressive damage. The symptoms of Hunter syndrome are usually not visible at birth, but usually start to become noticeable after the first or second year of life. Often the first symptoms may include hernias, frequent ear infections, runny noses, reduced growth rate and abnormal facial appearance.

As the disease progresses, a variety of symptoms appear including enlarged liver and spleen, heart failure, decreased endurance, obstructive and restrictive airway disease, sleep apnea, joint stiffness, and, in some cases, central nervous system involvement. If central nervous system involvement exists, the life expectancy for patients with Hunter syndrome is typically 10-15 years of age, however, some patients can survive into the fifth or sixth decade of life. There is currently no effective therapy for Hunter syndrome. Idursulfase is a human iduronate-2-sulfatase produced by genetic engineering technology, developed to replace the missing enzyme in Hunter syndrome patients. Idursulfase has been designated an orphan drug in both the United States and in the European Union. Shire believes there are approximately 2,000 patients worldwide afflicted with Hunter syndrome in countries where reimbursement may be possible. Shire is committed to helping patients and families with Hunter syndrome. Further information about Hunter syndrome is available at www.hunterpatients.com.

SHIRE PLC Shire’s strategic goal is to become the leading specialty pharmaceutical company that focuses on meeting the needs of the specialist physician. Shire focuses its business on central nervous system, gastrointestinal, general products and human genetic therapies - all being areas in which Shire has a commercial presence. The structure is sufficiently flexible to allow Shire to target new therapeutic areas to the extent opportunities arise through acquisitions. Shire believes that a carefully selected portfolio of products with strategically aligned and relatively small-scale sales forces will deliver strong results. Shire’s strategy is to develop and market products for specialty physicians. Shire’s in-licensing and merger and acquisition efforts are focused on products in niche markets with strong intellectual property protection either in the US or Europe. For further information on Shire, please visit the Company’s website: www.shire.com.

"SAFE HARBOR" STATEMENT UNDER THE PRIVATE SECURITIES LITIGATION REFORM ACT OF 1995 Statements included herein that are not historical facts are forwarding-looking statements. Such forward-looking statements involve a number of risks and uncertainties and are subject to change at any time. In the event such risks or uncertainties materialize, Shire plc's results could be materially affected. The risks and uncertainties include, but are not limited to; risks associated with the inherent uncertainty of pharmaceutical research, product development, manufacturing and commercialization; the impact of competitive products, including, but not limited to, the impact of those on Shire plc's Attention Deficit and Hyperactivity Disorder ("ADHD") franchise; patents, including but not limited to, legal challenges relating to Shire plc's ADHD franchise; government regulation and approval, including but not limited to the expected product approval dates of DAYTRANATM (MTS/METHYPATCH) (ADHD), SPD503 (ADHD), SPD465 (ADHD), MESAVANCE TM (SPD476) (ulcerative colitis), ELAPRASE TM (idursulfase) (Hunter syndrome) and NRP104 (ADHD), including its scheduling classification by the Drug Enforcement Administration in the United States; Shire plc's ability to benefit from the acquisition of Transkaryotic Therapies Inc.; Shire plc's ability to secure new products for commercialization and/or development; and other risks and uncertainties detailed from time to time in Shire plc's and its predecessor registrant Shire Pharmaceuticals Group plc's filings with the US Securities and Exchange Commission, including Shire Pharmaceuticals Group plc's Annual Report on Form 10-K for the year ended December 31, 2004.

Society for Mucopolysaccharide and Related Diseases

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