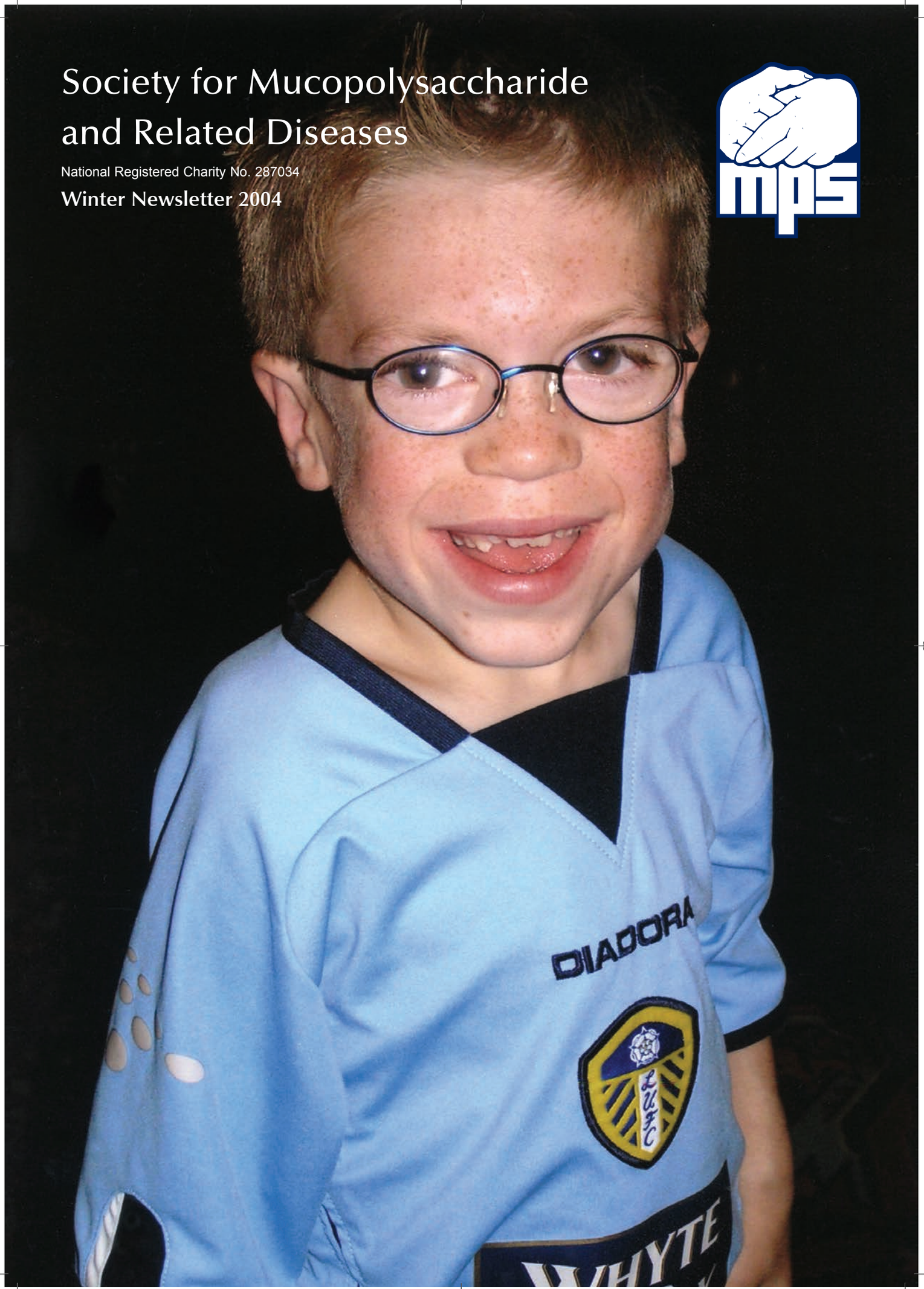
Society for Mucopolysaccharide and Related Diseases

National Registered Charity No. 287034

Winter Newsletter 2004



What are Mucopolysaccharide & Related Diseases?

Mucopolysaccharide & 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 1000 children and adults suffering from Mucopolysaccharide and Related Lysosomal Storage 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. The Society has the following aims:

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

How does the Society meet 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 Young People & Adults 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 including a video

Quarterly Newsletter

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

Front cover photograph: Oliver Moody (MPS VI)
Oliver's story is featured on pages 18-19

Newsletter

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Registered Charity No. 287034

Newsletter Deadlines

Spring	1 Mar 2005
Summer	1 Jun 2005
Autumn	1 Sep 2005
Winter	1 Dec 2005

Subscriptions

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

Contents

Editorial

Welcome to the Winter 2004 edition of the MPS Newsletter. Our cover star is Oliver Moody who writes, together with his father Paul, about living with MPS VI, Maroteaux Lamy Disease, on page 18.

This newsletter is a bumper edition featuring stories from the lucky winners of the Disney draw who attended the MPS Disney Conference in December. Everybody seemed to thoroughly enjoy themselves and have written about their time at Disney so that we too can share a little bit of the Disney Magic!

Please keep your stories, photos and news coming in for future editions. The deadline for the Spring newsletter is 1 March 2005.

Antonia Crofts, Editor

THE ROALD DAHL FOUNDATION

This MPS Newsletter has been funded with the kind support of a grant from the Roald Dahl Foundation.
www.roalddahlfoundation.org

Chief Executive's Report

For the last three months work at the MPS Society has been frenetic. The Advocacy Team has worked with a record number of our members offering support on a whole range of issues including special educational needs, home adaptations, child protection, disability benefits and accessing new therapies.

Accessing Enzyme Replacement for MPS I, MPS II, MPS VI and Fabry occupies the minds of many of our member families. In December 2003 the Society led a meeting between medical experts in these diseases and interested Members of Parliament. Those attending the meeting committed with the Society to taking our case for central funding for Enzyme Replacement Therapy for Lysosomal Storage Diseases to the heart of Government. This was achieved in March 2004 when Dr Ed Wraith, Dr Atul Mehta and I, accompanied by supportive MPs, met with the Minister of Health, John Hutton to put forward our case. Anxious months passed and during this time the Society supported members through appeals to their Primary Care Trusts (PCTs) and on the pathway to Judicial Reviews of their cases. Using these processes almost every one achieved funded ERT. In July 2004 the Department of Health announced that Lysosomal Storage Diseases are to be managed under the National Specialist Commission Advisory Group (NSCAG). This is good news for all children and adults with MPS and should result in a very clear protocol of the clinical, ethical and social management of these diseases. However, the costs of ERT were not to be included in this NSCAG designation so on we marched. Ellie and I attended many meetings in September and October ready to put errant speakers on the straight and narrow. Finally, whilst in Toronto attending a series of meetings, I received news that the Minister of Health back in London had agreed to include the drug costs of ERT in the NSCAG designation. It had taken ten months but it was worth it.

Some of you may be asking, but what use is NSCAG designation to us where there is no treatment immediately on the horizon? Well, NSCAG should be of considerable benefit. As I mentioned earlier, a document, rather like a road map, is being developed between the Department of Health (DoH), NSCAG designated centres and the Society. The Society has met with the Department of Health to comment. Areas of concern raised include clinical management at transition and in adulthood; admission policy for young adults with neuro-degenerative MPS conditions i.e. Sanfilippo Disease, where the young person requires 1:1 care management. We raised with the DoH situations where our young adults have found themselves accommodated as inpatients on geriatric wards. Other points are the need for accessible information during consultations. It may be that English is not the patient's or family's first language or that the patient is visually or hearing impaired.

As you will read further on in the newsletter the Society facilitated 39 members with Fabry Disease to participate in a 'Fabry Patient's Meeting' in Rome in early November. Many members were accompanied by family members and feedback from those who went to Rome was appreciation of the opportunity to increase their knowledge of this complex disease and meet others similarly affected from all over Europe, the United States and Canada. Closer to home 25 MPS families in the Yorkshire area enjoyed a seasonal and social get together at the end of November. The intrepid Father Christmas made it from the North Pole to Otley! You will also see included in the newsletter feedback from the lucky sixteen families whose names were drawn out of the sack and went to the National MPS Society Disney Conference in Florida.

Finally, on behalf of the MPS staff team and Trustees, we wish you a peaceful New Year. We all appreciate that for many members Christmas and the festive season is one of mixed emotions and our thoughts were with you.

Christine Lavery
Chief Executive

News from the Management Committee

The Society's Board of Trustees meet regularly. Here are the key issues discussed at their meeting in September 2004 at the Hilton Hotel Bromsgrove.

Advocacy Support

The Chief Executive informed Trustees of the high quality of work being carried out by the current members of the advocacy support team which has enabled a proactive approach to long-standing members for whom there are no active advocacy needs and a programme of support to these members was agreed.

The Assistant Director gave details of the work undertaken supporting individual patients achieve ERT which included appeals and judicial reviews. Trustees were informed of the Department of Health's announcement of NSCAG designation for Lysosomal Storage Diseases and the Society's determination to achieve funding for ERT under the Department of Health's NSCAG designation. The Assistant Director informed Trustees of the progress made in evaluating the MPS regional clinic programme and the Chief Executive tabled plans for the future management of the Childhood Wood.

Research Grant Update

The Trustees received reports on the progress of the MPS research projects at the Institute of Child Health, Addenbrookes Hospital, Cambridge and the University of Manchester. It was agreed that further visits be planned to all three centres to develop closer links and increased understanding of the research being carried out.

Advocacy Events

The Trustees considered an evaluation of the AGM Family Weekend held at Splash Landings Hotel in May 2004. 95% of respondents fed back that they thought the hotel was 'fantastic'. 79% thought the Gala Dinner was a highlight. It was agreed to hold a similar event for the Society's AGM in May 2006 at the Splash Landings Hotel, Alton Towers. Trustees also received feedback from the Sibling Weekend held in July 2004 and agreed the programme for the Society's National Conference on MPS and Related Diseases, 1-3 July 2005.

Policies

The Trustees reviewed and agreed the following policies: Equal Opportunities Policy, Conduct Policy for Chief Executive, Conduct Policy for Staff, Conduct Policy for Trustees, Mobile Phone Policy, Policy for Managing Abusive, Threatening and Insinuating Phonecalls, UK and Overseas Travel and Subsistence Policies, Relocation Policy, Volunteer Carers Conduct Policy.

Publications

The Trustees were informed that several publications are in progress including booklets on Anaesthesia and Inheritance for MPS and Related Diseases. The advocacy support team are working on a series of information sheets tailored to specific areas of the advocacy support service.

Sarah Cornwell says Goodbye...

Just a note to say goodbye to everyone. I have been offered a place at the London College of Law starting in September 2005. Having graduated with a law degree, this will be the next step along the road to qualification as a solicitor. Having accepted this place, my boyfriend and I decided that now would be a perfect opportunity to travel for 5 months, as once we both embark on legal careers it will be very difficult to find the time to indulge in such an experience. We leave on the 5th February for Thailand, Vietnam, Laos, Singapore and then to New Zealand, Australia and across to South America. This is all very exciting and my life will be very different then, to what it is now.

However I have learnt so much in the last six months, all of which I will be able to take forward with me into my future career. It has been a pleasure to work with the people I have had contact with, and I will not forget all the people I have met. I am looking forward to this future challenge and, you never know, I may end up supporting you from a different angle next time we meet. I wish you all the very best for the future.

From Clare…

We will be saddened to say goodbye to Sarah, such is the excellent work she has undertaken within the advocacy support team for the brief time she was with us. We employed Sarah knowing that her career aspirations were in the field of law and that she may not be with us for long, but felt that she could make an invaluable contribution to our work with our members, which she has. We wish her all the best for the future as she embarks on this, her chosen career path.

Who are the MPS Society Trustees?

What are they for and what do they do?

Barry Wilson, Chairman of Trustees, explains the role and responsibilities of the Society's Trustees. For more information on becoming a Trustee contact us at mps@mpssociety.co.uk.

Barry Wilson, Chairman of Trustees

Trustees are, for the most part, grey anonymous figures who merge into the background of a well-run charity for most of the time. Their main role is ultimately to be responsible for everything that the MPS Society as a charity does. The law is quite rightly tough with regard to the management of charities and is enforced, if necessary in considerable detail, by the Charity Commission.

First the Society has to find the money to make sure that it is running a business, (and make no mistake, we have to run it like that), which can pay its bills. To run the individual advocacy support service and fund research on an on-going basis takes a lot of cash.

The Society's trustees then have to make sure that there is somebody in charge on a day to day basis who has sufficient ability, experience and commitment to ensure the Society works properly. Doing this is the most crucial job that the trustees have to do, because a good Chief Executive makes the whole enterprise succeed.

The Chief Executive needs to know about the very rigorous legal framework in which we have to operate, with not only the Health and Safety laws and the Personnel legislation that all employers have to cope with, but some very demanding requirements relating to Child Protection, Data Protection and Care Standards. Our staff have to comply, our Chief Executive has to set up the framework so that they do, and it is ultimately the responsibility of the Trustees to make sure that they do.

So, who are we to carry this awesome and multi-faceted responsibility? Well, the Society is constituted in such a way that trustees must be members of the MPS Society, over 18 years of age and eligible to serve as Honorary Officers or members of the Management Committee. The Management Committee (Board of Trustees) need a broad mix of expertise to be represented at our meetings.

I am a retired businessman, bringing (I hope!) current knowledge of a range of management issues. I am supported by a geneticist, Judy Holroyd, a Health Service Manager, Ann Green, Sue Peach, a teacher, Wilma Robins, a civil servant and Bob Devine, Paul Sagoo and Judith Evans, who bring with them a broad and diverse range of knowledge and experience and all of whom are parents of a child who has or has had an MPS or Related Disease.

Getting three monthly meeting dates to suit us all can therefore be tricky, as Trusteeship is an unpaid add-on to what is already a busy life for all of us. I suspect we manage because Christine Lavery is just as capable of managing Trustees as she is managing staff!

I've Passed! Sophie is awarded her DipSW

After what seemed an eternity, I finally received my final result on the 22 December 2004 to confirm whether or not I had successfully completed all aspects of my course. As you will guess from the title I did and now have a Diploma in Social Work.

Once again I would like to thank everyone who has supported me over the two years. All that is left for me to do now, is to attend my graduation and accept my certificate, as well as implementing into practice everything I have learnt.

The Society sends many congratulations to Sophie on becoming a qualified Social Worker as she remains a valued member of the MPS staff team.

New Members

Hazel and Ewen's son, Luke, has recently been diagnosed with ML II. Luke is 5 months old. His family live in Scotland.

Nicole has been diagnosed with Fabry Disease just over a year ago. She is 44 years old and lives with her family in South West England.

Michael Anthony Burke has recently been in contact with the Society. Michael has Fabry Disease and lives in the North West of England.

Rebekah has recently been in contact with the MPS Society. Rebekah suffers with Fabry Disease. She is 30 years old and lives in South East England with her husband Justin.

Joy Duthie has been diagnosed with Fabry Disease. Joy is 61 years old. She lives with her husband, John, in the Home Counties.

Jane Ransome has recently contacted the MPS Society. Jane is 46 years old and has been diagnosed with Fabry Disease. She lives in Eastern England.

Michele Moore has recently been diagnosed with Fabry Disease. Michele is 31 years old and lives in South East England.

Lister and Jackie's son, Ben Cooper, has recently been diagnosed with Sanfilippo Disease. Ben is 10 years old. His family live in the South West of England.

Sarah has recently been diagnosed with Fabry Disease. Sarah is 25 years old. She lives in the South East of England.

Katie Smithers has recently been diagnosed with MPS I Hurler Scheie Disease. Katie is 20 years old and lives in Wales.

Saleem and Shehla Ali have recently been in contact with the Society. Their son, Sultan Ali, is 4 years old and has Morquio Disease. The family live in Birmingham, West Midlands.

Arran and Charlii's daughter, Willow, has recently been diagnosed with MPS IH. Willow is almost 2 years old and has an older brother, Ethan, and sister, Cleo. The family live in South East England. Willow has now had a bone marrow transplant and is doing well.

Agnita Oyawale's son, Michael, has recently been diagnosed with Morquio Disease. Michael is nine years old. The family live in South East England.

Steven and Naomi Shannon's daughter, Hannah, has recently been diagnosed with Sanfilippo Disease. Hannah is three years old and has an older sister, Kathleen, who is five years old. The family live in Northern Ireland.

Births

Matthew Gallagher was born on 22 September 2004 weighing 7lb, 1oz. Matthew is a baby brother to Ryan who has ML III, both of whom are shown in the photo to the right.

Suhil and Ruhil Chowdhury (MPS III) have a new baby sister. Her name is Pritti and she was born on 25 October 2004 weighing 7lb, 8oz.

Deaths

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

Ashleigh Tomes who died on 16 October 2004 at Little Bridge House, aged 7.

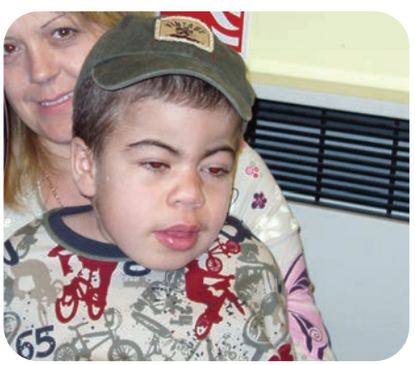
Alisha and Azaria Haji-Afzal who died on 18 November 2004.

David Andrews who died on 6 December 2004 at Rainbow House Children's Hospice.

Max Nyack, who died on 17 December 2004.

Congratulations to **David Oulton**, MPS II, who passed his driving test on 23 November 2004 at the age of 17 years and 4 days. Well done David!

Bristol and Cardiff MPS Clinics



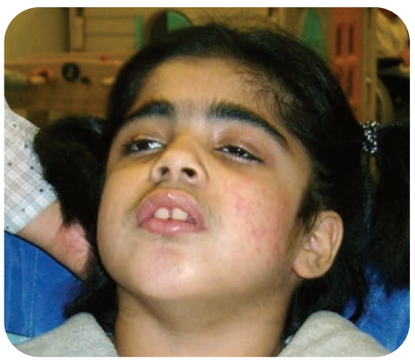
We are pleased to report that both clinics ran smoothly, if not a little chaotic. However, everyone we spoke to has found them useful. There were few hiccups and delays although an apology should made to all those who attended the Cardiff Clinic. We weren't intentionally rationing the biscuits, it was merely the case that Dr. Shortland took quite a fancy to them all, leaving the MPS staff to fight it out over the few that came with lunch! A special thank you must be made to Sarah McKnight for trying, in vain, to bring the biscuits back from the consulting room for us!

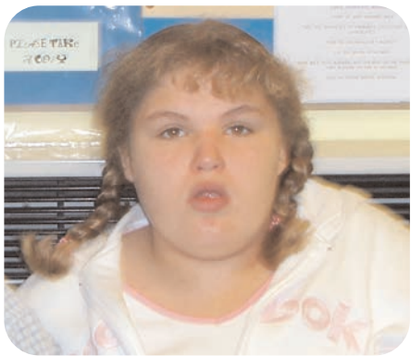
Another interesting moment was trying to find our hotel in Cardiff the night before. Ellie started navigating, as we managed to get lost in the centre! A quick phone call was made to the hotel, who proceeded to direct us out towards them, their main instruction being to turn off after the building with a huge 'Daffodil' on it. But... We were expecting a big monstrosity of a flower on top of a building, not the lovely painted one on the side of the hospital! Oh well, this ended the three and a half hour journey with a few giggles.

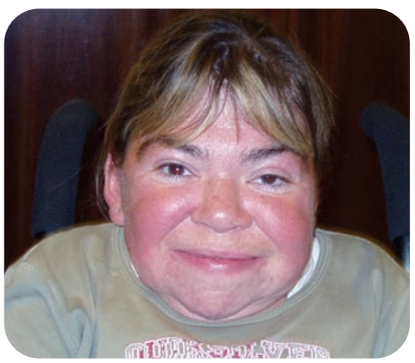
We thank all the doctors who gave up their time so generously for these clinics and also to those who put in a lot of effort to arrange them. By name they are Dr. Ed Wraith, Dr. Phillip Jardine, Dr. Graham Shortland and Sue and Deirdre. Thank you for all your continued support.

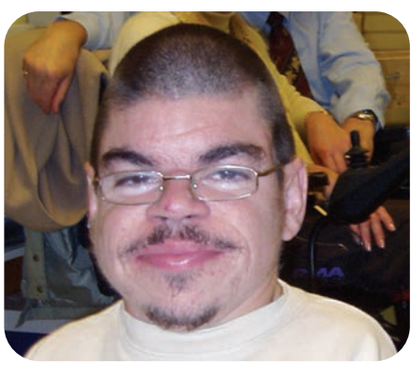
Photos left column from top: Andrew Hawkins (MPS III), Faye Longley (MPS IV), Sophie Clarke (MPS III) Archie and Isaac Eaton (MPS IV) at Bristol, Steven Young (MPS II) at Cardiff.

Photos right column from top: Ayesha Ghaffar (MPS III), Carly Dickinson (MPS III), Helen Skidmore (MPS I), Sara and Gavin Hyde (ML III) at Cardiff





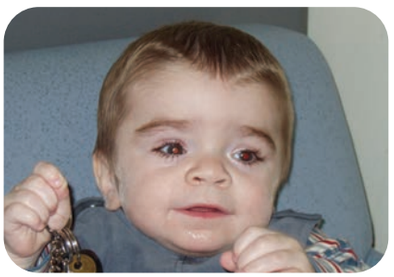




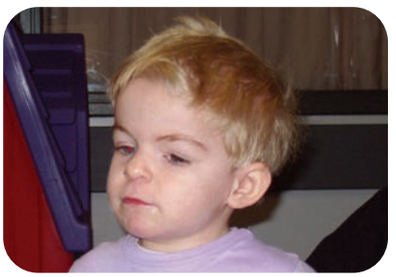
Northern Ireland MPS Clinic

Another wet and cold day, another Northern Ireland Clinic, this time held in Antrim Area Hospital near Belfast on 18th November 2004. As always, there was a full clinic list with Dr Ed Wraith and Dr Fiona Stewart in attendance. The facilities at this location were felt to be very good, please could families who attended let us know if they disagree or have any comments or suggestions to be made. We hope that next year's clinics can be held here.

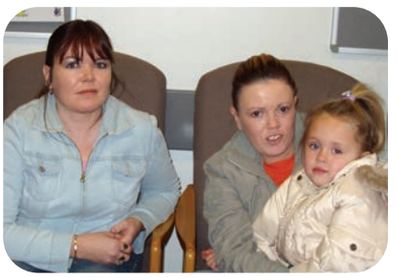
Thanks go to Dr Stewart, Dr Wraith and Dr Stewart's secretary, Sandra Smith, for ensuring the smooth running of the clinic and also to the staff in the maternity & antenatal outpatients department who accommodated us.



Charlie (MPS I)



Hannah Shannon (MPS III)



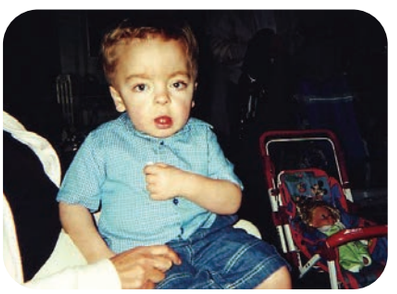
Fiona Devlin & Bernadette Maguire (MPS IV)

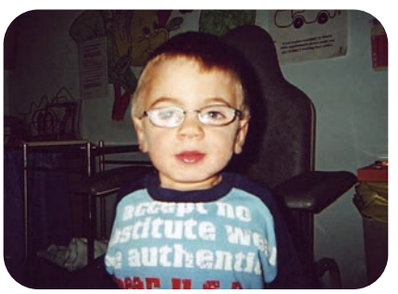
BMT Clinic By Cheryl Pitt

As you will now be aware the BMT clinic at the Willink has been split in two and spread over two days. The clinic has become so busy, it was felt that you would benefit from more time with the Consultants with this new arrangement. The under-5s come on one day, and the over-5s on another. This also allows for specialist Consultants to see children in the age group that is relevant to their field. So, I hope you felt you had more time to spend with the Consultants, and that you find this new arrangement agreeable.

One thing's for sure, the waiting room certainly seemed a lot quieter! As usual it was lovely to see you all again, and great getting to know the more recent additions to the clinic list. Please remember that, in future, if you require support with any issues related to your child with MPS I, please call a member of the Advocacy Support Team prior to the next clinic, so that I can bring any relevant information with me.

Thanks to Jean Mercer and Bernie for organising the clinic and arranging refreshments. Thanks also to Gill Moss and the Consultants: Dr Wraith, Professor Clayton, Mr Meadows, and Dr Wynn for their excellent work and a successful clinic. I would also like to wish Bernie the best of luck with her new job - she will be greatly missed!





BMT Clinic photos clockwise from top right: Jordan Mount, Leighton Barker, Rubina Jalani, Matthew Ingram, Keira O'Neill, Thomas Mett

Childhood Wood Planting 2004

On Friday 22 October, the MPS Society joined with five member families to remember and commemorate five special children who lost their lives this year to MPS diseases, at the Childhood Wood, in Nottingham.

We gathered at the Clumber Park Hotel for a buffet lunch where Paddy Tipping MP, a valued supporter of the MPS Society, welcomed everyone and spoke a little about the Childhood Wood and what it represents. Six-year-old George Wragg then thanked Paddy for attending, and presented him with a bouquet of flowers.

Everyone then made their way to the Childhood Wood in the heart of Sherwood Pines, where Sir Andrew Buchanan gave a touching speech at the Wood and read out the names of the children being remembered. Sir Andrew was also presented with a bouquet of flowers by Ellen and Louise Mayhew, as a thank you.

Photographs clockwise from top right: Jane, Richard, Louise, Ellen, Lloyd & Neave Mayhew & Teresa & Barry Mulcahy; Linda Smith, Christine Lavery & Sharon Birch; Ken & Margaret Naish; Sir Andrew Buchanan, Louise & Ellen Mayhew; Welcoming families to the Wood.

Each family went on to plant their own oak sapling in the Childhood Wood, with help from Byron from the Sherwood Pines Forestry Commission.

It was a peaceful and beautiful day, and the rain held off! We would like to thank all those families who came and planted a tree, and also a big thank you to Byron from the Forestry Commission, and Sir Andrew Buchanan and Paddy Tipping MP, who always support the Society.

Leeds Family Day By Sarah Cornwell

6am and I have just woken up knowing I have half an hour to make myself presentable for the day to come. The taxi is waiting outside the house at 6:30am and the journey begins there. I get to Heathrow airport for 7:15am and wait for Sophie. 7:30am and I spot her walking through the entrance. We are both in need of some serious caffeine, Sophie having been out for a 40th birthday the night before and myself, Sarah for a 21st!

We fly to Leeds Bradford where we are picked up by Ellie, and taken to the Craiglands Hotel. There we have another few cups of coffee and decorate the tables with Christmas crackers, drinks, hats and so on. 12:15pm and families start to turn up.

Having never met any of them before it takes a while to suss out who is who, but then get chatting away to the Heath family with Sophie, who has proceeded to take quite a shine to Jack's baby sister, Katie. The room has started to fill up and the noise level was slowly increasing, children are running around getting excited having seen the Christmas tree to one side of the room.

Lunch is served, and for a short time everyone is more engrossed with what is on their plate than what is going on around them. Out came the magic man and the balloon man. The children spot them and both men immediately become the centre of attention for the next couple of hours. This is fine with us as it gives us time to get our resident Santa dressed up to the nines with bags of presents. (I won't reveal who it was, as I don't want to be held responsible for spoiling a child's innocent perception of Santa so close to Christmas!)

Santa appeared and half of the children were mesmerised and the other half were trying to guess who it was that was hidden under the outfit. The presents were handed out and many photos were taken. It was a lovely end to what had been a very hectic day. People started to leave and things started to die down. All in all it was a very successful day and we hope to repeat it next year.

Merry Christmas Everyone!

ERT Christmas Party

By Sophie Denham

Addenbrookes Hospital, Cambridge

On 9 December 2004, Christine and I were invited to join everyone involved in the MPS II ERT trial for their Christmas party. Everyone was in good spirits and the staff made it extra special by dressing up in clothes relating to Christmas. Everyone had a good time and there was plenty of food and drink for everyone.

Before the entertainment arrived there was a special visitor to the party. Yes, you've guessed it, Father Christmas, bringing presents for all the children. Father Christmas even stayed for the entertainment, which for me and probably for everyone there was the main highlight of the party. The entertainment was in the form of the clown doctor who was fantastic and not only entertained the children but the adults too with her quick wit and superb balloon modelling skills.

We had a great time at the party and I would just like to thank everyone at Addenbrookes Hospital for inviting us. Here are a selection of photos from the day.

Oliver - A Celebration

A Poem By Shauna Gosling, Ollie's Grandmother

The anguish when thinking of that child
So innocent and pure
That baby that should grow and have a life ahead
The pain when remembering that enchanting smile
That gurgle of pleasure as the hand reaches out
To touch your face, your eyes, your hair
Reaches out for vivid colours and jingling toys.
Dear baby you are such a precious being
How could this terrible thing happen to you?
Why should you not have the chance to grow?
Why should you be so different to others?
Why should your life be so short and sad?
I shed my tears for you right now.
But little boy, you will be so loved
I will always be there for you.

The sky is blue, the white clouds are turning to a vivid pink
The countryside is beautiful, the light reflecting off the trees
With all the different colours and hues, the land is green
With appearing shoots that will grow oats and barley.
All around is still and clear.
The birds are having their last song before the night descends.
As I look around I marvel at nature's gifts
Then I stop, I look inside me and it is if my spirit has died
I feel quite dead and dark and want to go and hide away
There is anger too, how can nature be so beautiful on one hand
And so very cruel on the other, where is God?
How can that baby be born to have such a dreadful life
A life of pain and sorrow, no, it is not fair
It is not fair for his parents who will see him suffer
It is a sad, sad time of questions and queries
There seems to be no answer.....January 2003

Laughter and smiles, what an enchanting baby this is
He is attached to so many tubes and machines
Lethal chemicals are dripped into his body.
Nurses come and go attaching tubes,
attending to his bleeping machines.
He complains when it hurts
They tickle him and talk and he gives back a huge smile.
The people he loves tend to him,
play with him and stay with him always.
Then comes the big day, the day that can change his life.
From a black future to a promising worthwhile one.

Dear Ollie, You are given the chance of a better life.
That precious blood that flows in you comes from a baby just born
Whose cells enter your body, will multiply and
enter your bone marrow and become one with you
Replacing the enzyme that is missing.
Just as well, your body is being abused by so many chemicals.
So many different medications seem cruel right now.
But we keep looking at the bigger picture, looking to the end result
Turning despair to hope.
A wonderful happy boy full of life.
You are such a wonderful child
We all love you so much
Bless you Ollie.....April 2003

As yet again I fly across the Atlantic
My heart is full of hope, this journey is different
It is for a celebration, not an anxiety
That wonderful baby will be one year old
With all that heartache and the spirit of those around
That little boy has struggled and shown such courage
He is not through it all yet, but the worst has passed
He laughs with the excitement of being part of the outside world
No longer is he in just one room attached to al those machines
Yes, despair has turned into hope
Now it is even more than that.
I go to see a child that can play and smile.

Here comes the party, lets play the drums
This is a miracle to behold
It is Ollie's first birthday
So lets go out and celebrate
Put up the balloons, decorate the cake,
Wrap up the presents and all is well,
Here comes the birthday boy!
All dressed up in his best
A huge smile is upon his face
Gurgles of laughter and pleasure
As he grabs the paper and sees his toys.
Surrounded by cards, love and happiness.

Then on to the party
Where he sits at the head of the table,
A blue crown upon his head
For today he is the prince
And he certainly deserves that status
All he has gone through, the trials and the anguish
The pain and seclusion from the outside world
His disposition is one of joy and smiles and laughter
Dear Ollie, you are the prince today
We all celebrate this special occasion
With thanks and gratitude to have you here
You are an example to us all
Happiness Ollie, have a wonderful life
Our hearts burst with joy.....June 2003

Ollie Gosling, MPS I

Daniel wins the Community and Citizenship Award

Hi, I'm Madeleine, Harrison King's, mum. Harrison has Sanfilippo. I thought I would write in and share with you the outstanding achievements that my eldest son Daniel has achieved over these last two years. Up until Harrison was diagnosed in February 2002 Daniel was aiming to leave school to join the police force in London, but he decided that he did not want to leave Harrison and move away now that he was ill.

Daniel joined a voluntary charity organisation called CHAOS through his school; this is a group that help families who have children with disabilities. The last two years he has helped a boy with autism, another with severe cerebral palsy and a boy with severe learning difficulties. He has worked for a year, again voluntarily, in a school for autistic children for 1½ days a week, which he thoroughly enjoyed. While in the sixth form at school Daniel worked two days a week at a local Montessori playschool as a one-to-one for a young boy with special needs.

Dan at Work

In July 2003 Daniel was presented with the Rotary Club's Youth Endeavour Award for volunteer work, involvement in school and commitment to helping others in difficult circumstances.

In March 2004 he received the East Dorset District Council Community Service Award in recognition of his voluntary work and commitment and effort in the community. This was presented by Emlyn Hughes, ex England Captain and Sports

Dan with Emlyn Hughes and the local Mayoress

Personality. Emlyn was a really nice man and talked about his own difficult times dealing with his brain tumour, which sadly he died from recently.

Another certificate Daniel has gained is for attending counselling skills sessions from the NHS Trust. These include listening skills, child protection overview, bullying, raising self-esteem, assertiveness and stress.

Earlier this year we gathered quite a large group of people and went to Daniel's Graduation Evening, which was held at the Bournemouth International Centre. We were quite surprised that in front of hundreds of people, Daniel had to go on stage after receiving his graduation certificates to receive his CHAOS award, 10 girls and he was the only boy! Later in the evening the head teacher spoke about a boy who was going to be presented with the Community and Citizenship Award. As you can imagine the room was full of applause, cheering and tears as Daniel returned to the stage to receive his award.

Daniel has since left school and remains at the Montessori nursery as an SEN Support Worker now having his own group of children which include special needs at the same time studying NVQ3, moving towards a degree. I think you will agree that Daniel has done us proud. He has applied to be a Volunteer Carer for July's 2005 MPS Conference, which he is really looking forward to.

Here is an edited version of the speech for Daniel's award...

The Community and Citizenship award is given in recognition of an outstanding contribution to the community and to others, whether in school or beyond. This year's winner is characterised by his modesty. Many of his fellow students probably have no idea of the time he gives to others.

When he devotes his time to something, he has always had the determination and commitment to see it through. He lives by a very strong moral code and will stand up for what is right. Ladies and Gentlemen, staff and students, this year's community and citizenship award goes to Daniel Luckham.

Emma's Transition to High School: Part 2

John and Emily Slater

This article is the second of a two part series, the first of which appeared in the Autumn 2004 MPS Newsletter. If you need support with any issues around education contact the Advocacy Support Team for more information.

In preparation

On 2 September 2004, the day before school opened, Cheryl and Sophie gave a presentation to all the staff (not just the teachers) at Tottington High School, so now everybody at the school had some knowledge about Emma and her condition. We felt this was a good start, letting the staff know about her problems from day one.

First day

Emma was, of course, very nervous but she was looking forward to starting her new school, and wearing her new uniform with a tie and blazer. Due to the short time frame between our visit and the start of the new school term, there isn't a chair in each class yet so Emma has a chair carried from class to class for her. Emma has made a friend named Rebecca who now helps her around school when needed.

And now?

Emma seems to be coping well although she does find it more tiring than primary school as she has to move from class to class. As for the teachers promises at the meeting? They have done all they said they would do. Emma now has a chair in every department including science lab-style stools with a back to support her (rather than just the stool type). There is also a posture pack in every class.

Emma leaves class two minutes before the bell so she can get to the next class or dining room before the rush in the corridors. Emma also has an early pass so she can go into school early in the mornings, and she is happy with all of this as she is not madeto feel different from the other children. She has had a few questions from the other children but they have accepted her and just got on with school life.

Now we are well into the first term we have a better picture of how Emma is coping with high school, and she is coping well with the help of the RP unit and staff (she doesn't like all the home work!). We have now had our first parents evening and her form teacher said Emma has fitted in well and just gets on with it. All seems to be going well. We are now only waiting for her statement to be amended to include physiotherapy and psychological support if needed.

We are happy Emma has coped so well in high school, especially with all the changes she has had to adapt to. She is happy and enjoys going so we just hope this continues so Emma can enjoy her time at high school.

Life at Treloar College By Amy Pain

Hi! I am 18 years old. I have started at a college which specialises in physical disabilities and there are also some students there with learning disabilities.

I started Treloar College in September and have made loads of new friends. I get on really well with a girl called Sam who has had the same operation which I am going to have soon. This is a spinal fusion and should take place between Christmas and Easter. It will be done at the Royal National Orthopaedic Hospital in Stanmore.

Treloar is the best college around this area for me to attend. I like it a lot and feel very secure here. The course that I am doing is Health and Social Care. The first three units I am doing for this year are Investigating Health and Care Services, First Aid and Safety and Personal Development. The next three units will start in the next academic year.

I go to college every day from 9am – 4pm. I don't go home until the end of the day, but I do get Friday afternoons free to study. I also have regular physiotherapy sessions as well as weekly hydrotherapy.

Sibling Stories

William, aged 7¾, brother of Sophie who is 6½ (Sophie has Sanfilippo Disease)

William and Sophie

Sophie always stands in front of the television. She sings Old MacDonald, ring-a-ring of roses, wheels on the bus, 1,2,3,4,5 and her favourite is row, row, row your boat. She screams very loudly and throws things about. She also chews Leo's nose (my toy lion) and her toy bone! She chews puzzles and books as well. She smacks Daddy and Mummy on the back. She always chews her cups so she can't get any juice out of them. She poos loads and runs around. She has a special chair to eat in to stop her from escaping. We have to put special powder in her juice and fruit because it helps her.

I like to push Sophie in her wheelchair and Mummy or Dad will stop me rushing down the hill with her. She put her foot down the toilet and got her foot and sock and trousers wet! Sophie threw her duck bread in the lake when the bag was tied with a knot. Poor ducks! When we were littler, when we were in the bath, Sophie turned on the hot tap when I slipped on the bath floor!

I feel sad because she isn't in my school and because she has to go to hospitals called Keech and Great Ormond Street.

Living with Sanfilippo

By Hannah Donegani (16), sister of Amy (15), Daniel (12) and Josh (13)

I arrived back from a party with my brother Josh to find Mum in a right state. She had just managed to stop Amy from eating the whole bar of soap while Daniel was busy with his favourite game - playing with the plugs and switches in the lounge. Empty food packets were all over the kitchen and biscuit crumbs were scattered on the carpet. The bathroom floor was flooded, Amy again! This was a typical day seven years ago.

When I was nearly 9 and Josh was 6, our extension was built. Amy was 7 and Daniel was 4.

Hannah

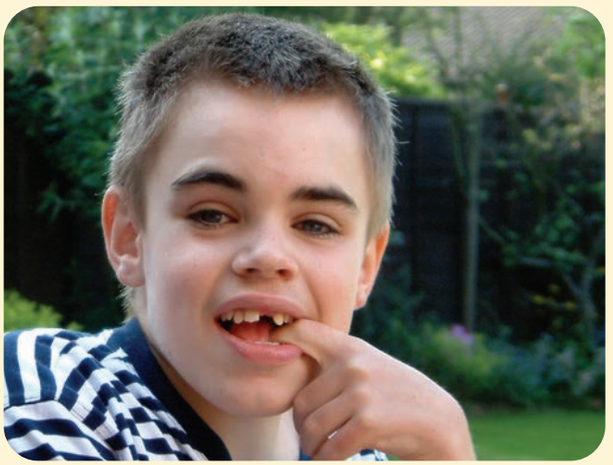
It was brilliant. I now didn't have to worry about them 'helping' me with my latest artwork when I wasn't there and Mum could get dinner ready without being scared of Daniel raiding the fridge, or Amy burning herself on the hot pans. Amy and Daniel now had their own safe play area, next to the kitchen.

Amy and Daniel have Sanfilippo Disease, a neurodegenerative disorder that's caused by a mutant recessive gene. Carriers like my parents aren't affected. They have a functioning gene, and a non-functioning gene. My brother Josh and I are obviously either carriers or have two working genes. Sanfilippo affects me only in that I live with a brother and sister who have two non-functioning genes. As a result, they cannot produce an enzyme to break down certain sugar molecules, called mucopolysaccharides (or MPS), which build up, inside the cells (mainly in the brain), damaging them.

Amy started the primary school I was at, but was moved to a special school for children with severe learning difficulties after 2 years. It was sad for me, as she had been my sister at school. She had been embarrassing at times though, for example getting told off in assembly for singing '10 green

bottles' over and over again. Sometimes I'd been bullied and teased because of her but I'd tried to stick up for her. It hurt because she was my sister and I loved her.

Now life is a lot quieter, Amy and Daniel are slowing down. The average life expectancy of a child with Sanfilippo is 15 years. Amy had her fifteenth birthday in June but is still quite well. The extension has an ensuite bathroom, which is very useful, as they find it hard to move around. We have just had a hoist and track fitted to the ceiling so that when she needs it Amy can be lifted from a chair to her bed and the bath. Daniel only says a few words now, and Amy doesn't talk at all. She finds it hard to swallow and if some food goes down the wrong way, she could get a chest infection. If it gets too hard for her to swallow she will need to have a gastrostomy, an operation to put a feeding tube straight into her stomach.



It's been hard work looking after Amy and Daniel, especially when we didn't have the extension and they could 'destroy' the house. Now they are older, they are totally dependent. They don't sleep very much, so Mum and Dad haven't had many good night sleeps for 15 years. But it has made our family very close, as we all have to help each other. Although they are hard work, looking after Amy and Daniel is very rewarding. They are both very loving and are always smiling even when they are awake in the middle of the night.

When Amy and Daniel were diagnosed Mum read in a book that siblings often complain of their parents not giving them attention, and that this can lead to difficult behaviour and emotional problems. Mum and Dad tried very hard to give me and Josh lots of attention, this was good, and we never felt left out.

In some ways Josh and I have had special privileges because of Amy and Daniel. Mum and Dad taught me at home for 7 years and Josh is still

home educated. 3 years ago, Gran and Granddad moved up to Loughborough, from London, to be near us. It's great to be able to see them often and to be so close to them. Every day after school, they come to help give Amy and Daniel their tea. Two or three times a year Amy and Daniel can go and stay at Rainbows children's hospice, where they are cared for and have a great time, while the rest of us can go on holiday and chill.

We've also had special days out with Rainbows and the MPS Society, who have taken us to 'Alton Towers', the 'Imax Theatre' and the 'Horse of the Year' show at Olympia, just to name a few. We went to a super party at 11 Downing Street with other families with children with MPS a few years ago, and one year we even got to go on the Loughborough Town Hall balcony so that Amy and Daniel could turn on the Christmas lights for the town.

Living with Sanfilippo is a normal life for me. Life has been interesting, frustrating, exciting and sometimes sad. I don't like to think too much about the future. Life will be very empty without Amy's smiles and Daniel's cheeky grins.

But for now I'm just content to be 'living with Sanfilippo'.

Photos this page clockwise from top right: Amy (MPS III), the Donegani Family, Daniel (MPS III)

What it's like having MPS by Oliver Moody

Oliver (MPS VI)

My name is Oliver Moody. I am nine years old and I have MPS VI, Maroteaux-Lamy Disease. My Mum and Dad found out that I had MPS when I was five years old.

One day my teacher came out of school to tell my Mum and Dad that I could not put my arms up properly in PE. So, we went to have some tests at the hospital and they told us that I had MPS.

I think I am fine but technically I am not.

My dad then found out where Dr Wraith was, and we went to see him in Manchester. I like Dr Wraith but he is a Newcastle United fan and I am a Leeds fan! Dr Wraith did some tests on me and he told us what type of MPS I had. But that was five years ago. This is how I am now:

I cannot put my arms up as far as everyone else. do not grow as quick as everyone else. That's why I am the smallest in my class and it affects my joints. A lot of my joints crack and I have had an operation on my fingers to make them work better.

I spend a lot of time in hospitals having tests. Sometimes it gets boring. All this does not stop me from playing football for my local team. This year I won a trophy for the club 'Man of the Year'. I also have lots of friends who I play with. People say I am a happy person and that I smile a lot.

My family have been wanting me on the enzyme replacement therapy because it will make me a lot better. A few weeks ago we got a letter from our local PCT saying that they will pay for my treatment when it becomes available. It should be some time next year. I feel glad because up until now it was incurable. I am very lucky and I hope all the other MPS children can get it.

Oliver is featured on our front cover. His father has written the following article from a parent's perspective.

From diagnosis by Paul Moody

It is exactly five years ago since we walked across the hospital car park after being summoned by the specialist. We were not too worried – we had asked for tests to be done on Oliver when we realised that he couldn't lift his arms very high. He had already been seen by one consultant who suggested he may just have a slight deformity in his shoulders. This new specialist had had many x-rays done and revealed that the radiologist had recognised the problem. Today he would tell us what the problem was.

Dawn and Paul Moody, Oliver's parents

Our only fear was that he might need some form of corrective surgery – how wrong we were!

On entering the hospital, the specialist broke away from his students and took us into a small side room. He seemed very sombre. He began writing an extremely long word on the white board on the wall, but abbreviated it to MPS and within the space of a few minutes, our whole world crashed beneath us.

We heard words like incurable, degenerative, fatal, but we were too dazed to take it all in. He told us we would need to bring Oliver down to give a blood test, to be sent away for analysis. He asked if we wanted a cup of tea. We refused and within fifteen minutes we were walking back across the car park as different people. Unable to understand or comprehend what we had been told.

When Oliver arrived home from school that afternoon, he was his usual cheerful, happy, energetic self, and we found it even harder to understand.

I look back now with anger on that day because the hospital sent us away with no help. The specialist was too blunt with us, and never told us about the MPS Society or about the Willink in Manchester.

I began trying to find information. I found the Royal Manchester Children's Hospital had a specialist called Dr Wraith and I got on the phone to try to locate him. We were still consumed with grief, and it took almost an hour to locate where Dr Wraith was based.

Eventually, I remember explaining that I needed to speak to Dr Wraith even though he would not know who I was. The next voice I heard was a friendly Geordie person saying 'Hi, Mr Moody, how can I help?' From that moment on things began to get better.

I explained what had happened and said I did not know what to do. Ed Wraith told us to contact our doctor immediately and ask him to fax through a referral and to drive over, the following day, for a consultation.

We took Oliver to Manchester the very next day and met Ed. He explained the condition in detail to us and even though he needed to carry out a blood test, he predicted that Oliver had MPS VI.

He explained to us that Oliver did not have severe symptoms and that a treatment may be available in time to give to him in the foreseeable future. Suddenly we could see a little light at the end of the tunnel. Something to aim for, a little hope!

Over the coming years we tried, unsuccessfully, to get Oliver onto the clinical trials of enzyme replacement therapy. At one stage we even offered to move to the States and live with relatives in L.A. to get him onto the trial there.

Throughout all this time we have become paranoid about his health. Every headache, every joint pain, any problems he has with his health are taken very seriously and cause us great worry, and yet these same symptoms would be almost dismissed as 'growing pains' in his brother and sister. We have tried not to spoil him or give him 'special treatment' but like many MPS parents, we are guilty of over-compensating by eating out, days away, holidays and generally 'living for today'.

This year, however, the third ERT trial finished and BioMarin announced their intention to file for marketing approval. Our application for funding needed to be submitted in good time to our local PCT because it is hoped the treatment will be available sometime in 2005.

We alerted our local councillor and began campaigning for the treatment. We expected a long hard battle and were already feeling low, because we were hearing about MPS I children being refused the treatment.

In July, however, we received a short letter from the PCT, saying that Oliver's case had been considered before the panel and that they were pleased to say he had been approved for funding!

This was the letter that we had been praying for for five years and was the best possible news we could have received.

We are still frightened to celebrate though. We worry that they might change their mind, something may go wrong, the treatment may not be approved… we still feel like we are walking on glass and will not believe it until we see the enzyme dripping into his body!

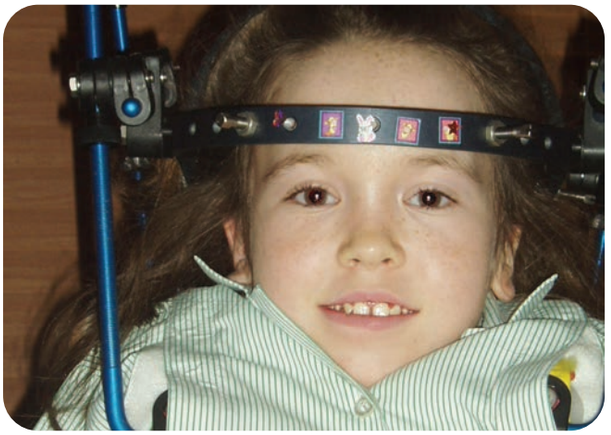
We are hoping though, that one day next year, we will find ourselves walking across a hospital car park with Oliver, in a much happier state of mind than five years ago, and that for the first time in his life, the missing enzyme will have been introduced into Oliver's body.

We also want to echo the final words of Oliver's story that we sincerely hope that all MPS children will soon receive this treatment. It is unbelievable that children are being refused ERT and we will continue to do everything we can to help the cause, raise the profile and make people aware of what is happening.

Oliver and his family have fundraised for the MPS Society. Check out the Society's fundraising newsletter to see how well they did!

Roma's Education

by **Bernie Drayne**



Roma (MPS IV)

In the Autumn 2003 Newsletter you may recall the article which I submitted relating to the problems we were having with regard to our daughter Roma and her transition from mainstream primary school to secondary school here in Northern Ireland.

I am now happy to report that Roma now attends a local grammar school, the school of her choice. The school has a very caring ethos and both Roma's academic ability and her care needs are well balanced and catered for. Problems with physical accessibility and access have been minimised, there are two lifts in the school and some classes have been rescheduled so that Roma can attend lessons which would have otherwise been in rooms which would have been inaccessible. There is an excellent pastoral care system and the special needs coordinator has been anxious to ensure that Roma has made friends.

I wanted to give you this update for several reasons. I would hate to think that other parents in the same position as ourselves would have to go through the dreadful stressful and negative experience which we found ourselves in when trying to find a suitable school for Roma. I also want to highlight the intervention which Christine Lavery and the MPS Society made on our behalf to our local MP, Mr Jeffrey Donaldson who rapped the knuckles of the local Education Board.

In hindsight, I probably should have spoken to the MPS Society earlier about this. I had been advised by many professionals from teachers to educational psychologists and occupational therapists to start looking around schools at least two years before Roma's move to secondary school. I took this advice and spent two years contacting and visiting schools. The majority of schools were inaccessible in terms not only of physical structure but also and, most depressingly, in attitude. The attitudes of some school principals was unbelievable, some suggesting that a special school would be more suitable, whilst others were more concerned that their listed buildings would be denigrated if lifts were added. Whilst some schools dissuaded me from visiting by telling me to contact the education board others said 'oh, there's no one 'like that' in this school'. It became almost comical if it was not so serious when I realised that when visiting three schools in particular, I was taken around the most inaccessible routes possible to prove the point that wheelchairs were just not simply welcome.

I may sound very cynical, but that is what the process did to me. Despite 'professionals' knowing that there is a very real problem for children with physical disabilities to access the education they are entitled to, no-one really seemed to care or do anything about it. Social workers kept their heads down and occupational therapists were suggesting that Roma should travel 40 miles to a school because it had lifts, the alternative being was to attend a local school renowned for its bullying.

There was no proper written information for parents involved in this process from the education boards. There was plenty of guidance issued for parents of children who did not have disabilities, but when I rang the education boards it was very apparent that the transition officers knew very little, if anything about the prospects for children with disabilities.

I really felt very isolated and exasperated by having to make a nuisance of myself as I visited the schools. The final straw came when the educational psychologists would not give me a date for Roma's assessment which would determine her academic abilities and type of post primary school she would attend. Roma's peers were all sitting the 11+ exam in mid November, but Roma's assessment, we were told, would not happen until much later – February at the earliest. This was unacceptable as it treated Roma so completely different from all the other 60 pupils in the primary school sitting the 11+.

Christine Lavery intervened on our behalf and wrote to our local MP who in turn spoke to the Chief Executive of the education board.

Within two weeks the educational psychology service both rang and wrote to us giving us a date for Roma's assessment! At last we were being treated courteously and, most important of all, Roma was being taken seriously and afforded the respect she deserves.

Roma is now at the school which she really wanted to go to. She is still in the settling in process, and has extra problems because she is wearing a halo having had a spinal fusion. That's a story for another day. I still feel bruised after the whole experience but the lesson to be taken from this is that you really have to stand up to be counted. We all want what is best for our children, and sometimes you have to really fight to get your voice heard.

I know that the intervention by the MPS Society definitely helped us, it was a shot across the bows for the education board. Following this intervention, the contrast in our treatment could not have been more marked. We were advised of progress very rapidly and education officials acquired real names!

We are very grateful to the MPS Society, and we know that in future should the need arise we will ask for their assistance again. I would urge any parents in a similar position to do the same thing. It certainly eased our burden and it has hopefully provided a wake up call to the education authorities that they have a very long way to go if they are to comply with the Disability Discrimination Act. ■

MPS Regional Clinics

Dates for 2005

Bristol

Tuesday 5 April
Tuesday 7 June
Tuesday 29 November

Cardiff

Wednesday 13 April
Wednesday 30 November

Birmingham

Wednesday 30 March

Newcastle

Tuesday 8 February

Northern Ireland

Thursday 12 May
Thursday 17 November

Irish Society for MPS Diseases Conference

1-3 April 2005

The Glenroyal Hotel, Maynooth, Co. Kildare, Ireland

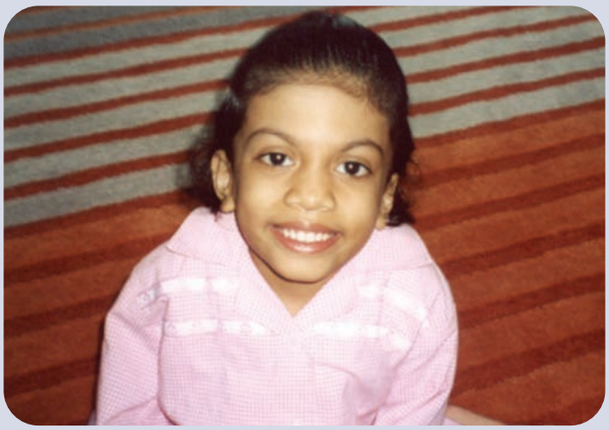
Among the speakers will be:

Dr Ed Wraith, Dr Fiona Stewart, Dr Anne O'Meara, Dr Eileen Treacy, Mr Fogarty, Mr Pat Doherty, Mr Paddy Flemming.

If you would like more information please contact Martin Lynch on 0035361351778 or email mps@mpssociety.ie

Do you have any stories, news or information that you would like to share with other readers? If so, contact the MPS Society on 01494 434156 or email us at mps@mpssociety.co.uk

Aisha's Story by Sharif and Asma Seedat



Aisha (MPS IV) aged 5 and a half

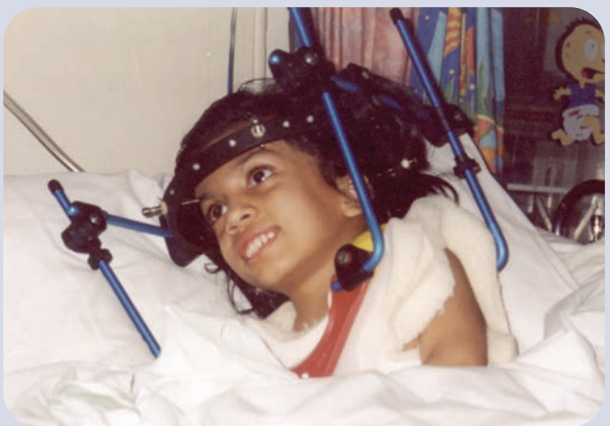
My husband I were looking forward to the birth of our new baby. Aisha was born in November 1997 and her diagnosis came when she was seven months old. At that time we were still residing in Malawi, Africa. These were no doubt the 'happy days' for us because Aisha did not know about the complications of her disease and we always prayed that she would get better as any parent facing a diagnosis does.

I had flown out to the UK with her grandfather and she was diagnosed at Queen's Medical Centre in June 1998. We returned back to Malawi to break the news to her Dad and the rest of the family. I don't know which was harder, to get her diagnosis or to tell people about it. As any parent in our position knows when your child is diagnosed with a condition, it seems you can't go through another day, but you do as we have. It has been seven long years since her diagnosis. We returned to the UK in 2000 for permanent residence. It has been a roller coaster of emotions for us. All parents reading this will understand how we felt, to be told that our beautiful child has this degenerative condition, to which there is no cure.

We remember the day very well when the MPS Society visited my husband and I in my parent's home in Leicester. It was the turning point in our life and one thing that keeps me going even today is the comfort that we received from the Society at that time. We were given the 'yellow booklet' and some other literature. For the first year we kept it hidden in our bedside drawer and when we went through the book we tried not to look at the pictures. Until one day it became easy to pick up the booklet and read it for information, as by then we were actually relating some of the things our daughter was starting to go through. Now we give the booklet to anyone who is interested in her condition.

We have been in contact with the MPS Society and Dr Wraith since 1999. Dr Wraith and his team have always been very supportive and we are grateful to have him there for us. It is reassuring to know that Aisha is receiving the benefits of worldwide research.

In February of this year, Aisha had her neck fusion. This was under the care of Mr Cowie and Dr Wraith at RMCH in Manchester. She spent four weeks in hospital. We stayed with her. She was on the Heywood Ward. The doctors and nurses looked after Aisha excellently and made her stay at the hospital really comfortable. It was quite a time as we now have another daughter Saffiya who is three and needs equal attention. The hospital had a multi-faith prayer room. We found a lot of comfort in that room especially during the time of her op. We practice Islam and our faith and prayers during this time kept us going.



Aisha - 10 days after her neck fusion

She was eventually discharged and we were glad to be back home. For the first three weeks Aisha's grandma from Malawi stayed with us but she had to go back. She helped out with the cooking whilst we saw to Aisha's needs. It helped me a lot and made Aisha happy to have her with us. Once her Grandma had left it was just the four of us and dealing with Aisha in her halo traction on our own. It was quite daunting at first but we slowly got used to it. Social Services in April appointed us to have Homecare to help with Aisha's nursing needs. This was mainly because Saffiya was not receiving the total attention and gave one of us a break from time to time to carry on with other things especially seeing to Saffiya.

Aisha has a Statement of Special Educational Needs. She attends mainstream school. Aisha accessed the services of The Hospital School whilst in her halo traction. They are based at the Leicester Royal Infirmary. She was on the outreach service, which enabled her to be taught at home every day for a few hours five days a week.

She did so well even in the first few months when she had no balance and could not walk. She was awarded with an achievement certificate and the Headteacher's Golden Owl Award at the end of her Summer Term. She was given the opportunity to carry on with her education, which no doubt she would have missed out on if this service had not been available. We will always be thankful to her

school and hospital school for this. On the day of her assembly at Hospital School she was wheeled and proudly received these special awards that no doubt she really deserved. We stood so proudly as everyone clapped for her. It was a day that we will never forget.

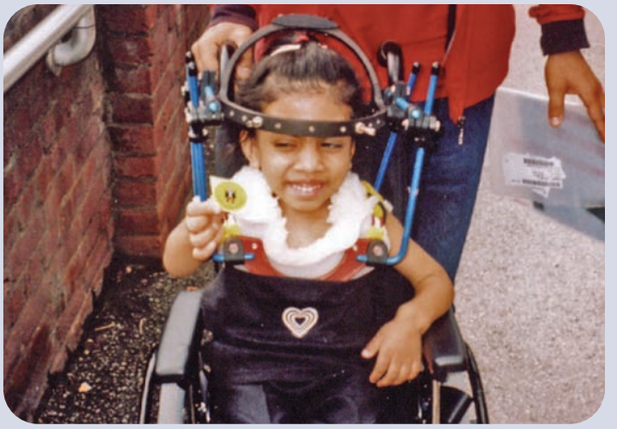
We have so much to say about our experiences since she had been in her halo. It had become part of our lives and wonder how we have gone through the six months. It was not easy but we stayed positive throughout and given her all the encouragement we could. She was not mobile for the first four months and one day she surprised all of us. She took her first steps; it was like when she took her first steps as a baby. Out of everything we think that the nights had been the most difficult as she constantly awakened to be turned or she felt too hot or too cold.

If you see us as a family we would look like we have dealt with it very well but, any parent that has gone through what we have, will understand that you have some days where you think you cannot do it anymore and you wake up the next morning and feel a little better and are ready for the challenges again. Aisha has probably seen us through these days. She has coped so well. Someone once said to us she is strong because she sees her parents strong and that is probably true and the key to many things.

We have passed the first hurdle and know that there are many more still to come. All we want right now is to get some normality back in our lives before the next surgery which we pray will not be for another year or two. As parents we have battled on with the Social Services. We have been told, as Aisha attends mainstream and does not having learning difficulties, the resources available to us are very limited. We have no respite for Aisha and we have had endless meetings regarding DFG for our property that we want to get adapted for Aisha. There are times when we just want to give up but our faith keeps us going.

Aisha is back at school now. She is gradually building up her hours at school. She uses a manual wheelchair to get her around, as her mobility is restricted. We have problems buying clothes for her because of her size. This is another difficult period which we are facing at the moment as her sibling who is almost four is much taller than her. Aisha always felt that by having the neck fusion she would grow in height. This is something that we always talked to her about but she always talks of getting taller. She admires all tall things. It pains us to hear this even though we knew from the start that her stature would be affected.

We hope that she can one day understand it and try and live a 'normal' life instead of admiring other children/people's clothes and wishes that she could



Aisha on the day her halo was removed

get into them. Her sister Saffiya is very restricted what she can wear because of this and we don't know how we will ever come round this, as Saffiya deserves a life too. As a sibling she has been affected greatly as we have had to leave her behind with her grandparents in Leicester whenever we have had to travel to Manchester or have had to be with Aisha whilst she has been in hospital. Saffiya has had six months away from us with her Grandparents in Malawi and that for us was the hardest decision we ever took. She was only twoat the time. When we think of it now and look at her we feel we were very selfish for leaving her in Malawi, but hopefully for her it was a time in her life that she received all the attention and love she needed.

We have attended the MPS Conferences since 1999 and for the three days that we are there we share each others experiences as parents until we get home and once again we are alone dealing with the day to day of living with Morquio. After seven years you could say we are still in the process of accepting her illness. The process is not easy and accepting does not happen all at once. Acceptance is not about being happy about Morquio Disease. To us it means that we have made room for Morquio in our lives. The important part for us as a family is we have very strong faith and believe that there is a merciful creator and we live in hope that there will be a cure for Morquio's. We believe that Allah (God) watches over Aisha and this is what has given us the strength and will power to go from one day to the next. ■

Aisha and her sister Saffiya

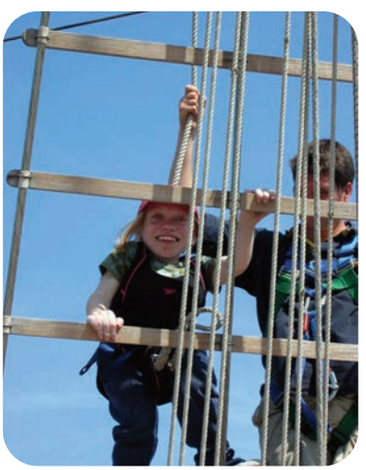
Tall Ships Adventure By Joanne Evans

After a very early start, a long flight from Heathrow to New York, and an interesting session with the neurotic American security and immigration services who couldn't seem to grasp that I wasn't staying in a hotel in America, because I was there to take part in the Tall Ships race, I eventually arrived in New London to board the good ship Tenacious and meet the rest of the crew - my home and family for the next 10 days.

I was truly amazed by the accessibility of the ship. The gangway is extra wide to allow wheelchairs to be pushed up, there are lifts everywhere and my berth is extra big to allow for my chair and you should see the showers! It just shows what is possible when people, in this case the Jubilee Sailing Trust, have the foresight to realise that disabled people have lives to live too.

I made friends with the crew really quickly, tried my first Hershey bar, was informed about the midnight to 4am watch duties (ergh!) and found the ship's dressing up box! I knew immediately that this trip was going to be quite an experience!

Typically, on my first day of watch, the weather threw everything it had at me - driving rain and gale force 6-7 winds! My wet weather gear was well and truly tested, but it just added to the challenge. Thank goodness for the waist harness so I could tether myself to the deck!



On only my second day came one of my proudest moments. I was down in my cabin, when one of the guys, Phil, came looking for me saying 'We're bracing the yards - we need you'. I don't think I've ever been 'needed' like that. So many times, jobs are just created for me because people think it will make me feel involved, but this time I really was one of the team.

The next day, the weather was a total contrast. Gorgeous hot sunshine and perfect weather for my first attempt at helming the ship. What an incredible feeling! You've got this massive ship, and you're in charge of the direction she steers in! The helm is a bit heavy, so my watch-leader Kristen helped me with it, but otherwise it was all 'little ole me'. Just to top it off, as we came off our wonderfully hot and sunny watch, a pod of dolphins appeared at the side of the ship and started showing off for us. Amazing!

The following days were spent between watches, helming the ship, having fun with the team and keeping the log up to date. The best bit of the voyage came on day 7 when I got to climb the mast (Dr Wraith - please don't read this bit!). This was the one thing I'd been waiting to do. At one point I almost cried with disappointment when we discovered that the body harnesses were all too big for me, but the crew were adamant that I could do it and, with their ingenuity and Nicki's swimsuit over the top of my clothes as a makeshift harness, I did it!

Nick, the first mate, did the climb with me and loads of people who were watching, said they'd never seen an assisted climber go up so fast and independently! The whole thing was filmed by a Canadian television crew who happened to be around that day, and was shown on Canadian television news that evening; then the press descended on me and I was in all the newspapers in Halifax the next day. As a result I was recognised whenever I went ashore - great fun while it lasted, but I don't think I'd like to be a real celebrity!

On the 8th night there was a big party at the Casino Hotel in Halifax for all the voyage crews competiting in the ASTA Tall Ships Challenge Series 2004. It was a great night but, of course, the highlight was Tenacious winning the New London-Halifax leg of the race and Best Overall. The evening ended with a spectacular firework display over the harbour.

It's hard to know how to sum up my experience and how to end this piece, except to say that this was probably the best time I've ever had in my life - so far! I really was part of a team where everyone was equal - it's so easy to do, if you think about it. I enjoyed every minute and cannot wait to return to Tenacious to do another voyage.

Conference on Lysosomal Diseases of the Brain

By Christine Lavery

In May 2004 a conference on Lysosomal Diseases of the Brain was held in Bethseda, near Washington, hosted by the Children's Gaucher Research Fund. Unfortunately neither Ellie Gunary nor I could attend, however, this is the feedback from those who did attend.

During the conference, representatives with experience in a range of lysosomal disorders, presented evidence clearly showing that what they had found in their research did apply to other disorders, as there are so many similarities in the presentations.

Tony Futerman from Israel and Kondi Wong from Texas started with the 'Pathology of Type 2/3 Gaucher disease' and 'Neuropathology of Gaucher disease'. This was followed by Steve Walkley from New York who presented his work on 'Secondary Glycolipid Accumulation in Lysosomal Disorders', showing that this is a common feature. Richard Proia from Bethesda finished the morning by looking at 'The Role of Inflammation in the Neurodegeneration of Sandhoff disease'.

The afternoon started with Ed Schuchman from New York looking at 'The Use of Bone Marrow Derived Cells for the Treatment of Brain Disease in a mouse model of Niemann-Pick Disease', followed by Charles Vite on 'Adreno-Associated Virus Vector-Mediated Gene Transfer to the Brain of cats with Alpha-Mannosidosis'. David Wenger from Pennsylvania ended the afternoon session by looking at 'Treatment Approaches for the Mouse models of Globoid Cell Leukodystrophy'.

Three more presentations from Oliver Morand (Switzerland), Pedro Huertas (California) and Greg Stewart (Massachusetts) finished off the evening.

On the Saturday talk turned towards therapies and whether what worked for Gaucher, for example might work for Niemann-Pick. Fran Platt from Oxford opened looking at 'Substrate Reduction Therapy', followed by Ari Zimran from Israel presenting 'Zavesca in Adult Patients with Type 1 Gaucher Disease: Implications for use in Neuronopathic Forms'. Late morning speakers, Jian-Qiang Fan from Amicus therapeutics and Greg Grabowski, a molecular geneticist looked at 'Active-site specific chaperone therapy: implications in Lysosomal Storage Disorders'.

In the afternoon David Begley from London explained why the blood-brain barrier was a problem - 'The Blood-brain Barrier, Delivery of Therapeutics to the CNS, the Problems and the Possibilities'. Finally William Partridge from California talked about 'Intravenous, Non-Viral Gene Therapy of the Brain'.

Roscoe Brady and Greg Macres rounded off the conference praising the work that all these scientists and clinicians were doing to bring together the Lysosomal Disorders with the common aim of finding therapies. Hopefully following the success of this scientific meeting, there will be future plans to encourage professionals involved in Lysosomal Storage Disorders to come together on a regular basis to share their knowledge and experience, with the aim to understand these diseases better and to identify possible future therapies.

Paris Workshop on Fabry Disease

Dr Uma Ramaswami, Christine Lavery and Professor Elizabeth Neufeld

By Christine Lavery

An Autumn workshop for professionals was held in Paris to which I was invited. The delegates heard presentations on the pathophysiology of Fabry Disease, how it affects women, the cardiac manifestations and Fabry in childhood.

The highlight of the workshop was undoubtedly the plenary lecture given by Professor Elizabeth Neufeld who gave a historical overview sharing how she discovered enzyme deficiencies in MPS I and MPS II in the late 1960s that have directly led to the current understanding of MPS and Related Diseases today.

International Fabry Symposium Rome, November 2004

By Sarah Cornwell

Virginia Laffin, Graham & Susan Soanes

Well, where do I start? The weekend in Rome came and went so quickly in comparison to the palaver of organising, in what felt at the time to be endless weeks, before hand.

Having managed to organise 70 members of the MPS Society to get on the right flights, there was the issue of sorting the rooms in the hotels. Initially many of you would have been in a double bed with your brother or sister, a single instead of doubles or even sharing rooms with a stranger. This was eventually sorted out, but communicating in differing languages always causes confusion. Somehow I had also acquired a 2nd 'boyfriend' who I was supposedly sharing a room with. This was, however, promptly sorted out at the lovely hotel reception on arrival.

Thursday 11th November - the beginning of the long weekend. Having managed to get everyone on a flight the majority turned up in Rome at their planned arrival time. There was, however, bound to be an incident. 'Reliable' Easyjet had delayed their flight from Northern Ireland by over 5 hours, meaning that two of our couples missed their connecting BA flight. This is the last thing you want to hear having just arrived in Rome yourself and been up since 4:30am. Luckily, I say luckily although I don't think this was the feeling from the couples, they managed to get onto another flight out to Rome arriving late that night. Once they arrived it was time to make the most of the weekend.

Paul & Margaret Black, Glenn Ming, Deborah McAlessee

After having our organisational skills, travel agency skills and patience been put to the test, it was time for a quick sight see of the Vatican City, a bowl of spaghetti in an old Italian restaurant and then back to the hotel for a meeting with the organisers and dinner with all our members.

Susan & Sarah Hill, Gemma Corder, Jams Rowbotham

Following a restful night, it was time to learn more about Fabry Disease. Friday 12th November. Firstly there was the buffet breakfast, we then had 2 ½ hours of doctors giving us a clinical update on ERT for Fabry. This was followed by a coffee break consisting of many hand made chocolate truffles, an assortment of cakes and juices and fruit for those feeling healthy. Then came another two hours of the conference this time it being a question and answers forum, where some of you asked some very interesting questions challenging the professionals from all countries. This was followed by a buffet lunch. After lunch there was another two hours of the psychological aspects of Fabry Disease, another coffee break and more questions and answers.

At the end of the day we all gathered for a group photo by the pool outside, strange for November, but having said this the weather was gorgeous, clear blue sky and mild. It was then time to relax! Dinner and a few glasses of wine later, everyone was enjoying themselves. A quick exchange of a white bottle for a red bottle occurred with the Camfields and the evening continued. We moved on to the packed

Piano bar and were pleased to see everyone enjoying themselves. So far it had been a success.

But work does not stop for the MPS Society. Having got our 2 couples safely from Northern Ireland there was suddenly a panic as to which flight they took back? Technically they had 2 choices. This is when Christine, who on occasions can only be described as being a Duracell bunny, leapt into action. Out came her laptop. Little did we know that the connection in her room would not work without a gadget called a WiFi. The pronunciation of this caused fits of giggles down the phone to reception, which combined with a language barrier made matters worse. Therefore a poor unsuspecting young man, from reception, came up to the room to sort out 4 giggling women and their inability to connect to the Internet after a 'long day'.

Ellie Gunary, Sarah Cornwell, Clare Cogan

Unfortunately he wasn't able to shed light on this situation, so we proceeded to follow him down to reception to use their Internet facilities. It was heading towards 1am and all we wanted to do was to go to bed safe in the knowledge that these two couples were on a flight home. After much perseverance we finally found what we wanted, though not before a few free cups of tea. Served to us on a silver tray with a smile at 2am.

Jack and Debbie Johnson

Saturday 13th November. Many members were less enthusiastic at breakfast following a long day before, therefore all were grateful of a ½ day conference with the afternoon free for sight seeing or a little respite. That evening was the Gala dinner, coaches booked for 7pm, everyone dressed up to the nines and looking forward to a lovely evening. The restaurant appeared to be outside under a large canopy, we were greeted with a glass of champagne and nibbles. Dinner was lovely although there were many courses and much food left. Surprisingly this was not the case with the wine!! Members were serenaded by a small traditional group, who were making their way round the tables. All in all everyone had a very enjoyable night.

Sunday 14th November. The end!! Many had early flights back home and those on the Heathrow flight grabbed a couple of hours after the mornings group meetings to browse round Rome and grab a pizza for lunch, before heading home.

The weekend flew by, we all learnt a lot from the conference and it was interesting meeting patient's from other countries. Let's hope that next year's hotel is as luxurious as this ones'! ■

Ian Hedgecock, Derek & Susan Evans

Claire & Thomas Harris, Ann Hedgecock

Michael Burke shares his experience of the Rome Fabry Conference

The 4th International Fabry Conference in Rome was a little more special to me than most. It was the first time in my life that I had ever been abroad, and at the tender age of 52 years, it was probably one of the best locations in the world to start.

I was first diagnosed with Fabry Disease in 1970-71 after about 15 years of different diagnoses such as Rheumatoid Arthritis, growing pains, malnutrition etc. The attacks of Fabry Pain were very severe and regular in the early years and the sun, heat and cold affected me very much. The only treatment for the illness then was pain management, regular pain relief, injections and hospital stays.

In my 20s and 30s, when all my friends were regularly going on their annual holidays from work, I remained at home. The reason for this was that I was afraid to go abroad in case I became ill with Fabry Pain. What would I do in a foreign country? I could not just walk into, or be carried into hospital and 'request' morphine or other pain relieving drugs. I know Fabry Disease is rare but it was a lot more rare 30 years ago when I was first diagnosed. I also suffered gastrointestinal problems and would not risk an attack which I believed was common on foreign holidays.

So, the conference in Rome was a very special 'working holiday'. It is Fabry Disease that kept me from holidaying for the last 30 years and it is thanks to Fabry Disease that I finally started to live a little. Thank you to the MPS Society for giving me the opportunity to attend the meeting.

Saved by a Wonder Drug by **Christine Doyle**

Jenny Dickinson's father died of Fabry when she was six, so she knew the life-threatening, hereditary disease was in her family. However, while she was growing up, doctors believed it did not affect women severely, if at all.

It was almost too late before she linked the childhood bouts of burning pain in her hands and feet and her frequent fevers with her father's condition. The symptoms had diminished as she grew older, but 10 years ago, more serious ones emerged.

'I had severe abdominal pains, nausea and diarrhoea, extreme tiredness and high temperatures,' she says. 'My GP gave me antibiotics in case I had an infection and, over the next few years, I saw a sequence of specialists. Few inquired into my family history and I still did not make a connection. I reduced my job as a travel consultant to two days a week, but the unexplained symptoms and exhaustion grew much worse.'

Not until Jenny decided, by chance, almost six years ago, to read some articles about Fabry did the truth strike home. 'I had all the symptoms,' she says. In Britain, the disease severely affects about 600 people. The accumulation of glycolipid, a product of fat metabolism, causes non-cancerous skin growths to form over the lower part of the trunk. Men tend to be most severely affected, though 60 per cent of women with the gene have some symptoms and, like Dickinson, can suffer severely.

When she was diagnosed the only drugs on offer were painkillers. 'It was a dreadful time for my family,' Dickinson says. 'We all felt depressed and cried a lot. Soon, I was in the first stages of kidney failure.'

Dickinson, now 44, knew that two drugs that make up the enzyme deficiency were being trialled and, three years ago, she was treated at Addenbrooke's Hospital, Cambridge, with one called Replagal.

'Within six months, I changed dramatically from being terminally ill to being my usual cheerful self. I even returned to work.' She will have to take this costly drug for life. To her relief, however, her two children, aged 17 and 13, do not carry the gene.

Dr Stephen Waldek, from Hope Hospital, Salford, who has worked on Fabrazyme, the other drug for Fabry, urges greater awareness. 'This is now a treatable condition. If we treat early, we could prevent the damage developing.'

This article is reproduced courtesy of the Telegraph Group Ltd.
The article first appeared in the Daily Telegraph 5 October 2004.

The Magic of Disney

at the MPS National Conference, December 2004 by Christine Lavery

Back in April discussions took place with the organisers of the Ollie G Ball that funds raised might go towards giving 15 families the chance of a lifetime to participate in the American National MPS Society Disney Conference. It all seemed very straightforward back then! Several hundred MPS member families applied and a sack of names were taken to Chenies Primary School where 15 names were drawn out, plus a few reserves. In fact, we took 16 families in the end.

In the run up to our departure for the Disney Conference Gina sorted everything from hotel, change of hotel, air flights, coaches, wheelchairs, oxygen, special diets and not least the mounds of paperwork required to enter the USA.

Wednesday 15 December and Ellie and I have arrived in Orlando. Help! Our flight was late leaving New York and now we have only a few minutes to get to the other terminal and meet up with the MPS families from the Manchester flight. There is disaster as one family missed the connecting flight in Atlanta! Several hours later and the Gatwick flight arrives and so, thankfully, does our missing family from the previous flight who arrive at the hotel.

From here on some of the families will take you on a tour of Disney over the next few pages. All there was for Ellie and I to do was to say farewell and wave the coaches off.

Manchester Airport - Clare Cogan

Wednesday 15 December 2004, 8:45am, standing at the Delta Airlines check in desk at Manchester Airport with tickets to Florida, was I excited?

Of course, but not for me, for the six families that were flying out that day from Manchester to meet up with Ellie and Christine in Disneyland and attend the American MPS Conference! I'd be lying if I said I wasn't a little wistful. One of my dreams has always been to go to Florida but the best part of it all was seeing the excitement on the children and adults faces as they arrived to collect their tickets and wishing them a happy holiday.

Then I got to go on a plane myself, but only back to Heathrow, never mind, maybe next time…

Our trip to the American MPS Conference in Disney World, Florida

By the Longley Family

Chris, Jacqui and Faye (MPS IV)

We were one of the families who were lucky enough to be picked to attend the national MPS Conference in Disney World Florida. We had never been to an MPS Conference in the UK so this in itself was a first for us. But to experience one in sunny Florida was a real treat!

We were travelling on the flight from Gatwick and the first opportunity we had to meet the other ten, or so, families was when we all turned up in the Departure Lounge ready to board our flight. As you can imagine, it wasn't difficult to spot the other families. I have never seen so many wheelchair users waiting to board a flight before. I'm not sure Delta Airlines knew what they had let themselves in for! By the time we reached the entrance to board, we had to pick our way through a variety of buggies and wheelchairs, littering the walkway. None of us really knew each other andwe all exchanged pleasantries to begin with, however after about nine and a half hours on the flight to Atlanta, we all got to know each other pretty well!

Our flight finally arrived in Atlanta and we had to endure the dreaded Immigration procedure where the Immigration officers are deadly serious and ask all sorts of stupid questions about why you are visiting! During this very serious process, there was a very light hearted moment where young Amie Oliver was sitting on the desk whilst the family were being 'interrogated' - however she was more interested in squashing her face up against the security glass, trying her best to make us laugh!

On arrival in Florida, we were greeted by half the Lavery family (Ben and Lucy) who did a fantastic job of rounding us all up and getting us on the coach without leaving anyone stranded at the airport.

At the hotel, the other half of the Lavery's (plus 'adopted daughter', Ellie) were there to meet us and get us checked in. Pretty soon (after a quarter mile hike to our room!), we were tucked up in bed and fast asleep. Well, we were, but after chatting to other families, some weren't that lucky. Unfortunately, some people's body clocks didn't adjust too well to the 5 hour time difference and at 3am (8am UK time), they were awake and ready to start exploring the parks!

In the morning, we experienced our first All American Breakfast - Gallons of coffee, fresh fruit, pancakes, waffles, muffins and eggs done a thousand different ways. What a way to start the day!

The first day was spent exploring Magic Kingdom, which was just a short monorail journey from our hotel. Despite the huge size of the park, we kept bumping into families we had met the day before and exchanging experiences of the day.

The evening was the first of the Conference Seminars preceded by a welcome dinner - for 800 guests - all in one enormous room. I have never seen so many people together for a dinner at one time. What struck us was the informality of the occasion - families strolling in at various times over a two hour period and 'grazing' on a huge hot and cold buffet. Following an introductory speech, we attended 'break-out sessions' for each of the MPS Types. Our family all trekked off in search of the MPS IV room, not knowing quite what to expect. We arrived to find just one other family there, whose young son, Ryan (aged 10), was

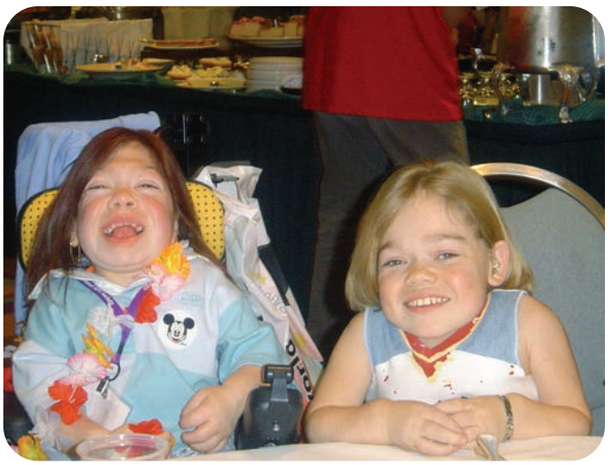
Part of the Florida group

affected by Morquio's. In the next few minutes just two other MPS Sufferers arrived - DJ (Donald Junior), a 46 year old from California and Sheryl, a 45 year old special needs teacher. This turned out to be an extremely interesting couple of hours, probably the most useful of the whole conference, swapping experiences (and e-mail addresses).

Day 2 was started with the customary mammoth breakfast followed by the day at Animal Kingdom, which was very different to the other Theme Parks. Dinner that evening was another unique American experience - a Box Meal which was a 'Subway' Roll, crisps etc. Accompanied by a rather welcome addition - a can of Budweiser (added, I suspect, by our UK hosts!). The conference that evening consisted of a talk about the MPS diseases and their diagnoses. Following the conference we all met in the Lavery's room and were treated to some liquid refreshment and an excellent view of the Magic Kingdom Firework Display.

Day 3 we decided to give the parks a miss and head for the Shopping Mall! With the Christmas Sales in full swing and the very favourable exchange rate, many bargains were to be found and we returned to the hotel laden with bags from every Designer Outlet you could think of - as a father of two teenage girls, I can tell you that they really know how to shop! The evening meal was a hot buffet laid on by the MPS Society at which we were subjected to another American 'experience' - following a very nice main course of chicken or pasta, we started on the sweet trolley, which was accompanied by a very large bowl of 'Cream', which we spooned on to our chosen sweet. What a shock we had when we discovered the 'cream' was, in fact, butter! Yuk! By the end of dinner, there were dozens of sweet plates, all with discarded butter. The American's must think we English have strange eating habits (but they can talk!). The Conference that evening was mainly about treatments and US Legislation, probably the least useful of all the sessions.

Day 4 was our final full day and started with an enjoyable 'Group Breakfast' with the entire UK contingent. The rest of our day was spent lazing by the pool and soaking up some very welcome (and warm) sunshine. The evening was the Gala Dinner, with yet another surprising 'twist'. We all arrived in the huge hall with tables (for 800 people) beautifully set and our eyes lit up at the sight of glasses of punch - the first alcoholic drinks that had been served with our meal all week. We all sat down and eagerly sampled the 'punch',



Amie (ML II) and Faye (MPS IV)

only to find that it was Iced Tea! Never mind, we did make up for it in the bar afterwards! The meal was lovely and it was a grand end to the Conference with a chance for everyone to dress up a little.

Day 5 and it was time to leave. Breakfast together again and then off to find the coaches for our trip back to the airport and our long flight home.

I am sure everyone came home with different but equally fond memories and experiences. We met some genuinely lovely people, we have all agreed to keep in touch and we're looking forward to seeing some of them at the UK Conference in June. We would like to extend our thanks to the Ollie G Ball who funded the trip and the UK MPS Society who were our excellent hosts throughout! The memories will last for a very long time.

Chris, Sophie and Faye (MPS IV)

Letter from America MPS Conference, Florida, December 2004

By Vic and Sue Lowry and Colin Holland (MPS II)

Dippity do da, Dippity da, wonderful feeling coming our way. There's a blue bird on our shoulders, in fact it's actual, everything is satisfactual thanks to all the organising and hard work put in by our USA friends and the dedicated team in Amersham. We felt extremely lucky to have been drawn to go to Florida along with 15 other families to meet up with our 'American Cousins' for their MPS Conference.

Never having been to that part of America we weren't too sure what to expect. We imagined it was probably more for children, but hey, if you weren't 8 years old when you got on the Monorail, you certainly were when you went through the turnstiles at the 'Magical Kingdom'. It just has that quality that transports you back to childhood. The euphoric feeling of being let loose in the Candy Store and you have all day to explore it! Wow! And explore it we did, and Epcot, and Downtown Disney, and MGM Studios. We had a very magical time and some good laughs along the way.

As it was close to Christmas, everywhere was decorated with the most beautiful lights and highly decorated Christmas trees, standing as tall as the Eiffel Tower, well they looked that tall from where we were standing. There were lots of stalls selling all sorts of interesting food. The speciality being roasted Turkey Legs! It left us all pondering what size the turkeys were, cos' the legs were like

something you would see on the Rosemary Conley Hip and Thigh diet, before the diet! Families of Americans were buying one each and walking round with these huge thighs in their hands. Fred Flintstone came to mind, but that's the sort of place it is - everyone can be a Disney character.

Every time Vic and I wandered off to find 'the bathroom', we came back to find Colin chatting away to someone. That was one of the very special qualities of Disney World, people smiling at you, engaging in conversation, genuinely wanting to talk. I think I can say on Colin's behalf it gave him a great boost of confidence.

We had a wonderful time, and met some very special people, made new friends who we look forward to staying in touch with. We want to thank our MPS Society for giving us the opportunity of discovering a little piece of Heaven on Earth where everyone of every nationality and disability and ability can feel 'the magic'.

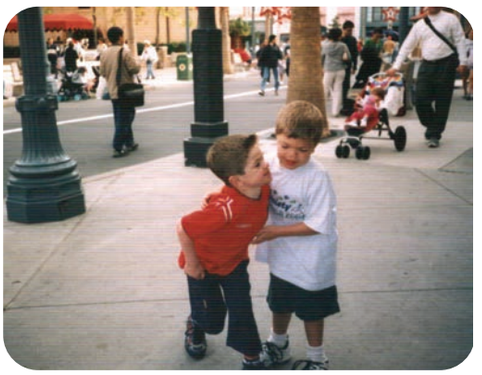
When you wish upon a star
Makes no difference who you are
Your dreams come true...

A big thank you from the Ingram Family

Hi there! Thank you for making it possible for us to experience the magic of Walt Disney World. We couldn't possibly begin to explain what a fantastic time we had because you would need all day to read about it! All we can say is that it was a fantastic, fun-filled holiday but also a great chance to meet other families from around the world, a couple of which we shall be staying in touch with.

The conferences at night were important for us as they allowed us to compare how the U.S. deals with issues which are relevant to Matthew's needs at present in the U.K. and by talking to families from the U.S. we could get an insight into the way their system works. The conference also allowed us to help give one particular family a positive outlook on an issue of theirs which, until they had spoken to us, was anything but positive and just knowing we had put a whole new brighter picture on their problem made us realise another reason why these conferences are so important for families with children with MPS.

Yes, we got a lot out of the conference but to be quite honest the highlight was the trips to the parks, the fantastic rides and 3D shows and Matthew getting pictured with all his favourite characters, Mickey Mouse, Donald Duck, Lilo and Stitch, The Incredibles etc,etc,etc .



Elliot Matheson and Matthew Ingram

We also made great friends with the families who travelled out from the U.K with us and will also be keeping in touch and meeting up at the various U.K. conferences in the future.

We would also like to thank Ellie and Christine who dashed around arranging things for us while we were out all day having fun in the parks. So, once again, thank you for all your hard work arranging the trip for us (that includes all the MPS team i.e. the ones who were not over there with us) and also a massive thank you to anyone involved in any fundraising or sponsorship which helped fund the trip. It was an unforgettable experience for us as a family. Thank you so much!

Our American Adventure by Alan and Linda Tucker

We just had to write and tell you how much we enjoyed the conference trip to Florida. Joe, Alfie & Eleanor had the most magical time there!

They woke each morning (unprompted mind, which is a first for them!) at 7:30am. They even helped each other wash and dress as they didn't want to waste a single moment. What was stranger was the fact that they were even civil to us!

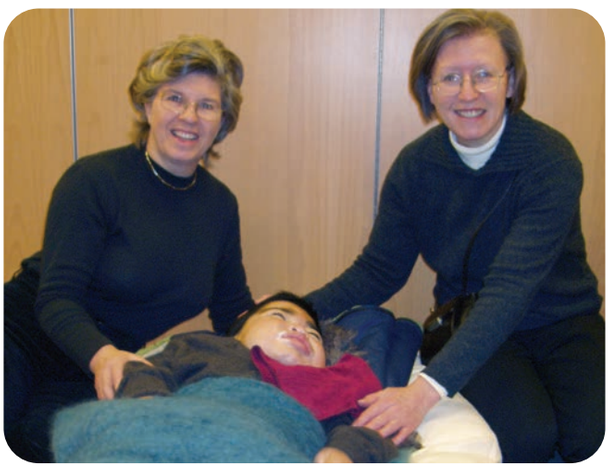
I was so proud of Joe as he ventured on some of the most terrifiyng rides, though I must admit we had to tell a few fibs to get him in there in the first place. After one such ride we feared he would need counselling for the trauma but then he would break into a beaming smile as he quietly congratulated himself.

I know that Joe, Alfie and Eleanor will hold on dearly to their beautiful and magical memories for a very long time, if not forever, and I wish to pass on our sincere thanks to our very generous benefactors, who gave our family the opportunity to enjoy this once in a lifetime experience that we will never forget.

MPS Week at Frambu - December 2004

By Cheryl Pitt

Christine Lavery, Dr Maureen Cleary from Great Ormond Street Hospital, and I arrived in Oslo on a cold and snowy Monday evening. The following morning, bright and early, we were whisked off to the hills a few miles outside of Oslo to a centre for rare diseases, known as Frambu.



We donned our heavy coats and boots and prepared ourselves for the sub-zero conditions. As you can see from the pictures, it really was a winter wonderland. Frambu is set in woodland, next to a large lake, which was frozen solid, and felt a million miles away from civilisation.

Photos clockwise from top right: Stian Pettersen (MPS II); Stine Rudi (MPS I); Norwegian Winter Wonderland; Christine Lavery, Oddrun Gronvik & Einar Gronvik (MPS II); Frambu.

It is a wonderful centre however, which includes a conference centre, a school, which the children attend while their parents are busy at conferences, a play area, accommodation, and two dining areas. There is also a lovely warm swimming pool, and an information centre.

Over two days we met Norwegian families who had children affected by MPS disorders. The families met with Dr Cleary for a medical consultation, and then with Christine and I to discuss any pressing issues of disease management, treatment, or social issues.

On the third day we attended, and presented talks, at a day conference held at Frambu. All the families we had seen over the previous two days attended the conference, as did professionals in the field of MPS diseases. It was a privilege to meet all the families and share their experiences, and we hope that they found our input at the clinics and conference useful. We felt that our week had gone very well, and that we had been well looked after.

So, thanks to Oddrun Gronvik of the Norwegian MPS Society, and all the staff at Frambu for their hospitality (and translating skills!).

Morquio Children in India survive the Indian Ocean Tsunami

In the Spring 2004 MPS Newsletter we told the story of Radhika, Marimuthu and Manikandan who have Morquio Disease, MPS IV, and live in India. Elizabeth Herridge, who accompanied her husband, the British High Commissioner, for a four-year posting to Chennai (Madras) in Southern India tells us how the children survived through the Indian Ocean Tsunami.

You may have been wondering how the three children in Chennai survived this huge and terrible disaster. Maybe Uma (Ramaswami) won't have been able to get to them as I imagine all doctors available are called upon...

Anyway, the news isn't too bad.... Which is a miracle as Cheshire Home is not too far from the sea. Doubly miraculous is that Maureen Murari who 'runs' the home suggested that Manikandan always misses out on Christmas treats and that for this year he should stay at the home over Christmas and Boxing Day! He has a mother, ancient grandmother and sister who live on a palm hut on the beach (no father around).

Radhika and Marimuthu (both MPS IV)

Immediately it happened, our ex-housekeeper went to the beach to look for them and found them sitting on the sand crying their hearts out as they lost absolutely every single thing, including their house of course. But they were still alive.

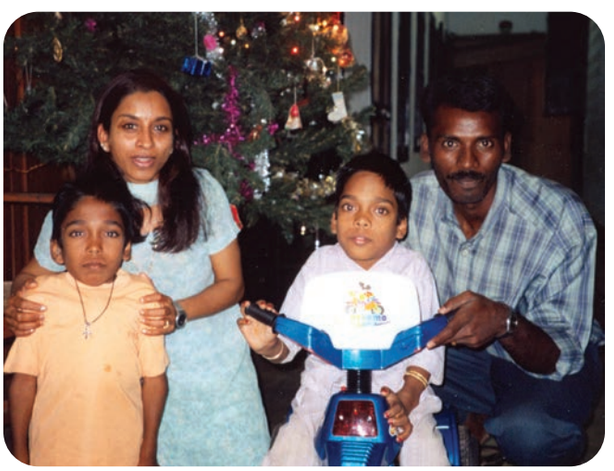
I am starting a bit of a collection from our very long suffering friends and we hope to get enough to rebuild their house and get them the few essentials they need to live. God knows, they had absolutely nothing in the first place. The Government may provide some sort of relief but it may never get directly to those that need it.

Dr Uma Ramaswami, Radhika and Marimuthu (both MPS IV), Ellie Gunary

Marimuthu and Radhika were travelling home when it happened and they are OK and so are their parents so that is good. Everyone at Cheshire Home is safe also.

There is lots of water around in Chennai and now disease warnings have been issued because of the fresh water. Ironic isn't it? The water came up the rivers.

Anyway, I thought you might like an update on the children and the situation. If only I was there. I have suffered severe frustration these past two days! Michael said probably just as well or we would have filled the Residence with the homeless!



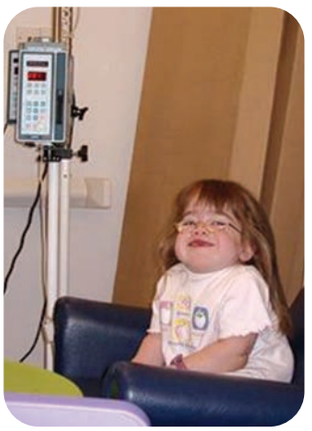
Radhika, Dr Uma Ramaswami, Marimuthu and staff member

If you would like to make a donation to the family please send us a cheque made payable to the MPS Society and we will forward this on. Please indicate clearly that your cheque is for this purpose.

www.mpssociety.co.uk

MPS VI Update (One year on...)

Jane Roberts, Willink Biochemical Genetics Unit, RMCH



Martina Lynch

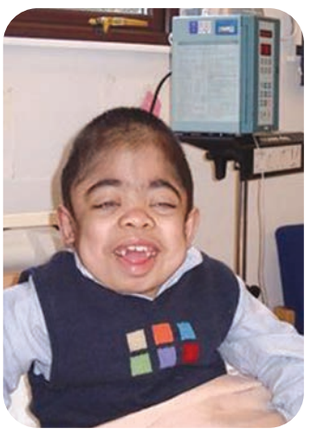
'Did you know that it is a year since we started the ERT Trial?' said Martina's mum casually during her week 52 infusion... Well, yes, subconsciously I did, but it has only now really dawned on me what those words really have meant to the patients, families and study staff.

A full year of... Religious weekly attendance (still only one missed infusion at our site despite some very poorly episodes!), regular canulations/blood and urine collections, travelling... Whatever the weather/delays/illnesses/family woes etc, never-ending paperwork, data collection and transcribing (black pens only of course!), recurring full and rigourous weeks of investigations and tests, seeing gradual improvements in the trial patients health, amazing continuing commitment, motivation and support of all involved and too much coffee and chocolate! (I could go on and on!)

However, at present, it is a little quieter at our Manchester site as the overseas patients will not return until next summer. They have already been back twice for investigations and still lose their luggage each time! Also, we are working hard to get our other site patients to centres nearer to their homes... sighs of relief all round, fingers crossed!

As a point of interest, you may or may not be aware that during a trial we all have to adhere to a strict protocol but despite this, we managed a surprise new recruit at week 36 as our research nurse, Sarah, had a baby girl called Ellie! Both are doing very well.

So, there you are for now. But keep watching out for any news from the regulatory authorities which will hopefully lead to licensing of the drug and then no doubt some of you will be getting on the carousel of weekly infusions!



Junadal Islam

My New Job! Jean Mercer, Clinical Nurse Specialist (MPS I)

I have been asked to write this article to introduce myself in my new role. Some patients may already have met me either in the Willink or at Conferences.

I have been working in the Willink for four years as the Senior Trial Coordinator on the Genzyme studies which included the MPS I Patients. I had previously worked with Ed prior to having my own children and returned after experiencing life elsewhere in the hospital. My time at the Willink has been hectic and fulfilling as the Enzyme Replacement for MPS I was given a license in June 2003, thanks to the enormous effort and commitment shown by the patients and families.

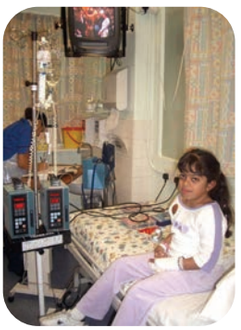
It was based on this that I applied for the post of Clinical Nurse Specialist in MPS I. This role covers all of Ed's patients with MPS I. I will be involved in the clinics at Manchester including the Bone Marrow Transplant Clinics. For all the patients who need it I will remain the contact for queries and problems.

In line with the recognised treatment of MPS being Bone Marrow Transplantation and/or Enzyme Replacement Therapy, I am working with all the relevant professionals, both here at Manchester and in the hospitals nearer home, to ensure the efficient treatment of the patients. Ellie has the pleasure of me being in contact with her, and we do keep threatening to meet over a meal!

The annual assessments of the patients will continue to be done by Mrs Caveney in the Willink Office. Gill Moss (Clinical Nurse Specialist in Metabolic Disease) and I liaise closely and the care of her MPS I patients is now transferred to me. The Willink is a team and will continue to work as one. The patients should be able to contact the relevant professionals, as will other professionals involved with the care of you or your child. I look forward to meeting more families.

Simran gets ERT

We are delighted that Simran has now started her ERT treatment. The photo here shows Simran at Great Ormond Street Hospital getting her first infusion which takes just over 4 hours. Simran will be going to GOSH every Friday for the first 6 to 8 weeks and then hopefully move to Northwick Park Hospital.



For Sale!

A family are wanting to sell an electronic 'go talk 6' book (purchased from liberator.com). It is a book where pictures can be inserted and on each page you can record your own message. The family are asking for £70 (Retail price is £150). Please contact Sophie at the MPS office for more information.

In the next edition of the MPS Newsletter we will be offering an update on Enzyme Replacement Therapy and newly published papers. Watch this space!

Child Tax Credit and the Working Tax Credit

Contact a Family have recently produced an article entitled 'Moving into Credit' which details that, during 2004-2005, payments for children that are currently included with Income Support are to be replaced by Child Tax credit awards.

Many parents on Income Support are likely to have questions about how this process will affect them. This article explains the process in more detail and offers contact details for further advice.

In April 2003 the government introduced two new tax credits to help families with children and people in work - Child Tax Credit and Working Tax Credit. Initially parents getting Income Support have been able to choose whether to claim Child Tax Credit or not. However, the Government's intention is that all claimants will eventually be transferred onto the Child Tax Credit.

This article looks at how payment of the Child Tax Credit will affect parents currently receiving Income Support. Similar rules will apply to families claiming income based Job Seekers Allowance.

What is Child Tax Credit?

Child Tax Credit is a means tested benefit that can be claimed by anyone with a dependant child. You can apply regardless of whether you work or not and it is paid in addition to Child Benefit. The amount of Child Tax Credit you receive depends on your personal circumstances, including the number of children. An extra amount is added to your tax credit calculation for each child who gets Disability Living Allowance (DLA), or who is registered blind. If your child gets the high rate DLA care component, a further amount is added.

Can I get both Child Tax Credit & Income Support?

From April 2004 onwards CTC is set to replace all Income Support payments for children. Anybody making a new claim for Income Support only gets amounts for themselves and any partner, with a separate CTC claim required for children. Between April 2004-05 all existing Income Support claimants will also have the amounts they receive for children replaced by a CTC award. Some parents will continue to receive a reduced amount of Income Support but for others it may stop altogether.

When will I start to be paid Child Tax Credit?

Parents on Income Support will automatically be awarded CTC. This will happen during the course of this year on an area by area basis. Some parents may be better off claiming CTC now, rather than waiting to be transferred.

If you stop receiving Income Support you will have to reapply for Housing Benefit and Council Tax Benefit. The amount of any new rebates will depend on your income (including the CTC payment). You should still qualify for help with NHS costs and free school meals, so long as your income remains below certain annual amounts. However, you will no longer be able to apply for community care grants or budgeting loans from the Social Fund.

Contact a Family employ a welfare rights specialist who can provide advice on any aspect of Tax Credits. Please call the Contact A Family freephone helpline on 0808 808 3555 (weekdays 10am - 4pm), or e-mail to: helpline@cafamily.org

Coping with Benefits
when a Partner Dies

Coping with bereavement is never easy and the last thing a bereaved person needs is a confusing mass of bureaucracy sorting out benefits. There are some key benefit issues to consider and things to do when a partner dies. The following article may be of some help.

Informing the Department of Work & Pensions

Firstly, the Department of Work and Pensions (DWP) needs to be informed that a partner has died.

A form BD8 will be given to complete. Information about the person who has died, their name, date of birth and date of death, will already be entered on the form. Information which needs to be entered includes the deceased person's address, their national insurance number, details of the state benefits the surviving partner receives if they include an element for the deceased person, whether the surviving partner wants to claim any benefits owed to the person who has died, the name and address of the person dealing with the estate and whether bereavement benefits are to be applied for. Once completed, the form should be posted to the local social security office, which will ensure the information reaches the appropriate offices.

If means tested benefits were received as a couple, only one partner would have been the claimant. If this is the partner who has died, these claims will end when the DWP receives the form BD8. A new benefit application will therefore need to be completed. If the surviving partner receives benefits, they will continue to receive them but the amounts paid may be reduced as soon as the DWP learns of the death.

Carers Allowance

If the surviving partner was claiming Carers Allowance as their partner's carer, they will be able to continue to claim this benefit for eight weeks following the death of the person cared for, provided they continue to fulfil the remaining criteria for eligibility for Carers Allowance.

Bereavement Benefits

Bereavement benefits are available to those who are eligible. Except in Scotland, these are only payable to partners who were married to the person who has died. In Scotland these are also awarded to a co-habiting partner, though substantial evidence is required.

There are three main bereavement benefits to which an individual may be entitled: Bereavement Allowance which is payable for up to 52 weeks to a spouse aged 45 or over when the partner died; Widowed Parent's Allowance which is payable if there are dependent children, (an individual cannot receive this and Bereavement Allowance at the same time); and Bereavement Payment which is a lump sum payment of £2,000.

All these bereavement benefits have extra conditions and further information will be required on each in order to be certain whether an individual is eligible.

Means Tested Benefits

When a person dies, as at any other time, eligibility criteria for other means tested benefits may be met. These include Income Support, Job Seeker's Allowance, Council Tax Benefit and Housing Benefit. Eligibility will depend on individual circumstances, such as whether or not a person is working or has children.

Funeral Expenses

If a partner is in receipt of a qualifying benefit, it may be possible to claim a funeral expenses payment from the Social Fund. Qualifying benefits include Income Support, Income based Jobseeker's Allowance, Housing Benefit and Working Tax Credit.

Additional criteria include that the funeral must take place in the UK, the deceased person must have been 'ordinarily resident in the UK', the claim is made at any time between the date of death and three months after the funeral and a funeral expenses payment must not have been paid in respect of the deceased to another claimant.

The DWP may not pay all costs of a funeral and the amounts awarded may be reduced by other monies received, such as any lump sums which may be due from an insurance policy on the death of the deceased.

www.dwp.gov.uk

New Employment Rights for Disabled People

From October 2004 it has been against the law for an employer of any size to discriminate against a disabled person because of their disability.

The Disability Discrimination Act (DDA) has given employees with a disability a new right to access to buildings. The Act also gives new rights to disabled people who already have a job, to those who become disabled during their working life and to disabled people who seek employment.

Under the DDA there are two distinct types of discrimination: Firstly, a person who receives less favourable treatment and secondly, failure to make reasonable adjustment to accommodate a person's disability. This is known as the duty to make 'reasonable adjustments'. For example, if an employer does not take reasonable steps to prevent a disabled employee from being at a substantial disadvantage because of any working arrangements or physical feature of the work premises.

For employers, the duty to make 'reasonable adjustments' is assessed against various considerations which include: How effective the adjustment is in preventing disadvantage, the financial and other costs of the adjustment, the ability of the employer to meet these costs, the availability to the employer of financial or other assistance to help make the adjustment and the extent of the disruption caused.

Large employers, (for example Local Authorities), should have developed 'Equalities Access Plans'. These include actions to promote and address all equality issues including those relating to access.

These may include: Changing the location of equipment so an employee who uses a wheelchair can access it, acquiring special equipment e.g. computers which have screen reading technology or phones with a loop system and adapting working hours to accommodate treatment needs.

Potential employees are also covered by the Act and reasonable adjustment is required at interview stage. For example, the building and room the interviews are to take place in need to be accessible for an interviewee who is a wheelchair user.

The following organisations provide information about the DDA and employment issues.

Disability Rights Commission (DRC)
Tel: 0845 7622 633
Web: www.drc.org.uk

Employers' Forum on Disability
Tel: 0207 4033 020
Web: www.employers-forum.co.uk

Centre for Early Intervention Feasibility Study Latest News

In the previous MPS Newsletter plans to develop a National Centre for Early Intervention for children with disabilities were introduced. The Centre for Early Intervention Feasibility Study has recently sent out its Autumn Newsletter. Here are some of the key issues announced.

Parents' Survey Results out Now!

Following over 200 replies to the parent survey the results have now been analysed. 90% of parents believed a national centre for early intervention would make a difference for families with a young disabled child; 97% thought it was important to have good links between research, training and practice. These findings have been used to inform the Parents' Views Report, which was submitted to the Department for Education and Skills in September.

Your Views!

Views of parents are still welcome. You can either email them to info@earlyintervention.org.uk or write to Ellie Gunary at the MPS office.

Launch of Parents' Report!

From April to September 2004, the Feasibility Study focussed on the views of parents. Along with the Parent Survey, a programme of Parent Consultation Days was carried out across the country and the study has been promoted by organisations that work with parents and represent their views.

The Centre for Early Intervention Feasibility Study has now collected together the findings of these events and produced a report that deals with the following issues: What parents say about the overall concept of a national centre; what the centre should do; how the centre could deliver its information function; views about a virtual or buildings based centre; views about central and local/regional sources of help.

The report and newsletter are available from www.earlyintervention.org.uk or by phoning 020 7976 6954.

Independent Living

Employing a Personal Assistant

Employing a Personal Assistant may be a key means of support for a person with a disability, enabling independent living. There is a vast difference between the role of a Personal Assistant and a carer. Whereas a carer will look after needs within the home, addressing health and cleanliness issues, a Personal Assistant (PA) will assist over a much wider area of requirements. These include things like driving, washing the car and filling it with fuel, grocery shopping, clothes shopping, organising travel and being a travelling companion. A PA may provide support at college, answer the phone, take care of bills and perform other administrative tasks.

To find a PA the local social care services may be approached or a PA can be employed by the person with a disability themselves. If an individual is looking to become the employer of their PA, the first task is recruitment. Registering with a private agency is one option to locate candidates, advertising in local and/or national newspapers and magazines, or listing the vacancy at the local job centre are others.

When recruiting it is important to consider what is wanted from the PA and the skills and qualifications they need to perform the specific tasks required. If, for example, moving and handling is required, it may be of importance that PAs have relevant training and expertise to perform these particular tasks. Alternatively it may be more important that the PA shares common interests with the employer.

Whilst a PA can really change the life of someone with a disability, there are issues which need careful consideration. It is important to find the right PA as they will spend a lot of time with their employer. It is also important to ensure references and police checks are fully completed and genuine. Although an agency may have 'vetted' an applicant, this does not guarantee that all the information given is correct or that references were nor forged or exaggerated.

Employing a PA is a two-way street and, as a PA is expected to be with an individual employer for a long time, it is important to consider the PA's long-term needs. Addressing issues such as living costs, personal telephone calls and time off from the start, can prevent misunderstandings ensuing. Agreeing allowances when agreeing a salary, to cover things such as food and laundry, may also be helpful.

The employment contract should include clauses on issues such as drinking whilst on duty, damage to personal property and smoking etc. Whilst an employer will want their PA to be on duty during times most suited to them, it is important to consider that the PA has sufficient time off to pursue their own interests and lead a personal life.

Funding for a PA is available through the Direct Payments Scheme, an assessment for which is carried out by the local social services department or health body. Other sources of help may include the Independent Living Fund.

Help in employing a PA is available from various sources including local social services departments. The National Centre for Independent Living, (NCIL), has information on the 'Direct Payments Scheme' which provides funding for PAs and information about local support schemes. The NCIL issues a newsletter. Log on to www.ncil.org.uk or phone 0207 587 1663.

Guides to Direct Payments are also produced by local social services departments and the Department of Health.

Government to Provide Heating Grants

Government to provide up to £2,500 heating grants for which disabled people on certain benefits can apply. Funded by the Government, the Warm Front Grant provides up to £2,500 to make homes warmer and more energy efficient. Eligible households receive a package of insulation and heating tailored to meet the requirements of their home.

In England, more than 4 million households live in fuel poverty, the main cause of which is a combination of poor energy efficiency and lower incomes. Fuel poverty is defined as the need to spend more than 10 per cent of the total household income to maintain an adequate standard of warmth. Lack of adequate heating can exacerbate existing health and mobility problems. Respiratory diseases, such as flu, pneumonia and bronchitis, are made worse by the cold and the risk of heart attacks and strokes is also increased.

The Warm Front Grants, managed by the Eaga Partnership are specifically aimed at people who own their home or rent from a private landlord. Disabled people on certain benefits are eligible for the grant as this group can have specific mobility barriers and cold-related health problems.

For further information on Warm Front or the Benefit Entitlement Check, householders should telephone Eaga Partnership free on 0800 316 3018 or apply on-line at www.wfteam.co.uk

Addenbrooke's Hospital Department of Paediatrics

Dr Uma Ramaswami MD, MSc, MRCPCH, is a consultant paediatrician with an interest in metabolic diseases. She has a special interest in MPS and related lysosomal storage disorders. She has recently been awarded a Genes for Jeans grant to further her interest in MPS and related diseases.

The Dept. of Paediatrics at Addenbrooke's Hospital is housed in the main building. In-patient care is carried out on three wards with a total of 64 beds. The Paediatric Intensive Care Unit (PICU/HDU) has 8 beds, 6 of which are staffed as ICU beds. The paediatric day unit provide a service for all planned day case activities, for example admissions for MRI scans, investigations, and enzyme replacement therapy infusions.

As well as the general paediatrics provided for the local population, the department also runs regional/local services for a number of sub-specialties, including neonatology, oncology, neurology, endocrinology, paediatric surgery including neonatal, ENT, orthopaedics and neurosurgery, metabolic, cardiology, allergy, hepatology, gastroenterology and renal medicine.

The department also runs a Child Development Centre (jointly with the local PCT). There are 30 paediatric consultants working in the various specialties. The paediatric service is supported by six paediatric anaesthetists.

Addenbrooke's has recently been NSCAG (National Specialist Commissioning Advisory Group) designated as a centre for management and treatment of lysosomal storage disorders in both children and adults. The department of Paediatrics at Addenbrooke's Hospital is one of three UK designated centres for the management of MPS and other lysosomal storage disorders, in particular, enzyme replacement therapy or substrate reduction therapy.

The Department of Paediatrics is also located within the main hospital building. The Departmental areas of academic interest include clinical and laboratory based research centring on endocrinology, diabetes, metabolism and neurosciences.

Addenbrooke's Hospital has a dedicated research unit, funded by the Welcome Trust and GSK. This unit is available to all consultants working at Addenbrooke's Hospital performing research, and has facilities for families to stay overnight if necessary, and is staffed by experienced paediatric and adult nurses.

Addenbrooke's Hospital is in a unique position with both adult and paediatric metabolic units on one site. The aim of this service would be to develop a comprehensive service for both children and adults with MPS and related diseases. Current paediatric research activities include clinical trials of enzyme replacement therapy in Fabry Disease, MPS II and substrate reduction therapy for Sandhoff's Disease.

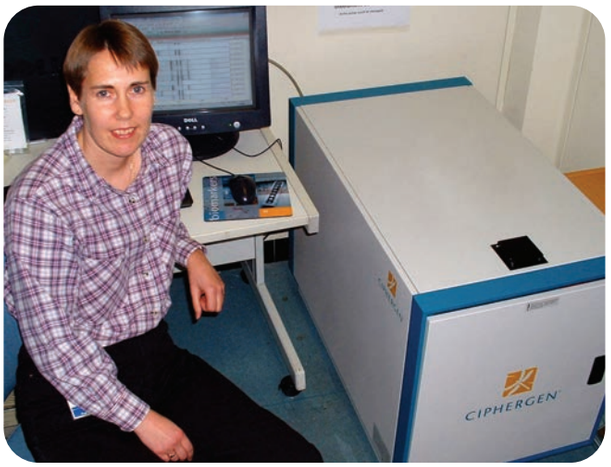
The paediatric metabolic team at Addenbrooke's Hospital consist of Dr. Uma Ramaswami; Junior Doctor Support, Paediatric SpR, HO (cover general, neurology and metabolic patients); Chief Biochemist, Dr. Jacqui Calvin; Chief Dietician, Mrs Lynne Radbone; Fabry Nurse, Mrs Marie Corcoran and MPS II Nurse, Ms Kathleen Woolverton. The adult metabolic physicians, Professor Timothy Cox and Dr. Patrick Deegan.

The Identification of Biomarkers in MPS Disorders

at the Institute of Child Health funded by Jeans for Genes

by Clare Beesley

In January 2004, the MPS Society awarded a grant of £72,827 over two years to the Institute of Child Health, London, to undertake a research project 'The Identification of Biomarkers in MPS Disorders'. The project is carried out by Dr Clare Beesley and overseen by Professor Bryan Winchester. Clare's end of first year report follows.



Clare Beesley next to the SELDI machine

Lysosomal storage diseases (LSDs) result from a fault in a gene that normally produces a functional protein/enzyme in the lysosomal system. This leads to the accumulation of specific molecules within the lysosomes of affected cells.

The mucopolysaccharidoses (MPS) are a group of LSDs in which the primary enzyme deficiency leads to the accumulation of partially degraded glycosaminoglycans (GAGs) inside the lysosomes of the cells of patients. The accumulation of storage products is progressive and therefore clinical symptoms deteriorate with age. Very little is known about the 'knock-on' effect of this accumulation on other components of the cell.

It is envisaged that the increased storage of GAGs inside cells will result in the appearance of storage products and other components of cells in bodily fluids, such as blood, at much higher concentrations than seen in normal unaffected individuals.

The disease may also cause a decrease in the amount of some proteins that are normally found in unaffected people. Measurement of these proteins (termed biomarkers) which are either increased or decreased in patients may provide biomarkers for a specific MPS disease or it may be found that some are common to all MPS diseases.

Identification of these biomarkers will enable us to understand more about what effect the storage is having inside the cells and how this is leading to the disease symptoms.

For example, are the cells dying prematurely, a process known as apoptosis, or are the cells becoming infiltrated with molecules involved in inflammation? The rate of appearance of these biomarkers in an individual will depend upon the underlying mutation in the gene and other genetic and environmental factors but they should provide a good indicator of the course of the disease.

As the novel forms of treatment being developed may reduce the storage in the lysosomes, the biomarkers could be used to monitor the effectiveness of the treatment and the improvement in disease symptoms.

In the Biochemistry, Endocrinology & Metabolism Unit at the Institute of Child Health our aim is to identify possible biomarkers in MPS disorders and to investigate the 'knock-on' effect of GAG storage inside cells by analysing differences in protein and gene expression between normal and diseased cells. The work that has been carried out so far has focussed on the identification of biomarkers in MPS I, IIIA and IIIB patient samples.

Our department at the Institute of Child Health recently purchased a machine from Ciphergen called a surface enhanced laser desorption/ionisation machine (SELDI) which helps identify putative biomarkers in patient samples. The technology involves the binding of proteins to the chemical surfaces of 'ProteinChips'.

Proteins exist in the human body as globular molecules which carry charges that are dependent on their structure and environment. There are also proteins which are hydrophobic (avoid water) and some which will bind to metal

ions. There are several types of chips that have different chemical surfaces and therefore bind different sub-groups of proteins. Some chips bind positively charged proteins, some negatively charged proteins, others bind either hydrophobic or metal binding proteins.

After a sample has been applied to a specific chip, only proteins that interact with that chip surface will be retained whilst unbound proteins are washed off with a buffer. The SELDI machine then analyses the proteins bound to the chip and generates a fingerprint of the proteins that is based on their size and which is representative of those proteins contained in the sample that have bound to that chip. Analysing the same sample on a different chip will generate a different protein fingerprint because it will bind different types of proteins e.g. hydrophobic proteins.

The software on the machine then allows you to compare a protein fingerprint from a MPS patient with that from a normal individual and any differences are highlighted. The machine only gives you an indication of the size of the protein, its charge, solubility or metal binding properties. Unfortunately it does not identify it for you!

Over the last nine months at the Institute of Child Health, blood samples from four normal individuals, four MPS I (Hurler), four MPS IIIA and four MPS IIIB patients have been analysed, in duplicate, on the SELDI machine using four different chip surfaces. So far, potential biomarkers have been identified in all three MPS groups. In MPS I, three proteins are increased in patients compared to controls and two proteins are decreased. In MPS IIIA, one protein is increased in patients and another (found also in MPS I) is decreased. In MPS IIIB, three proteins are increased in patients and one is decreased.

Only a small number of patient samples have been analysed so far so a lot more samples need to be screened in order to confirm and validate these findings. It will be interesting to see if potential biomarkers in MPS IH are present in the less severely affected Hurler/Scheie and Scheie patients. Also, with the availability of enzyme replacement therapy for MPS I, it will be interesting to see if the levels of a potential biomarker approach normal values during treatment. Once confirmed and validated then the next step will be to identify these potential biomarkers. ■

Psychosocial Outcomes of BMT for MPS I

Cheryl Pitt

Research Update

A very big thank you to all the families that have volunteered to take part in the research so far! I have already visited most of you who volunteered, and your participation is now complete. Those of you that I have spoken to, but not yet visited, will be contacted early in 2005 as arranged.

I am very pleased with the progress of this research so far, and am extremely grateful to you for giving up your valuable time to support this important work. I am also grateful to the children and young people that have taken part. You all put in such great effort, and had some fun along the way as well, I hope.

Some families are not yet involved in this research project, but further information will be sent out to you, so every family with a child affected by MPS I who has undergone a bone marrow transplant will be given the opportunity to participate.

If you feel that you should have been sent information about this research but have not, please contact me at the MPS offices. Similarly, if you have received information, but have difficulty understanding the aims or procedure of the study, please contact me and I will be happy to answer any questions you have. I look forward to seeing you soon.

Society for Mucopolysaccharide and Related Diseases

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