MPS

Care Today, Hope Tomorrow

Winter 2007

Society for Mucopolysaccharide Diseases

Mucopolysaccharide and Related Diseases are individually rare; cumulatively affecting 1:25,000 live births. One baby born every eight days will be diagnosed with an MPS or Related Disease. These multi-organ storage diseases cause progressive physical disability and, in many cases, severe degenerative mental deterioration resulting in death in childhood.

CARE TODAY HOPE, TOMORROW

What is the Society for Mucopolysaccharide Diseases?

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

What are the aims of the MPS Society?

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

To bring about more public awareness of MPS and Related Diseases

To promote and support research into MPS and Related Diseases

How does the Society achieve these aims?

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

Telephone Helpline
Includes out of hours listening service

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

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

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

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

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

Information Resources
Publishes specialist disease booklets and other resources

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

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

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

Cover photograph: Childhood Wood Planting 2007

This edition of the MPS Magazine is being funded using money raised by the 2007 Jeans for Genes Appeal

MPS Magazine

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Newsletter Deadlines

Spring	1 Mar 2008
Summer	1 Jun 2008
Autumn	1 Sep 2008
Winter	1 Dec 2008

Become a Friend of MPS

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

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CHIEF EXECUTIVE'S REPORT

Typically the Autumn is a continual round of collaborative meetings and specialist conferences providing unique opportunities to promote Mucopolysaccharide and related lysosomal storage diseases. This year has been no exception.

I would like to thank the members and their families who have allowed me to show their pictures and tell their stories to personalise the individual diseases and the impact on the affected child or adult in my presentations.

Major highlights of this round of meetings have included the invitation from EPPOSI to make the key note presentation on MPS diseases in the Danish Parliament in the presence of HRH Crown Princess Mary of Denmark. Equally important was the opportunity to present the MPS patients' perspective of accessing therapies at the Welsh Assembly in Cardiff. Some of our Welsh members will be well aware that whilst there are free prescriptions for all, accessing ERT for Fabry disease, MPS I and MPS II continues to be a lottery. It is to be welcomed that during this meeting Dr Graham Shortland, Consultant Paediatrician at the University Hospital, did announce that there is more money being released to fund a clinical service and therapy in Wales.

However, it is still most concerning that in October 2007 the All Wales Specialised Medicines Group (AWSMG) and Health Commission Wales (HCW) refused to recommend Elaprase for MPS II, Hunter Disease, for reimbursement in Wales. Elaprase joins Aldurazyme for MPS I and Naglazyme for MPS VI on the list of orphan drugs not approved for reimbursement in Wales. If there are members with Fabry disease who are still denied access to ERT in Wales even though Fabrazyme and Replagal are approved, please do let the advocacy team know.

As you will read further in the MPS magazine there is important new information on the progress of therapies for the other MPS and Related Diseases and we expect to be able to bring you news of further developments in the year to come.

The opportunity to participate in the pharma company lysosomal storage disease meetings are invaluable not just for what we learn from the presentations given by experts in Fabry and MPS diseases but the informal networking that allows us to learn in greater detail of the activities of the many scientists and physicians working throughout the world.

It was in just such a meeting attended by myself and Neisha Hall from the advocacy team that we found ourselves at the same table as Dr Joseph Muenzer, Consultant Paediatrician in Chapel Hill, North Carolina. Over dinner we learnt that Dr Muenzer is planning a clinical trial for intrathecal enzyme replacement in young patients with MPS II who untreated are going to develop progressive neurological disease. It is hoped that any success from this clinical trial may also lead to a similar therapy for MPS III, Sanfilippo Disease.

During the American Society for Human Genetics Meeting in San Diego I was able to meet with Dr Tomatsu to discuss progress on ERT for MPS IVA, Morquio Disease and to invite him to join us for our first conference specifically for those affected by Morquio disease 29-31 August 2008. We are also leaving space in the programme for any breaking news. During an LSD meeting in Brussels we met with Jenny Noble who runs Lysosomal Diseases New Zealand and is the mother of a son and daughter with ML III. Jenny has kindly written an article featured later in this magazine.

Moving into 2008 we have a very full and exciting events programme so please do be careful to read not only the MPS Magazine but any enclosures and we look forward to seeing as many of you as possible at these events in the coming year.

Finally I want to wish all our members, their families, the scientists, doctors, nurses and industry representatives a Happy New Year and thank you all for your support to the MPS Society over the past year.

Christine Lavery
Chief Executive

News from the MANAGEMENT COMMITTEE

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Management Committee Meeting held on 19-20 October 2007.

Governance

The Trustees confirmed that there were no changes to the Risk Register. The Trustees agreed the new Data Protection Policy unanimously.

Research

The Chief Executive updated Trustees on the projects being undertaken by student researchers over the Summer. The MPS II longevity study has been completed. Over 100 interviews for the MPS I Management Survey were undertaken and the data anonymised for analysis. The MPS I longevity study requires some more work and will be completed by Christmas. The Trustees considered a request for additional funding to maintain the MPS IIIA mouse colony vital to Dr David Begley's work on the Blood Brain Barrier. A grant of £9,000 was agreed.

Personnel

The Trustees were provided with the results of the indpendent salary review undertaken by freelance consultant, Harry Marsh, on behalf of the MPS Society's Trustees.

Advocacy Support

The CEO informed Trustees of a proposal for enhancing the individual advocacy service. It was agreed the Senior Advocacy Officer carry out a consultation with the advocacy team and feedback to Trustees at their December Trustee meeting.

Having achieved four grants over three years to support a palliative care advocacy officer and a PND advocacy officer, plans were agreed to recruit.

Clinical Management and Treatment

The CEO updated the Board on the progress of the Health Technology Assessment study being undertaken by Peninsula University. Judy Holroyd tabled a summary of discussions from the All Wales Medicines Strategy Group (AWMSG) on the appraisal of Elaprase for use within NHS Wales and were advised that the decision of the panel was that Elaprase should not be recommended for use within NHS Wales. The CEO advised that Enzyme Replacement Therapy for MPS II had been agreed for a boy in Glasgow but denied for a boy in Aberdeen. The Trustees were advised that the MPS Society is supporting the family to appeal. Trustees agreed that the aim is for Scotland and Wales to adopt a national reimbursement policy. The CEO advised that MPS had facilitated a joint meeting at MPS House with Genzyme, BioMarin and Shire with this objective in mind.

Overseas Collaboration

The Trustees agreed the Chief Executive's work plan.

International MPS Symposium, 26 -29 June

This Symposium is taking place in Vancouver, Canada. Trustees were advised that an expression of interest form was sent out with the MPS Magazine. The Board agreed that the Society take a small group of families and MPS adults supported by volunteers to the Symposium. It was agreed the CEO undertake risk assessments and make provisional reservation during her forthcoming visit to the USA. Trustees acknowledged that without the funds received from the Ollie G Shoot held in September 2007 it would not have been possible to give even a few families and MPS adults this opportunity.

You are important to us, please keep in touch.

Please remember to let the Society know if you are moving and your new address and telephone number. In addition to helping keep the printing costs down, you will help us keep our database up to date. Keep us informed of new addresses, telephone numbers, email addresses and any interesting news about yourself, your child or your family.

UPDATING YOUR INFORMATION WITH US

We have recently sent out a questionnaire to each household asking our members to update their information. It is really important for us and you that you complete this form and return it to us!

If you require us to send you another questionnaire because the first one has gone missing or for some reason you have not seen the questionnaire, please do contact us at the office so we can send you a new one. It is not too late to send it in!

Contact us now on 0845 389 9901

ANNOUNCEMENTS

New Members

Mrs Nicola Carnall has recently been in contact with the Society. Nicola and her son Michael both have a diagnosis of Fabry Disease. The family live in the South East.

Mr and Mrs Astle have been in contact with the Society. Their son Mikko has been diagnosed with Hurler Disease. Mikko is seven months old. The family live in Derbyshire.

Miss Moran has recently been in contact with the Society. Mackenzie has a diagnosis of Hunter Disease. Mackenzie is four years old. The family live in the East Midlands.

Mr Simner has recently been in contact with the Society. Mr Simner has a diagnosis of Fabry Disease. He and his family live in Wales.

Mr and Mrs Hawkins have recently been in contact with the Society. Aisling has a diagnosis of Sanfilippo Disease. Aisling is 6 years old. The family live in Northern Ireland.

Deaths

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

Luke Morrison who suffered from ML II and who died on 17 October 2007 aged 3 years.

Derek Evans who suffered from Fabry Disease and who died on 24 October 2007 aged 59 years.

Rachel Harrison who suffered from Hurler Disease and who died on 21 November 2007 aged 5 years.

Samantha Preece who suffered from Sanfilippo Disease and who died on 9 December 2007 aged 10 years.

If you would like help, guidance or information from the MPS Society's advocacy team please do call us on 0845 389 9901

Congratulations to **Sam Wheeler**, who has Morquio disease (MPS IVA) and is aged 16 years. Sam got all his GCSEs this summer and achieved two A grades, 6 B grades and 3 C grades. His family are really proud of him.

One of the A grades was in PE which is really excellent. Sam has now started sixth form and is hoping to study for four A levels.

Congratulations to **Caroline Harding**, Chief Executive of Jeans for Genes, and her husband on the safe arrival of their baby daughter.

Augusta Merle Violet was born on 12 November 2007 weighing 8lbs and is a new sister to Columbus.

The Journey

Life can change at any time,
Our path leads one way,
But can be jolted to another

Sometimes wishing we could erase with a rubber
Things that you thought were important are not!
Other priorities rise to the top

Having a happy child is one
Whether it be your daughter or be it a son.
We as parents must stay mighty strong!

We never thought in a million years
News that would fall upon our ears
I could no longer hear what the Doctor had to say
Our life changed on that very day!

Our son has a Rare Genetic Disease
Maroteaux-Lamy is its name
My husband and I carry a Gene that is exactly the same
But we know we are not to blame

The cloudiness in the cornea gave Doctors the clue
Blood tests and investigations were followed through
So what the Doctors said was actually true!

My heart was very heavy and quite bruised
This really can't be the news?
Work I couldn't do for a while
I couldn't even raise a smile.

The MPS Society were straight on the phone
We knew we didn't have to go this alone
They even offered to visit us at home!

The Manchester team are never far away
Without their help what can I say?
They bought the news upon a day
That the treatment Thomas needed was
Finally on its way, hey, hey, hey!

The sun shone on the new corner we would take
This would be the big found break
Enzyme treatment was drawing near
This was good news we were glad to hear
I let out an all mighty big cheer!

I can remember contacting all those near and dear
But although we would probably have to wait another year
It took away the dread, and the fear.

In May 2006 there was a very special Day
We must drive down to Manchester without delay
The infusion was now waiting for us
Hurry, quick, quick, travel we must
Good job we were not relying on a bus!

As the procedure began
There wasn't a dry eye across the land
It felt like Christmas, Birthdays, and all
I could touch the sky I felt so tall.

Last Christmas Tom's treatment came home
The Healthcare Team rang on the phone
We were all excited like a dog with a bone
Thomas does not have to go this alone.

He shows great strength in what he goes through
This treatment is helping him, this much is true
To remain positive we must always do
That goes to all of you too!

By Kim Coney, mother of Thomas (MPS VI)

MPS Awareness Day
15 May 2008

To find out ways in which you can help email **fundraising@mpssociety.co.uk** or **phone 0845 389 9901**

Your news and views

We are always pleased to receive news, information, letters, stories and poems from all our readers, especially our members.
We welcome letters on any subject and your views and comments would be very welcome or perhaps you would like to share some information? Email us at **newsletter@mpssociety.co.uk**

Celebrating Jeans for Genes Day

5 October 2007

www.jeansforgenes.com

Jeans were worn by teachers and pupils in every part of the UK last Friday to support a charity which helps people with genetic disorders.

Children at Hazlemere CoE Combined School spent Jeans for Genes Day learning about the cause and contributed £1 each, aiming to raise £200 in total.

The annual campaign urges thousands of school children to make a difference to the lives of children with genetic disorders simply by wearing their jeans and donating £1.

This year the charity's aim is to raise £3 million to help seven national charities, including Great Ormond Street Hospital, the National Deaf Children's Society and the Sickle Cell Society.

Issy Baxter, 10, a pupil at the school in Amersham Road, Hazlemere, said: 'In assembly today we were told how all of us have genes and that some people have things wrong with their genes and it affects the way they live and sometimes it can affect their brain and their muscles.'

'I liked today because we got to wear jeans and it also means that we have helped to make it easier for people who have genetic problems to live a normal life. It is unfair if they get treated differently.'

Chloe Horst, 10, said that she learnt about people who have walking disabilities and special needs owing to genetic complications which is why the money is being raised.

She said: 'I want to help them so that they will not be treated differently and it makes me happy inside to know that I am helping somebody.'

'It's good that when I come home I have a smile on my face because I know that I have helped somebody.'

Tommy Dixon, 10, said 'I like the fact that you get to wear jeans and help a charity at the same time, and it was fun seeing the teachers in jeans. It was weird because they are usually really smart.'

Headteacher Nick Waldron said 'We are a church school so we have been talking about the way God has made all of us special and that our genetic make up is part of that.'

He said teachers had explained to students how some children have genes which makes life more difficult and that this event is to raise money to help them.

Mr Waldron added: 'I think it is important that we are good citizens and I think it is important to understand that despite our differences we have similarities and that we all share a sense of common humanity.'

'And I think this is a very practical and fun way of showing children that they can help others and enjoy it as well.'

This article written by Sarika Sharma is reproduced courtesy of the Buckinghamshire Advertiser www.buckinghamshireadvertiser.co.uk

Photo left: The MPS team celebrating Jeans for Genes Day at MPS House

Events Calendar 2008

Friday 25 January	Royal Manchester Children's Hospital BMT Clinic
Sunday 17 February	Dartford Family Get Together
Friday 22 February	MPS Clinic at Birmingham Children's Hospital
Saturday 3 – Sunday 4 May	Alton Towers Family Weekend & AGM
Thursday 15 May	Great Ormond Street MPS III Clinic
Thursday 15 May	MPS Awareness Day
Thursday 29 – Friday 30 May	Northern Ireland Conference & MPS Clinic
Friday 6 June	MPS Clinic at Birmingham Children's Hospital
Friday 13 June	Scottish Conference
Saturday 14 June	Ollie G Ball
Tuesday 24 – Monday 31 June	10th International Symposium Vancouver, Canada
Sunday 13 July	Childhood Wood Remembrance Day
Friday 25 – Monday 28 July	Sibling Weekend, Longleat, Wiltshire
Friday 29 – Saturday 30 August	MPS IVA Conference, Hilton Northampton
Friday 3 October	Jeans for Genes Day
Friday 24 October	Childhood Wood Planting Day
Friday 21 November	MPS Clinic at Birmingham Children's Hospital
Saturday 29 – Sunday 30 Nov	MPS Adult Weekend

Bristol MPS Clinic

10 July 2007

The third Bristol clinic was again held at the Children's Hospital in Bristol. Despite difficulties parking and a hectic waiting area the clinic was quite successful. We have taken on board the comments regarding the venue and the hunt is on for a more suitable space to accommodate us.

I would like to say thank you to all those who attended for their patience and to the doctors and hospital staff for supporting this clinic.
Steve Cotterell

Bristol MPS Clinic

2 October 2007

The fourth Bristol clinic was for the first time held at Tyndall's Park Children's Centre. This centre provided an ideal environment for the clinic, having a kitchen to make tea and coffee, a smaller waiting area for our members with toys and also private space should this be required.

The clinic ran for half a day and went very well and we are hoping that clinics will be held here in the future; dates for 2008 are yet to be confirmed. We would like to pass our thanks to the team at the Children's Centre and to Dr Wraith and Dr Jardine for making the clinic possible. Should you wish to have an appointment at the next clinic please contact me at the MPS office.
Steve Cotterell

BMT Clinic

19 October 2007

The BMT clinic at Royal Manchester Children's Hospital on this occasion was split over two days in order to allow families more time with the medical team. I attended the second day on 19 October. We had a full day scheduled and fortunately we ran pretty much to time. Jean had been kind enough to provide refreshments and soon all the toys were scattered on the floor for the children to enjoy.

It was great to meet the families there and our thanks go to all the staff at the Willink for supporting this clinic.
Steve Cotterell

Photos clockwise from top right: Cardiff Clinic - Christopher and Steven Jones (MPS III); Northern Ireland Clinic - Aaryannah Lever (MPS III); BMT Clinic - Metabolic Team and Nandha Kishore (MPS I BMT)

N. Ireland Clinic

22 November 2007

The second Northern Ireland clinic of the year was held at the Antrim Area Hospital. As usual Dr Fiona Stewart provided us all with an abundance of tea, coffee, cakes and biscuits which were all gratefully received by the families that attended. We had a very busy day but all went well and families had a good opportunity to meet and catch up. We would like to pass our thanks to Dr Fiona Stewart, Dr Ed Wraith and to all the staff at the hospital without whom we could not hold these clinics. **Steve Cotterell**

Birmingham Clinic

23 November 2007

This MPS Clinic was held at The Birmingham Children's Hospital. The day was full and Dr Hendriksz, Dr Chakrapani and their staff team had their work cut out with the very busy clinic. Despite the busy day, the clinic ran very smoothly, and it was very useful for me to have been kindly allocated a private room to speak to families about any issues they had. I would like to take this opportunity to thank Dr Hendriksz, Dr Chakrapani, Catherine, Louise and Sat for all the support and for making my attendance at the clinic a pleasurable one. **Neisha Hall**

Cardiff Clinic

30 November 2007

The Cardiff clinic took place at the University Hospital of Wales. It was another busy clinic for all concerned and we would like to thank all those families who had to wait for appointments for their patience. We felt that the day was very successful, with some families re-making contact and others taking the chance to meet new people. Our thanks go to Dr Graham Shortland and Dr Ed Wraith for supporting this clinic and to all the staff at the hospital who help to organise this day. **Steve Cotterell**

Photos clockwise from top right: Birmingham Clinic - Thomas Kynaston (MPS VI), Mohammed Yousef (MPS IV); Cardiff Clinic - Sarah McKnight (MPS I); Northern Ireland Clinic - William Todd (MPS I HS)

Childhood Wood Planting 2007

We couldn't have wished for a more perfect day. The sun was shining and the rain stayed away - Barry, Wilma and I certainly breathed a sigh of relief. We all congregated at the Clumber Park Hotel and took a few minutes to greet the families who were attending the day and the dignitaries who had very kindly agreed to attend this special event.

After a lovely lunch and some calorific desserts we set off in convoy to Sherwood Pines and made our way to the Childhood Wood. After a lovely opening speech from MP Paddy Tipping, a touching speech from Barry Wilson and the poem 'Remember' beautifully read by Wilma Robins, the families proceeded to plant the tree for their loved one.

I must say that the releasing of the balloons was a very special moment, and we all took some personal time to remember our loved ones and watch the balloons drift upwards.

Following the planting everyone had the opportunity to have some time for themselves and to walk through the wood or to return to their cars and make their way home. I would like to personally thank a few people: Byron and Becky for all their hard work at the Childhood Wood, Paddy Tipping MP for his attendance at the planting, Jenny Mellors and Mr John Allin. I would also like to thank Barry Wilson and Wilma Robins who continue to support this important day. **Neisha Hall**

Remembering William

We were pleased to join the group of families at Sherwood Pines planting the oak saplings to remember their special children. The event had been a positive focus for us coming at the end of our first year without our son, William, who died on 26 September 2006, aged 17 years, with Sanfilippo. What a lovely place to come to remember William and all the other children.

The woodland setting reminded us of the walks we enjoyed in Germany where William had lived for most of his life. William had enjoyed running through the Grünewald forest in Berlin and over the Lüneberger Heide, north of Hanover. As a young boy he ran very quickly and we often had to chase after him. As he got older he loved strolling hand in hand through the countryside. It was lovely to remember these happy memories as we all made our way to the Childhood Wood.

William's older sister, Lindsay, wrote a moving inscription for the Memory Board and we are very proud of her. She has learned a lot from being William's sister and has just started her second year training to be a Learning Disability Nurse at Southampton University.

We would like to thank the MPS Society for giving us such a peaceful and beautiful place to visit and a special place to remember William. The photo on the left is of William and his sister, Lindsay, walking through the woods in Germany. **Bill and Caroline Ferrier**

Childhood Wood Remembrance and Planting Days 2008

The Childhood Wood is now 15 years old and some of our members will remember planting those first trees in memory of their loved ones when the Childhood Wood was a woodland clearing back in 1993. With the growth of the Childhood Wood there have been developments and it is now a thriving woodland habitat with some new additions such as the woodland animals hidden in amongst the trees and along the path that surrounds the Childhood Wood. The Memory Boards displaying short memorial inscriptions of the children and adults names that have passed away are a focal point in the Childhood wood and much read by passers by.

Remembrance Days and Planting Days are held annually and are an important time for families to gather together. If you would like to take part in the Remembrance Day on Sunday 13 July 2008 to remember a family member that has passed away from an MPS or related disease please do complete the booking form enclosed with the MPS Magazine.

An annual planting day is arranged each year for families that wish to plant a tree in the wood for those that have passed away and this is the time that we update the memory boards in the wood.

If you and your family are unable to attend the Planting Day you can still be a part of it by putting an inscription on the memory board which you can visit at your own leisure and with your permission we can plant a tree on your behalf.

There are forms that we can send out to you regarding the memory board which you can fill in with a personal message to add to the board.

Invitations are usually sent out closer to the event to those that in the past year have recently lost a loved one, although of course this is not exclusive and if someone has lost a loved one many years previous they are very welcome to contact us and be a part of the day as well.

If you have any questions or queries regarding the Childhood Wood Planting taking place next year on Friday 24 October 2008 please contact the office and speak to one of the advocacy team or Miriam Blowers the Events & Volunteer Coordinator.

Childhood Wood Planting 2007

London Family Day

It was an early start to catch the tube to London, the sun was shining although a little cloudy. As I queued for the tickets for the London Aquarium the families were arriving one by one, the MPS mobile was ringing once, twice, again and again, but everyone arrived in the end.
One by one the families disappeared into the darkness of the aquarium, only to be seen every now and again around a corner.

When the morning had gone it was time for our flight on the London Eye, the cloud was gone and the sun shone bright. We queued for a while here and there and then there again. The staff were great, we all got on in the end. The sights were amazing, a minor security hitch with a pair of scissors was soon explained and we were on our way. We were half way round and we could hear this voice. "Is there an emergency?" "Is everyone alright?". Oliver Robinson (MPS III) had pressed the emergency button, how cute. It was soon over and everybody said good bye until the next time.

I hope everyone had a great time, a huge thank you to 'Help a London Child' for sponsoring this event. The photos below show everyone enjoying themselves.

Yet another wonderful day out with the MPS Society. These days out are like a ray of sunshine in a cloudy day for me. It's fab to see old friends and make new ones. We are only sorry we didn't meet up with you guys at Pizza Express, we could have spent more time with you all. Thanks once again Gina for you and your family's patience waiting for us to arrive, and for giving up your Saturday. Can't wait for the next trip. I wonder if we'll be invited after Oliver's "little trick". We "Robinson's" always end up doing something out of the ordinary, only hope the staff can keep coping with our "special events?" We seem to have left our mark on each trip so far.
Karen, Stu, Oli (MPS III) and Sam Robinson

Many thanks for another fantastic MPS day out at London Aquarium. We arrived to the South Bank, where the children were delighted by the many street entertainers. Friends greeted us at the Aquarium and we had a raucous time devastating the calm and quiet of the many beautiful tanks! After stroking the rays and fish in the touch pool, we headed to Pizza Express for a delicious lunch. The London Eye was wonderful, made all the more exciting by our brief stop after someone pressed the emergency alarm in our carriage! A fun day was had by all and we are as ever grateful for your organisation and care and the time you and your family gave up to look after us all so well on the day.
Jessica, Tim, Jamie (MPS III), Emma & Megan Hooper

The Aquarium

The Aquarium was the place to be
The fish swimming round so gracefully
They came to the glass and pouted their lips
Looked like they were expecting a great big kiss!

We met up with a family we hadn't seen for a while
Their son had such a fantastic smile
It lit up the room which was dark inside
The mother's love she did not hide.

Their daughter took a liking to Tom
She kept saying come on Tom, come on
Walk with me we must move on!

The Stingray bopped to the top of the water
You could touch if you dared
All the kids touched it and were not scared.

The trip was good that's for sure
All the fish we did adore
We will come back and visit
That is for sure!

The London Eye

We came to London to see the Eye
The views were fantastic
And we went so high
There wasn't a single cloud in the sky

We climbed on board and took a seat
This was such a pleasant treat!
Taking photos of all around
This was better than being on the ground.

Thomas suddenly let out a cheer
Look I can see Wembley Stadium right from here!
"It seems that it is so near".

Chantelle stood up with me
Gazing around silently
Oh, it was so lovely!

To the left was the Tower of Big Ben
I said to Neil, we must do this again
As we have now come to the end.

The wheel stopped and we got out
It's been a really good day
Without a doubt!

Poems by Kim Coney

Day out in London

On a recent Saturday, we got up early and prepared for a trip into London for a day out at the London Aquarium and a spin on the London Eye.

Having decided to drive rather than take the tube into London, it was a pleasant surprise to find the only free parking space outside the London Eye. Such good fortune also resulted in us turning up on time!

We collected our tickets from Gina for the London Aquarium and met some friends on the way in, so we took the opportunity for a brief stop to take on coffee and cake before venturing into the depths!

The Aquarium was amazing, I can honestly say it was not what I was expecting in the basement of County Hall. The fish ranged from the almost invisible to mighty sharks and there was even an opportunity to touch some rays, quite a strange sensation if you were lucky enough to be chosen by the rays! Both William and Sophie (MPS III) enjoyed the experience, all areas were accessible by wheelchair but this did lead to us following our own route and not the order advised but I don't think we really suffered.

After the Aquarium it was lunchtime and we met up with two other families at Pizza Express, I don't think the restaurant was ready for three MPS children, but they took it all in their stride - eventually! The meal passed off very well with only one broken glass, one near escape through the fire exit and one bemused waiter. It is amazing how all these things become entertaining when you are not the only one trying to explain the situation. Oh for safety in numbers!

We walked back along the South Bank and watched the street entertainers, they ranged from buskers with doubtful musical talents to living statues which were incredible, how anybody can wear such costumes and remain so still is beyond belief.

We were slightly anxious about going on the Eye with Sophie, once the doors close you have thirty minutes to wait until you can get off again. That would be a long time if Sophie didn't like the sensation. We passed the security checks with ease, luckily not having any sharp instruments or anything else deemed to be dangerous.

When we arrived at the front of the queue they stopped the Eye for us to get Sophie into the capsule, then it was clunk as the door closed and we waited for Sophie's reaction - Phew, she was sitting quietly in her chair looking around quite happily. We spent the next half hour picking out the sights of London, from the Wembley arch where England were winning, to St Paul's Cathedral and Canary Wharf. It was an amazing view from the top, where we paused for several agonising minutes, before we started the descent.

Once back on terra firma we returned to find the car and beat the football traffic up the M1. We all agreed it had been a wonderful day out, a great experience and wonderful to catch up with other families and share stories.

Thank you to everyone who arranged the day out and a special thank you to Gina and her family for being there for us all. **Tim Summerton**

Our visit to London

We looked at each other at the side of Jack's hospital bed and said 'let's do it, let's go'. We had had eight months in hospital by this stage with Jack and were due to hopefully have a full and final discharge in June provided everything was ok with the agency that would be coming in to give us the 24 hour care package we would need since Jack's surgery the October before.

I left the hospital and telephoned directory enquiries for the disabled helpline for Virgin trains. As I waited to be connected to an advisor, all sorts of thoughts were running through my mind as to how we would accomplish this mammoth task of getting Jack, our 8 year old with Hunter's and a new tracheostomy and all the equipment that on its own entailed, our three year old daughter Katie and the luggage we would need for ourselves on a train to London and then from the station to an hotel! Would they allow oxygen on the train? In the hotel? Or would the new health and safety laws not allow it?

'I need wheelchair access from Manchester Piccadilly to London Euston please and one toddler and four adult seats.' I prayed as I waited for a response that my Mum and Mother in Law would come with us as I had not even asked them!

'I'm sorry Madam there is no availability for those journeys until the end of July!' 'But how can I book a hotel to make sure that is ok if I can't book the train?' I replied.

'Don't worry Madam, as long as you book in the last week in July as the tickets are released it shouldn't be a problem.'

I began to ask all the questions in my mind – oxygen – not a problem, wheelchair – we take those all the time, don't worry we'll meet you with a buggy in the taxi rank to take all your luggage and there will be another buggy waiting at the carriage door in Euston to take you to the taxi rank. Fantastic, I can push Jack, Jason can drag the sewing machine bag we had purchased to hold the spare suction machine, the emergency trachy box, ambibag, nappies and wipes that we would need on the journey and the two Grandmas were on hand to get help in an emergency and ensure that one very independent three year old was safe and sound.

That evening I spoke to my Mum and Mother in Law and they agreed to go on the trip, little did they know how much we would depend on them.

As we were due to depart on Friday 12, Jack was poorly on the Monday, had his infusion of ERT of which he reacted badly on the Tuesday and had to attend on the Thursday at RMCH for a dental appointment, so getting everything organised was quite difficult.

The disabled mini bus arrived ten minutes late on Friday lunchtime and we loaded up our large suitcase, large holdall, and emergency trachy case (we looked like we were going on holiday for a month with everything we were taking!) and then set off down the road to collect the two Grandmas and their luggage. We arrived at the station one and a half hours early too. I went to the desk as per my instructions to get the buggy to meet the taxi and spoke to a Virgin member of staff to just set my mind at rest that in the event of an emergency we would contact the driver of our train to stop and contact the paramedics. He went to consult with his manager and came back with a gentleman who said he would put us on a faster train, he would upgrade our extra-value tickets to first class, and we were literally installed on the train within 15 minutes and on our way within 20. They were so kind and couldn't do enough for us, we had just finished our meagre picnic when they came through to take our food order and offer us complimentary drinks – it was amazing and even though we declared that we were not really travelling first class they said it didn't matter and to just enjoy the journey and if we needed anything to just ask.

We had an enjoyable and uneventful journey down with the only problem being that Jack performed in his nappy twice. We had taken some paper roll so managed to manoeuvre the wheelchair into a position where we could hide what we were doing from the rest of the carriage of travellers, so at least it wasn't too embarrassing for Jack.

The train entered Euston two hours before we were originally due to arrive, but the buggy and ramp were waiting for us and took us through the underground passages to the taxi rank where I recruited two unsuspecting drivers to load Jack into one, and the luggage into the other and take us to the Premier Inn at County Hall which would be our home for the next three nights. As we unloaded Jack at the front door a young gentleman came and pushed him away and we shouted him back as we were still in the process of paying for the taxi. I don't think he had realised what having a child with a tracheostomy entailed in that there had to be always two of us with him, one resus and trachy trained and one trained in suctioning and that you always had to keep the emergency box with you. As we entered the hotel through the basement entrance, as there is no ramp up to the front door, I thought what have we done, they said they were disabled friendly but we have to go up in through the basement! We were allocated our rooms and unpacked before dinner and as we set up Jack's sats monitor and suction chargers we realised that there were no electricity sockets close enough to his bed! Could things get any worse? We rang down to reception and within five minutes, we had the maintenance department's extension lead set up in our room. We went downstairs for our first evening's dinner and the pureed diet we had ordered came and nearly choked Jack to death as it was that lumpy. When I asked if they could liquidise it again I was told they didn't have a liquidiser and were just using

a hand blender. (I wouldn't have minded if they had told me upon booking but they had said it wasn't a problem, but I could have taken one with me!).

The first evening after dinner we went for a walk around the block to find the entrance into the London Aquarium where we would be meeting the other MPS Society members at 10am in the morning and then going on to the London Eye in the afternoon. We found that it would be quite accessible and went back to our rooms feeling like we had really accomplished something and that we would not have as much of a panic as we thought the next morning as it was only a five minute trip around the corner. Day one was over! We were safely installed in the hotel and we could not have been any closer for the early start the next day unless we had pitched a tent on the embankment!

We got through the night without the sats monitor alarming but I was up about 40 times suctioning and resettling him so I was very tired. The grandmas arrived as arranged with Katie all dressed and just waiting for her hair putting up and we descended for breakfast where we caused havoc as England were playing at Wembley that afternoon. As we finished, the manager came and asked if everything was ok and we expressed that we had struggled with the meal the night before and that we would need a larger table in future as we had blocked the main thorough fare. She advised that she would reserve the large table at the front for dinner and try a different method of pureeing that evening.

We set off and met Gina outside the aquarium as arranged and met a couple with their two children who we had been to America with the MPS Society (you know who you are!). It was lovely to see them again and see how well their son was doing (also an MPS sufferer) and we had a chat and updated them on what had happened with Jack since we last saw them when Katie was only four months old. We then entered the Aquarium and had a lovely visit – it was so calming that Jack slept half way round in his wheelchair! We went back to the hotel and had a very small lunch of cup a soup, and changed Jack ready for our afternoon visit to the Eye. We were all really excited and set off at 2.30pm ready to meet Gina again to take our places in the queue for our 3pm flight on the Eye.

As we approached the front of the queue a security man stopped us and asked what was in our very suspicious large-wheeled case, just the stuff for an emergency trachy change, nappies etc we responded. He then asked if there were any sharps, scissors etc, yes, we responded. It was then that I thought we were not going into the pod, he wanted me to leave the emergency trachy kit with him. As we tried to explain that we could not and that we may need it at any moment a sense of doom came over me and we had had such a lovely day so far. I shouted Gina and explained and she said that I did need the equipment to the guard and managed to make it so that we could go on our flight.

Up in the pod my heart calmed eventually and we had a fantastic view of the city on a very clear day, the highlight for me was seeing horse guards parade with Buckingham Palace in the background, and Jack grinning like a Cheshire cat at so much to see. The highlight for Jason was seeing the MI6 building where they had filmed James Bond 007 in the World Is Not Enough. Katie just thought it was great to be so high in the sky.

Thank you Gina and everyone at the MPS Society for making a lovely day out come true for us.

When we reached the ground, we decided to pay a visit to a very famous store and caught two very expensive black taxis to get there. I don't think Harrods new what had hit them when we arrived (large suspicious bag in hand again!). No fewer than five security guards accosted me and asked what we had in the case and how had we managed to smuggle it into the store without being stopped – we certainly educated them as to what is required for a child with a tracheostomy! Although it was very embarrassing – I was as red as a beetroot! As we left the store a couple of hours later with our £1.95 purchase of an hair bobble for Katie in the token Harrods bag, we joined the queue for the taxi and returned through the basement to the hotel and went to dinner again. This time Jack's puree resembled soup with things floating in it and we had to request a sieve and thicken it up. We decided at this point to purchase some baby food the day after as we were starving the poor child. As we arranged to meet in our room the next morning at 8.30am to change Jack's trachy, we all went to bed exhausted and nervous of performing our first change out of hospital or in the familiar environment of Jack's room – a first for us all! We had lived through day two and had a tale to tell everyone!

Jack had another rough night and ended up in bed with me, and Jason slept in Jack's bed! We bathed him and just had his nappy on when reinforcements arrived and the change proceeded without a hitch. We went down for breakfast and planned our outing. The most expense we had had been with taxis and we had to have two every time, so we decided to try a bus today and we enquired at

reception and they gave us a rough plan as to what the routes were and the destinations. We set off en masse at 12 noon and walked over Westminster Bridge and proceeded towards St James Park where we would pick up the bottom end of Regents Street. However, when we got there it was a flight of about thirty steps! They weren't on my map. Luckily, there were two policemen sat on two motorbikes at the bottom of the steps and I went and asked them how we could get where we wanted to go without going up the steps. A half an hour's walk either way came the response, so I very cheekily asked if I carried Jack up the steps would they help carry the wheelchair with Jason. One of the officers kindly agreed so off we went up the steps child in arms, wheelchair carried by a policeman and Jason, and the grandmas with emergency kit and one Katie (who also requested carrying at this point and was told in no uncertain terms no, you're a big girl now even though she had been walking well over an hour already!). What a sight!

As we got going up Regent Street, we looked for some shoes for me as my heel had fallen off just before setting off! How much bad luck could a girl take? Any excuse to buy a pair of shoes on the London high street! Moreover, I managed to buy some quickly, we then went to Hamleys toy store where we managed to buy a toy for Jack's entertainment for Christmas. We decided we would like a brew and went into the Mamas and Papas shop where we bought Jack some baby food and fed him so that he could have his lunchtime medications and then proceeded up Oxford Road in search of the Disney store. I spent a fortune on Jack buying some pyjamas and a dressing gown and Katie a lovely red pinafore for Christmas day. We ventured a little further up the road where Jason also got some shoes and then found that the shops were starting to close and that the time had got to 6pm. We dug out the bus route, found a stop and waited hoping that the bus would be an accessible one.

Some hope! The ticket machine swizzed us for £2 it didn't issue and the bus pulled into the stop. When we said to the driver that we had a wheelchair to get on he gave us a filthy look and did not attempt to get any nearer the kerb as he was about 3 and a half feet away. A young man at the stop offered to help bridge the gap and lift the chair on board with me (I think he is probably still sat in a hospital waiting room with a hernia!) and nearly dropped it and we were on our way, blocking the main passage and causing a riot. Eventually three men got off and vacated the wheelchair space and I managed to sit with Jack and another lady, who started asking me questions about Jack. The whole bus was silent as they learnt all about MPS II, a tracheostomy, and our interesting trip to London. The others in my party had their own tales to tell once we disembarked after sending a message to the driver that a wheelchair was getting off in a similar manner to the final scene in Crocodile Dundee in Grand Central Station!

Laugh, we could not stop, Katie had had the front of the bus in an uproar with 'my eyes are knackered and my feet are knackered Grandma', talk about out of the mouths of babes! Jason had had a young Chinese man touching him! (I will leave that to your imaginations). In addition, Grandma Joan had had an interesting conversation. We went to a small restaurant across the road that evening and had everyone there in stitches as well as we regaled our tale to a couple who had asked us about Jack and we even had Americans taking pictures of our two beautiful children.

Time to pack, we leave in the morning, so we got the children's clothes out ready for the journey and started the mammoth task of repacking all Jack's equipment and other purchases. Jack slept a little better the last night and we arose at 7am to get out for breakfast at 8.30am then we could take in a bit of fresh air before we left. We booked two taxis and gave up one of the rooms and nipped around to the little park adjacent to the Eye to keep Katie happy. Jack watched with interest as she sprung backwards and forwards and eventually decided it was quite funny and was sniggering (oh to hear the giggles again). We returned to the hotel and waited for the luggage trolley to arrive and went outside only to find that the two taxis they had ordered were private hire and would not fit a wheelchair in, let alone our luggage! We stood there for a couple of minutes wondering what to do, as there was a taxi rank adjacent to us but it was empty! Just as we had decided to send one of us back inside to phone again, a black cab rolled in so we asked him to radio for another one, as he said he could not, another one approached so Grandma Lynn flagged him down and we commenced loading everything and everyone in and arrived at Euston 45 minutes later. We found the special assistance desk and were put on a train earlier than the one we had booked (unfortunately not first class this time!) only to be usurped from the seats by people booked into them! I searched for a member of the Virgin team and caught one with three minutes to spare and he said he would have put us in first class also if there hadn't been so much to relocate, so he just very kindly explained to the people pre-booked what had happened and rearranged them accordingly.

After an uneventful journey home to Piccadilly, the assistance team greeted us and escorted us down to our mini bus and we arrived safely home.

For anyone who has a child with MPS it is daunting enough to venture far from home to start with but with the tracheostomy as well it is a mammoth undertaking. Be organised and do not be afraid to ask for help and tell them exactly what you need. Our experience with Virgin was amazing and I would definitely use their Journeycare service again (Tel: 08457 443366) and Premier Travel Inn could not have been more helpful even though we struggled with Jack's special diet.

When Jack first had the tracheostomy inserted we as a family were devastated. However, as the months have passed we are more capable of dealing with it than some of the professionals we have met. It isn't the end of the world and hope is now on the horizon that he can have it removed in December as the ERT treatment has removed all the deposits of the disease from his lower airway. I hope our little story can help another family who is going throught what we have gone through in the last twelve months. Be brave, insistent and live life to as full a capacity as you can for our special children – they deserve it. **Elizabeth Heath**

Changes to the MPS advocacy service

The Advocacy Support Team is changing how it currently supports individuals and families. At present the advocacy work is split by the four countries of the United Kingdom and the counties of England with each advocacy officer being in charge of a certain area. However, after a number of consultations it was agreed that given the increase in membership and specific needs and requests for support, it would be better for advocacy workers to become specialists in a specific area. It was agreed that this area should be disease specific. Other controlling factors were that it has become increasingly more difficult to secure funding for generic workers and funding that has come in has been very prescriptive in what area of work it is to be used for. We have also taken into consideration that other support groups / medical teams have named persons for certain groups of diseases.

Our initial proposal was put forward to the trustees who agreed it at their recent trustee meeting.

Current membership comprises the following diseases: MPS I, MPS II, MPS III, MPS IV, MPS VI, ML II, ML III, Mannosidosis, Fucosidosis, AGU, Gangliosidosis, Multiple Sulphatase Deficiency (MSD), Winchester, Sly, Geleo Physic Dysplasia, Sialic Acid Disease and Fabry Disease.

Current Team Set Up

Currently we have four advocacy workers and each worker supports a different area of the country. We are also hoping to recruit another worker in the New Year.

Sophie Thomas
North West and Midlands

Chris Murphy
South East, London, East Anglia and North East

Neisha Hall
Scotland and Northern borders, Home Counties

Steve Cotterell
South West, Wales and Northern Ireland.

Proposal for Change

Listed below are the proposed splits and which worker will be supporting each disease group.

Chris Murphy and the new worker will be supporting the following disease groups: Bereaved families, MPS III, severe MPS II, MPS I H non BMT, ML II, MSD, Sly, Gangliosidosis and Sialic Acid Disease

Sophie Thomas with be supporting the following disease groups: AGU, Fucosidosis, Mannosidosis, Winchester and Geleo Physic Dysplasia

Neisha Hall will be supporting the following groups: Fabry, MPS I and MPS VI

Steve Cotterell will be supporting the following disease groups: MPS II, MPS IV and ML III

How will this be implemented?

The Advocacy Support Team has already started looking at their current workload and are planning to handover pieces of work in the New Year. We are mindful that some pieces of work are ongoing or that there are families who may not welcome a change in worker immediately. With these individuals and families a more detailed and timely handover may be required and this may not happen immediately.

We appreciate that there may be a few teething problems in the initial few months and this change over is going to take some time but we hope that it will be welcomed by our members who should feel secure in the knowledge that the worker supporting them will become much more specialised in their field of work, therefore providing a better quality and more informative service.

As a team we will continue to support each other and there will be ongoing training in all disease groups and workers will have opportunities to keep updated on any changes and to ensure that they all have a basic knowledge of every disease group. This is to ensure that during any absences, we can still provide an equitable service to all our members.

If you are currently receiving support from a particular Advocacy Support Officer, you should have received a letter or are due to be sent one, outlining the change of worker and the current support needs required. Anyone who has any concerns regarding the change in work patterns should contact the MPS office to discuss their concerns, so that we can have a look at how this can be managed.

We hope that our members will look on this change in a positive light and that the transition will be smooth with minimal effect on the high level of support we currently provide. **From all the MPS Advocacy Support Team**

Do you need support from the MPS Advocacy Team?

Please remember that should you wish to speak with a member of the advocacy team do not hesitate to pick up the phone or email if you find it easier. Please bear in mind that at the moment we are a small team covering the entire UK, however we will always return calls and respond to messages as quickly as possible. **advocacy@mpssociety.co.uk** or **0845 389 9901**

Ollie G Summer Shoot

The hugely anticipated Ollie G Summer Shoot went with a bang on Saturday 8 September and raised a more than impressive £65,000.00! With over 100 guests, there was an amazing atmosphere which reflected everyone's excitement for the day.

The Shoot was held to raise money in aid of MPS children and the MPS Society and the day was enormous fun with a range of 80 clay targets from specially built stands and a 100 target flush. Tom Garthwaite, who has Hunter Disease, from Bramley joined his father and guests on the 'Garthwaite Guns' team. Tom came 77th with a fantastic score of 25 overall which was an amazing result and earned him the award for best newcomer.

After the shoot came a hearty lunch washed down with some fabulous wine to help get the guests in the mood for fines and auctions. The main auction, presented by Toby Kilner raised a staggering amount; the main item being a shooting trip to Argentina for 10 guns raised an astronomical £17,000.00. Also during the afternoon, Toby auctioned off a ride in one of Country Wide's amphibious max cats for £300.00 for Tom who had a superb time going full speed around an undulating and thrilling course and taking a dip in the river!

A fantastic day was had by all and the event raised a breathtaking amount of money which will go towards helping those affected with MPS diseases and provide funding for families to attend an educational trip to Vancouver next year.

Ollie G Charity Events would like to say a very big thank you to everyone who took part in the day; the generosity of all the guests and donators was phenomenal so thank you to everybody for their kindness and support. **Ollie G Events Team**

The Ollie G Event Team is looking to consider 50 wishes between £250 and £1000. The individual child or young adult or the family representative should set out what the wish is, the difference it will make and how much it will cost and send this to the MPS Society no later than **28 February 2008**. These will then be considered and the decision will be made known prior to the Ollie G Ball on 14 June 2008.

If your wish is chosen, you will need to agree to providing a story and the wish being publicised in the MPS Magazine and on the Ollie G website. For further details please contact the MPS Society.

A bed for Hayleigh

We are Robert and Heather Reynolds and we would like to say a big thank you to the Ollie G Summer Shoot.

Our daughter Hayleigh needed an electric adjustable bed to help her mobility getting in and out of bed and also to help raise her legs slightly to avoid stiffness overnight and "it's great for reading my books" comfortably before she goes to sleep.

Before getting this bed Hayleigh always wanted someone to sleep with her in case she needed to get up through the night etc. Now she can get up herself, which is great for her independence and also great for us.

The money raised exceeded the value of the bed so helped Hayleigh to be able to choose which bedwear, pillows, sheets etc she wanted to go with it.

Here is a photograph of Hayleigh in her bed (with the back raised and the bottom slightly raised to read her book) before going to sleep.

Once again we would like to thank you very much for your kindness and generosity. It goes without saying that we all give our thanks too, to the MPS Society for all their help and support. Thank you very much.
Robert and Heather Reynolds

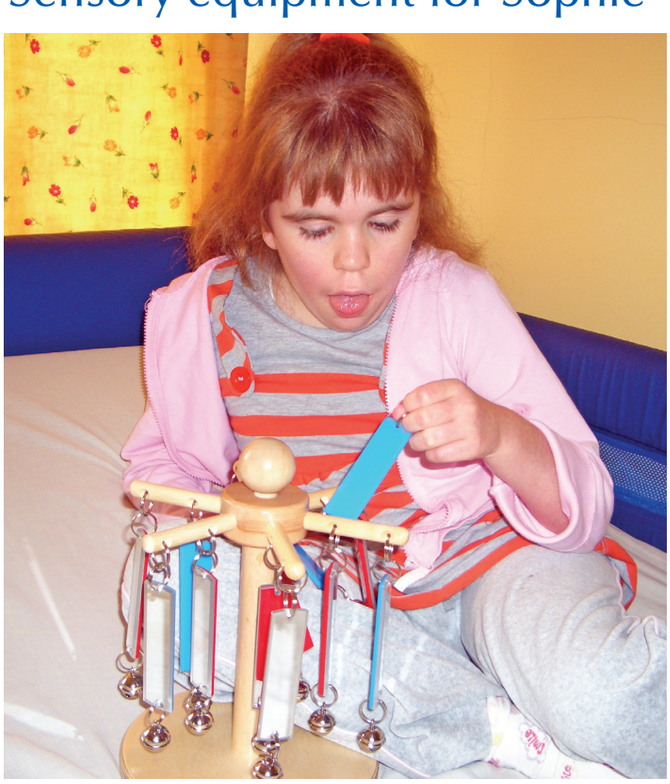
Sensory equipment for Sophie

Please will you pass on our sincere thanks to the organisers of the Ollie G Clay Pigeon Shoot and all the guests on the table who supported Sophie.

We were overwhelmed by the amount of money they so kindly raised. We have used the money donated to buy some items of sensory equipment for Sophie. As you can see in the photograph, she particularly enjoys the Chimeabout which spins, jingles and reflects light on its many mirrored surfaces. It is well suited to a Sanfilippo child as it is very robust and chew-proof! It does hurt, however, when it is launched across the room at you, again this is an occupational hazard and we are learning to dodge it!

We have also ordered a light projector which will allow Sophie to be entertained when she is lying in her bed. She will be able to relax and watch the changing patterns and colours on her ceiling.

The equipment you have enabled us to buy will stimulate and entertain Sophie, this will become even more valuable as her condition progresses.



Many thanks once again for your generosity.
The Summerton Family

Tom wins Best Newcomer Award

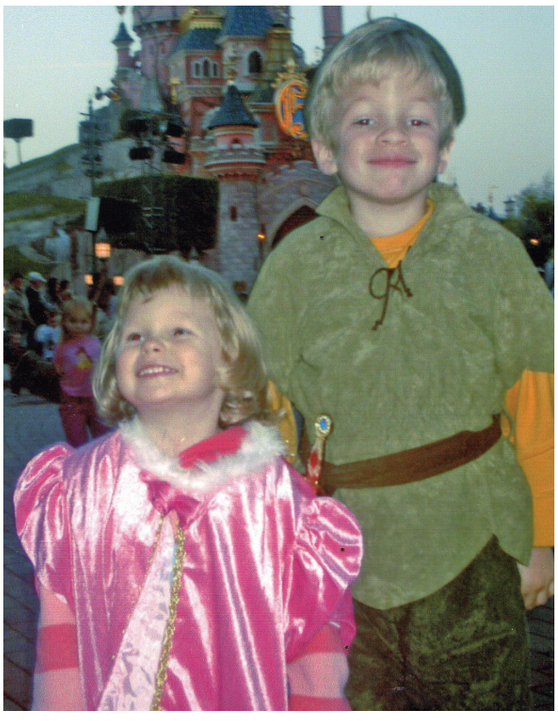
In August Dad said to me that David Gosling was organising a clay pigeon shoot to raise money for MPS, and that I was invited. I said that I really wanted to go, but I was a bit worried that I didn't know how to fire a shotgun. You can imagine how excited I was when he said that David had thought of that, and had therefore offered me the chance to have some private lessons before the charity day with the shooting coach at his activity centre, Countrywide Special Events (CWSE - what a cool business to have!).

I had two lessons with Mike, who was really a fantastic teacher - very patient and good at explaining things. Down in the woods at CWSE Mike showed me how to hold the gun, and how to shoot a clay pigeon. You have to follow the clay with the gun, and shoot as you overtake it. Sometimes he got me to shoot at an imaginary coke can on top of, or below the clay. It was an amazing feeling to hit the target and see the clay shatter. He gave me an oven glove to protect my shoulder from the 'kick' that the gun gives as it is fired. Whilst the gun was a bit big for me, I could quickly feel that this was a sport that I would really enjoy.

The big day finally arrived and I made up a team with Dad, my uncle and two friends. There were over 100 of us shooting that day and all of them had a really good time. There were also some famous people, including Roger Taylor (from Queen) and Gary Brooker. When all the teams had finished shooting, we had a fantastic lunch over which more money was raised for MPS. Dad gave a speech about what MPS was and how it affects families. Then I presented some prizes to the winning teams. I was really surprised and pleased to be presented with a cup by Christine Lavery for being the best newcomer! It was an experience that I won't forget - and I'm hoping that I can continue to learn to shoot. **Tom Garthwaite (MPS II)**

We would like to thank the MPS Society for contacting the Ollie G Events Team on our behalf, who have provided the funds needed for Alice's specialised bed.

The money so kindly raised by the Ollie G Events team and those who attended the Clay Pigeon Shoot will buy Alice a 'Kindersafe' specialised bed, which will make a big difference to Alice as she experiences sleeping problems. The bed is designed to ensure her safety and also be fun for her to sleep in. We would never have been able to purchase this item, and would like to give our sincere thanks to those who organised the event, and to those who so kindly gave to help Alice – we are astounded by such generosity and never imagined such an amount would be raised. **Karen, Clive and Alice Coombs**



A special thank you

We are writing to thank you for putting Harry's name forward to the Ollie G Events Team to say a special thank you to them as they kindly made a donation to help us make our son Harry's dream come true. Harry is an avid Disney fan and this October with the help of the Ollie G team it became possible for us to take Harry to Disneyland Paris.

Harry was diagnosed with Hunter Disease, MPS II, and has endured many hospital appointments and often painful tests, undergone major heart surgery as well as weekly infusions all within the space of 18 months.

We have found Harry a true inspiration. He always has a smile on his face and is such a happy positive and brave little boy who takes everything in his stride. We felt he truly deserved a special treat. We all had a fantastic time and Harry and his sister Olivia didn't know where to look first as there was so much to do! But Harry's absolute favourite was the Star Wars ride. He is now convinced he has been to space! Once again thank you to all that were involved in helping this special little boy's dream come true and creating wonderful memories for our family. **Paul, Amy, Harry and Olivia Robinson**

Dear David Gosling and the Ollie G Summer Shoot

We cannot thank you enough and are overwhelmed by the very kind generosity from the organisers and the people on the tabled named after Hannah. We are deeply touched to think that people who have not even met Hannah would like to help her and make things better for her.

We will be able to buy the DVD players for the car which will help enormously on those weekly, sometimes more, trips on the motorway. Even when the waiting time for appointments have been known on several occasions to be up to three and a half hours, at least we can go back to the car to watch a film! We have also brought a reliable and more high tech thermometer. We need to have accurate readings for Hannah as she is unable to receive her treatment if she has a temperature. So now we will be able to spot earlier if we think she is unwell and get her to the doctors to get treated and then she won't miss her vital enzyme treatment. This in itself is a little battle to make sure she is well enough to have her treatment. When Hannah does receive the enzyme we can notice the difference remarkably.

Some days are very hard to cope with what Hannah has got, but she is always a happy child and always makes the best of what she is able to do, which keeps us going. However, this generous gift will help make things sweeter for her with all the travelling we do because of the MPS VI, Maroteaux Lamy Disease. For us, knowing that there are very kind people out there who have helped our little girl – we will always be very touched.

Our very best wishes and gratitude to everyone concerned.
***Lucy and Edward Brock (parents of Hannah MPS VI)**.*

Dear MPS

A sincere and heartfelt thank you to all concerned and to the Ollie G Events team for your very generous cheque we received for our wonderful little son Nathan who was diagnosed with Hunter Disease, MPS II.

With the money that was raised Nathan now has a fire engine, police bicycle and a great bouncy castle, and really smart clothes to go with it all. You have all made a little boy so very happy.

Yours sincerely
Eamonn and Michelle Worsford

Thank you to Ollie G

Here are a two photos of our recent weekend away at the Lake District. As you can see the boys had a great time, James thoroughly enjoyed himself as did we all. As the Ollie G event raised money so generously we were also fortunate enough to be able to buy the boys a 'Playstation 2'. We can't thank everyone involved enough, the money has made a real difference. Many thanks to all involved. **Ruth Hall**, mum of James (MPS II)

6th International Fabry Patient Meeting - Munich

18 - 21 October 2007

After a slight delay for most of us flying in from England some of the British Fabry members arrived in Munich to the Hilton Hotel.

There were 53 from the United Kingdom and there were 400 delegates at the conference including all the doctors and speakers.

There was slight confusion about where dinner was to be held but once we all made it to the 15th floor we were soon staring at each other's badges trying to find the other British delegates. There was yummy food and nice wine and before too long it was past most people's bedtime. We needed sleep before a very full day of speakers and listening to a head set to understand the four or five different languages that the speakers spoke. The programme was well managed and most speakers spoke in a way that was clear and well explained.

We found the information really informative but we know for some of our newer members it was a lot to take in. It was great having the support of some of the older members and their partners to support the newer ones. The one thing that we found so amazing was how important it is for our Fabry members to gather together to be a support to each other to share and understand what they are going through. We were both really touched by this.

From an advocacy perspective (for Chris Murphy) it was such an invaluable experience. It was great to put a name to a face, hear personal experiences and also being able to explain the role of advocacy and how we can support our members. We hope this will continue by email and phone so that we can stay in touch, as well as support you with whatever crops up.

A full day of talks and some proud moments with three of our group speaking for the personal testimonies part of the programme, Liz Carnie, Sharon Chatting and Laura Davenport. The testimonies were brilliant and we know many of the countries found it amazing to see a photo of Liz Carnie having a home infusion, which she administrates by herself.

We were coached into Ratskiller in the city centre of Munich for dinner on Friday night (thank goodness it wasn't the night of the Rugby!) to a very chaotic evening of more German meat (I really felt for the vegetarians). It was a huge restaurant that seated all 400 delegates as well as most of the population of Munich. Chris and I raced around to find all 53 British delegates to make sure everyone had arrived safely and had some food. Then back out to the street of Munich and back on to coaches! By the time we got back to the Hilton it was time for bed for most of us, although many other Brits stayed up a little longer.

Now onto the Rugby! Oh dear. I couldn't help the score but I could sort out with the amazing support of a friendly Antonio in the Business Services Centre of the Hilton to get a wide screen TV to watch the game! It was a shame that the Gala dinner started so late but that didn't stop some dedicated fans who exited after the second course, escaping with a pint of Beer! It was a good, if not very quiet atmosphere, only Anastasia in the background crying at the end of the game (she grew up in South Africa, and is very proud of her boys).

By the time the Conference was officially over we then had the option to go site seeing! It was cold outside and when we all got on the warm bus it was heaven to be sitting down, so warm and cosy that a few of us that will remain unnamed fell asleep. The city was beautiful! Miriam did see most of it.

Well, we think that sums up the trip well. It was a great time, loads of friendships built, loads of time to chat, learn more about Fabry and of course the beer and Rugby! What more can you want in a conference!

Miriam Blowers & Chris Murphy

Laura's Story

Hi, my name is Laura. I am 15 years old and I have Fabry Disease.

At a family meal when we were altogether with my brothers, Mum and Dad told us that we needed to go to the hospital for a blood test. I cried all over my dinner because I said that I didn't want to have a disease. I didn't know what to think about it as I had never heard of it but I found out that I had Fabry's when I was 11 years old.

When Mum told me that my eldest brother Thomas didn't have it I was jealous. Not because I wanted him to have it, but because I did. My other brother Matthew was also diagnosed and my cousins, Glennon and Lyndon. I felt that everyone around me had it as I am very close to my cousins. They used to live with us when I was younger and I see them very often.

I usually don't mind going to hospital in Cambridge because I like Uma (Dr Uma Ramaswami) and the staff because they are kind, friendly and look after me. Mum arranged the visits to Addenbrooke's with my two cousins and Matthew and we have a fun time doing lots of different things. We would go punting on the river at Cambridge, bowling, cinema and Duxton Air Museum. We would always make our hospital visits a family time of being together and having a good time which made the tests and the waiting around in hospital more easy going.

Now that the boys are older I am the only one who goes to see Uma as the boys are now under the Royal Free Hospital in London which is where I will go in a few year's time.

I do not have many symptoms but sometimes I get shooting pains in my hands and a ringing in my ears. I sometimes get chest pains as if someone is sitting on me but they go away after a while. I also often get dizzy when I stand up but it soon goes and I feel fine.

I didn't want my parents to tell the school because I didn't want them to treat me different from everyone else. I didn't think that the school needed to know but maybe it is a good thing that they do know because if I do feel unwell they have a history of my background.

I hadn't told any of my friends about Fabry's, no one, because it is my business and they don't need to know

about it. I am not ashamed of it, it isn't my fault. It is just a part of life and you have to learn to live with it.

Everyone should know whether they have a genetic condition because sometimes if you are ill then it's good to know an explanation for it. If you don't know, then you are deprived from treatment.

I know it is a decision for parents whether they tell their children or not, or when, but I believe that children have a right to know and that it can be talked about openly within the family. It shouldn't be a stigma or making it out to be a big thing. The more you talk about it, the easier it is to accept it and it becomes like everything. If you don't talk about it you make it harder for everyone and escalate everything that is associated. I would feel angry if I found out in adult life that my parents knew I had a condition and never told me, whether I was suffering symptoms or not.

I know that I can talk to anyone in my family about it and get help.

Matthew has ERT treatment and he just gets on with it and I suppose I would be the same.

I enjoy these international conferences because I meet young people my age with the same genetic condition as me and I have a good time. I have made friends and have kept in contact during the year and look forward to seeing them again. These conferences enable me to do this.
Laura Davenport

My name is Leslie Hilliard and I have just attended the meeting in Munich. I would like to say how much I appreciated being invited. I think Miriam and Chris are a credit to the MPS Society with how kind, caring and supportive they were. Again I would like to thank you very much for inviting me and my family to the 6th International Meeting.

Sharon's Story

This year's International Conference on Fabry took place at the Hilton Hotel in Munich, Germany.

The weather was noticeably colder when we first arrived in Munich and we immediately noticed how clean everywhere looked. The streets were devoid of litter, with wide open streets and the hotel was located close to a beautiful park interlaced with streams and lakes called the 'Tucherpark'. The trees were an array of Autumn colours like a Canadian fall.

We soon settled into our surroundings and went down for dinner at 7pm. There were many people gathered from all over Europe and everyone was busy chatting away. I recognised many familiar faces as patients, medical teams and carers mingled together. I also met Miriam and Chris from the MPS Society and it was nice to put a face to someone I had often spoken to on the telephone.

Chris and Miriam both a did a splendid job and looked after everyone extremely well. They went to the 'top of the class' on Saturday night when they managed to arrange a wide screen television to be set up to watch the Rugby World Cup final for the rugby fans amongst us.

The Conference was informative and interesting but I felt the main theme and the speeches, including mine, was aimed in particular at females with Fabry and did not encompass male patients in the audience. This gave the Conference an imbalance. Dr Patrick Deegan and Dr Waldeck did give very interesting and informative presentations, as did Dr Roland and Dr Ramaswami on children and young adults with Fabry.

My contribution to the Conference was to read my sister's speech who was unable to attend. It would have helped if I had remembered to blow my nose first and take a tissue up on stage with me. Having to wipe your nose discreetly with two huge video screens focusing on your face is not easy. Sadly, there was no airbrushing for me, and with baggy eyes and wrinkles shown in high definition on two, 6 foot screens for all to see, I persevered with the presentation.

My niece, Laura, was standing next to me and prodded me in the ribs when I gibbered too quickly. The wavering voice, which first emitted from my mouth surprised me, as I didn't recognise it as mine. I hoped the audience didn't think that I 'wobbled' normally and I hoped that the interpreters were 'wobbling' with me during the translation.

I have never been very technically minded and this was clearly evident when I could not work out the complexity of clicking a button when I wanted a new overhead picture on the screen. This made the photographs get a little bit out of sequence. When I was talking about London we were in Wiltshire and then a blank screen seemed to keep the audience focussed for what must have seemed an eternity until I accidentally clicked the button, realised my mistake and 'Hey Presto' we were back in London! Oops.

I always thought women were supposed to be multi-skilled and that I was no exception. But talking and having to press a button was apparently a bit beyond me… but then I was speaking in front of several hundred people!

My presentation was written by my twin sister, Juanita, who wanted to point out the diversity of Fabry, and how it affected each patient differently. She wanted to present a positive view how we as a family have dealt with the diagnosis in a positive and constructive way.

Next, it was Juanita's daughter, Laura, to speak. Laura is 15 years old, and so other than the worry about 'what jeans shall I wear, and is my hair OK' she wanted to advocate that it was important to her that she knew of her diagnosis and that this information was not kept from her. From her own experience she felt it was a positive decision that she had been told, particularly now that treatment has moved forward. The response she received from people of various nationalities was very supportive, particularly as this viewpoint came from a young person with Fabry.

An important aspect of the Conference is the opportunity to talk to people who share a common understanding and being able to share experiences. I learnt an awful lot from talking to people from different places in Europe in a very relaxed atmosphere and delightful surroundings.

I believe the Conference is extremely beneficial in uniting patients and sourcing information as a positive way forward and that there is an opportunity to be heard and listened to by both patients and experts in their field.

I would like to say on behalf of my family a very big 'thank you' to the MPS Society in arranging for us to attend and to Miriam and Chris for their wonderful support whilst in Munich; and to the Royal Free Lysosomal Disorder Unit, London Heart Hospital and Addenbrookes, Cambridge.
Thank you, Sharon and Family

Left: Liz Carnie and family; Right: Deborah Ruffley and partner, Alan Milligan (Royal Free) and Jane Banks.

Sarah's Story

Firstly I would like to say thank you to the MPS Society for allowing my Mum and me to attend the Fabry patient meeting in Munich this year.

Even though my father died from Fabry's nearly four years ago, Mum has always discussed things with me and she felt that it was important to keep the family involved and up to date with all the advances of the disease and treatment. I'm glad that she did this because it has given me a more positive outlook on my treatment and how I have coped with the symptoms up until now, now that I am old enough to understand the genetics and more scientific explanations, maybe more than Mum can. I feel that the Fabry patient meetings are a good way of getting the information across, I know that doctors keep an eye on everyone when you go for checkups and try their best to explain what is happening but sometimes it isn't the same as chatting to people with the same disease and similar problems as you.

Over the past few years that Mum and I have been to the conferences it has given us the opportunity not only to see places we would possibly not have been able to visit on our own, but for us to make friends with people who have the same condition as I do, and we have been keeping in touch through the years with a few people that we have met. Although the first meeting that I attended in Barcelona (when I was only 11) didn't have many young people at all I became friends with a German girl and we still occasionally write to each other (even though I do not speak German so it is a little tricky).

As the years have gone by more and more younger people have been attending the meetings from many other countries as well as the UK. I have now made many friends who I keep in contact with and feel that this is important because where they are the same age as me, we can not only talk about Fabry's but also music and clothes and things like this, and with technology becoming more popular it is a lot easier to keep in contact with them.

Also over the past two years there have been special workshops for the young people where the doctors make things a little easier to understand and we can ask questions to them about how we feel and get answers which are easy to understand. This year not only were all of those who attended able to watch a great performance from 'More balls than most' but all of the young people had the chance to attend a workshop where we were all taught how to juggle and got our own set of juggling balls. I think that most of us did more laughing than juggling but some say laughter is the best medicine.

I think that getting people with Fabry's together in one place is important as everyone lives far away and you cannot always meet with people (although me and Mum are lucky as we are now good friends with another mother and daughter, who is of similar age to me, who we met at these meetings and we are able to meet with them when I visit Addenbrooke's Hospital as they live near there.) Even Mum enjoys these meetings as she is not very good with technology so finds emailing difficult and sometimes has to get help from me.

Once again I would like to thank everyone involved for giving us a chance to attend the conference this year and I do hope that another meeting can be arranged somewhere next year. These meetings are so important to everyone who goes. **Sarah Hill**

We attended the meeting in Munich and found it to be excellent. The venue, presentations, organisation, only one fault was the poor weather but we can't expect you to get everything right! After being diagnosed with Fabry disease for over 30 years I am amazed I can always learn something new from these meetings. Thank you once again. **Mr and Mrs Burke**

A big thank you to you all. Especially to Miriam & Chris for all the hard work and effort that you put into the Fabry conference in Munich. It was one of the best I have attended and you two girls worked your socks off to look after us and care for us. It was a very comfortable conference for us as well as being informative. **Ian Hedgecock**

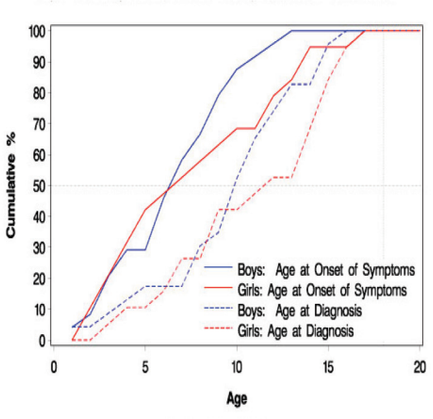
The pros and cons of Enzyme Replacement Therapy in Children and Adolescents with Fabry Disease

At the recent Fabry conference in Munich, Germany, Dr Uma Ramaswami, Consultant Paediatrician from Addenbrooke's Hospital, Cambridge spoke about the pros and cons of Enzyme Replacement Therapy in Children and Adolescents. Here we give a brief overview of her presentation.

Clinical Presentation In Children

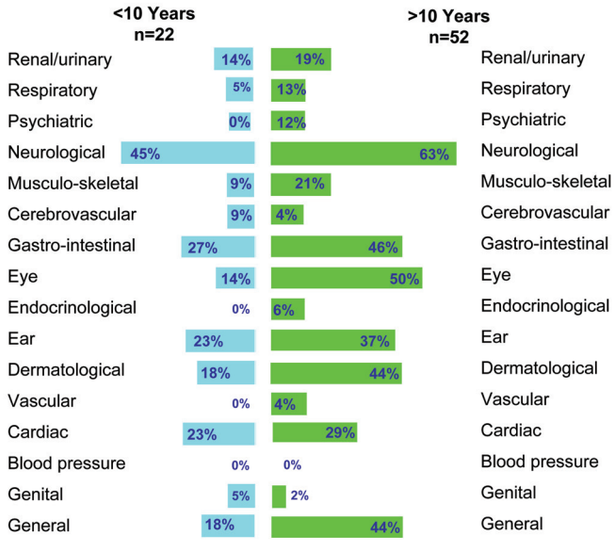
- Acroparaesthesia
- Gastrointestinal
- Hypohidrosis
- Angiokeratoma
- Fatigue
- Decreased physical activity
- Cornea Verticillata
- Proteinuria –in later childhood

Strokes/Cardiomyopathy/ Renal Failure rare



Data taken from the FOS Registry demonstrates how organs are affected by Fabry Disease in two age ranges, under 10 years (left hand column), and over 10 years (right hand column)

Affected organs



A number of children are in receipt of enzyme replacement therapy and the following observations were made from two paediatric trials:

ERT with agalsidase alfa

Both Multi Centre open label studies.
Agalsidase Alfa 0.2mg/kg fortnightly for six months

Ries et al, Paediatrics, 2006

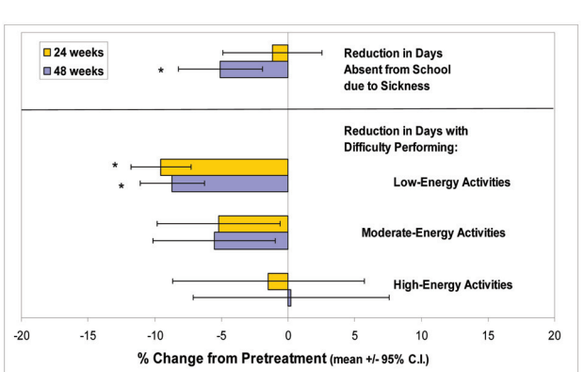
- 19 boys and six girls over 6 years of age
- ERT well tolerated
- One boy developed transient IgG antibodies, which resolved without stopping ERT
- Overall Infusion Reaction Rate 5.4%
- Microalbuminuria in 4 patients halved at 6 months
- Both abnormal heart rate variability and sweating improved at 6 months

Ramaswami et al; Acta Paediatrica 2007

- 9 boys and six females between 2 to 18 years
- ERT with agalsidase alfa is safe and well tolerated in children
- One patient developed IgG antibodies at 12 weeks. Continued to show clinical improvement
- Plasma and urinary Gb3 reductions at 12 weeks
- Improvement in pain scores noted
- All patients continued on ERT at the end of clinical trial

The key findings were a significant improvement in pain and general health and there was evidence of improvement in school attendendance and peformance in activities.

Improvement in School Attendance and Activity performance (% change, e-diary) Agalsidase beta



% Change scores are derived from a Logistics Repeated Measures model
95% C.I. = 95% confidence interval on mean change from pre- to post-treatment

Genetic Counselling

My name is Judy Holroyd, I am a Trustee of the Society. I was asked to write an article about genetic counselling for the magazine, particularly by some of the families who attended the Fabry Conference in Munich.

I have been working as a genetic counsellor in the NHS for the last six years. I hope the information below is helpful and answers some of your questions. I will start with a personal story of how genetic counselling has helped our family:

Our eldest son William was diagnosed with MPS IIIA Sanfilippo disease when he was 10 years old in 1984. We were living in Australia at the time and my husband and I will always appreciate the genetic counselling we received. Following testing that confirmed William's diagnosis we had an hour long appointment with the Geneticist at the Melbourne Children's Hospital. He took time and care to give us a full explanation of the disease and to answer all our questions and concerns. Every point he made was clear and he was honest when he did not have an answer. We were able to understand fully the genetic nature of the disease and the implications of this for our family. At that time, when we were both still reeling from the shock of the diagnosis, there was an overwhelming amount of information for us to absorb. However, the letter we received after our appointment covered everything that we had discussed. For many years after we passed on copies of the letter to teachers and health professionals to help them understand more about Sanfilippo disease. Sadly, William died when he was 22 years old.

Since then the genetic basis of Sanfilippo A has been determined. A blood sample was taken from William in 1984 and his DNA stored. This has enabled his brothers and sister to find out if they are carriers for the disease and two of them have now received genetic counselling.

What is genetic counselling?

To quote from the website of the Human Genome Project:

"Genetic counselling is a health care service available on the NHS to individuals, families or couples. It aims to bridge the gap for them between the often complex and fast moving field of genetics and their everyday world. It helps them understand the nature of the disease and what having it will mean in practical terms, what options there might be for prevention/testing, the risks of recurrence and the implications for other family members."

Crucially, genetic counselling is non-directive, supporting people in reaching their own decisions, based on their own unique medical and social circumstances.

Genetic counselling touches very deeply on human emotions of guilt, grief and fear, and on deeply felt moral beliefs. Counsellors are trained to help people through the inevitable emotions that a diagnosis arouses and which ripple through the whole family because of their shared genetic inheritance. No two patients are the same, and genetic counselling has to be sensitive to the fact that a diagnosis can have very different meaning to different people."

Genetic counsellors usually have a scientific or nursing background and complete a masters' level training programme in genetic counselling before coming into the profession. After teaching science for many years I decided to retrain as a genetic counsellor and in 2001 I joined the All Wales Medical Genetics Service. I work in Cardiff as part of a medical team and together we provide a clinical, diagnostic and genetic counselling service for families covering a large range of genetic conditions. In my job I meet parents and children, grandparents, other relatives, pregnant couples, other teenagers and adults. In the rest of the UK, including Northern

Ireland, there are a number of regional genetic centres that offer a similar service.

How does the service operate?

Some people are referred to the Genetic Service by a hospital consultant or their GP following a diagnosis. Other people may be referred because they are concerned about:

The chance of either inheriting or passing on a condition known to be in the family

The risk for a current or future pregnancy

Their child who may have a genetic condition

In Wales we discuss all referrals to the Genetics Service at a weekly meeting and a management plan is put in place. Some of my referrals I will look after independently and others will be seen by a Genetics Consultant after an initial appointment with me. Many people are uncertain about what to expect from an appointment with the Medical Genetics team and we send an information leaflet about the Genetic Service with their appointment letter.

For children, adults and families newly referred to the Genetics service, I am usually the first point of contact. I will arrange an appointment either in the hospital or at home whichever is more convenient. The purpose of this first appointment is to explain about the service, identify any questions and concerns and what the individual/family hope to gain from the service. I will draw up a family tree and identify further relevant family medical history that will help us assess whether there is an inherited condition in the family. This is important preparatory work for the appointment with the Genetics Consultant.

For individuals or families with a previously diagnosed genetic condition or a known family history of a genetic condition, I usually manage these independently. My aim is to help them understand the medical facts about the condition, how it is inherited and the risks to them and/or other family members. I go through the options available so that they can choose the most appropriate course of action for their family circumstances. If genetic testing is available for the condition, this may include taking a blood sample for testing. Following the clinic appointment I write to the individual or family going over the points we discussed and let them know the results of any tests. If appropriate any ongoing follow-up can then be arranged.

Requesting a referral to the Genetics service

If anyone feels that a referral for genetic counselling may be helpful, they can ask their GP or hospital doctor for a referral to their local Regional Genetics Centre. **Judy Holroyd**

Enzyme Replacement Therapy at Home

Those of you fortunate to be on Enzyme Replacement Therapy (ERT) and receiving your treatment at home will appreciate how important the people making the delivery of the drug and the nurses are to you or your child. The provision of Homecare for ERT comes through one of several independent commercial health care providers. Currently there are two major companies providing support for receiving ERT at home - Careology and Healthcare at Home.

The article that follows describes the work of one company, Healthcare at Home. In the next MPS Magazine we will share the work and ethos of Careology. Everyone involved in providing Healthcare is expected to work to set stringent standards. Like all services, from time to time, these standards fall short. If you have any problem or concern about the performance of your home care provider for ERT that are not or can't be resolved, please do contact the advocacy team.

In the New Year we will be carrying out a stakeholder evaluation of the Homecare services and look forward to our members receiving home care participating. **Christine Lavery**

Healthcare at Home

"Be it ever so humble - there's no place like home"

The last decade has seen a dramatic shift of what were once traditionally seen as hospital-based treatments, to safe and effective administration in the home environment. Home care offers significant benefits to those patients who can be treated at home - addressing the inconvenience of time-consuming visits to hospital, transport difficulties/expense, absence from work and alleviating the stress that can be associated with a hospital visit. More importantly, it allows patients to retain control of their lives - leading as normal a life as possible with the minimum of disruption to the family unit.

Lysosomal storage disorders represent a group of diseases which can mean a lifetime of therapy. Due to constraints on the number of clinical specialists within this area, patients travel long distances for review and treatment. Difficulties of travel, associated time commitment and limited hospital resources can impact on the quality of life of the patient and their family. As a result, specialist centres are exploring the different treatment options available for each patient, as determined by their individual circumstances and preferences - one option being home administration of enzyme replacement therapy (ERT).

Established in 1992, Healthcare at Home provides high quality specialist nursing and pharmaceutical services that reduce and prevent the need for hospital-based treatment. With its main operating base in Burton-upon-Trent, Staffordshire, and supported by an additional 24 regional offices located throughout the UK and Ireland, Healthcare at Home is uniquely positioned to provide a parity of service to all patients, regardless of their geographical location.

Healthcare at Home employs over 250 highly skilled and experienced nurses, who work as an extension of the hospital team to ensure the highest standards of care in the provision of specialist infusion services to adult and paediatric patients at home. No agency nurses are ever used, and implementation of stringent Company protocols and operating standards ensures delivery of a controlled, high quality service at all times.

Each patient is allocated a named nurse, thus allowing continuity of care and the development of a close working relationship between the patient, their family, and the nurse, thereby facilitating an environment where individual patient needs can be communicated, understood and actioned. Patients are supported by a 24-hour, 365 days-a-year, nursing on-call service - where advice or visit, if required, can be discussed directly with a nurse from the regional office local to the patient.

HOME TREATMENT

Complementing the specialist nursing teams is a comprehensive and integrated support system - including pharmacy, aseptic compounding, logistics, home delivery and customer services. With the high cost of ERT, and to avoid unnecessary levels of stock-holding by the patients, regular deliveries are made by Healthcare at Home's own fleet of refrigerated vans on a day, time and to a location convenient for the patient. A small dedicated team of ERT customer service administrators co-ordinate the service requirements on an individual patient basis.

Delivery of medication, ancillaries and infusion of therapy can be arranged for patients who take holidays within the UK, thereby minimising the disruption to family life and allowing as near normal activities as possible to be maintained for the family unit. Additionally, assistance with refrigerated transportation of medication and associated customs documentation can be provided for patients who wish to take holidays overseas.

Healthcare at Home is fully accredited with the Commission for Social Care Inspection (CSCI), holds ISO 9001:2000 and Healthcare Accreditation and Quality Unit (HAQU) accreditation across the full range of services.

In December 2000, Healthcare at Home was named the second fastest growing company in the UK in the Sunday Times "Fast Track 100". This achievement of Fast Track status was followed up in 2001, 2005, 2006 and again in 2007. Further to this, Healthcare at Home was presented with a National Customer Service Award presented by Sir Richard Branson.

Healthcare at Home has 10 years' experience of home infusion of ERTs and currently provides home treatment programmes for both adult and paediatric patients with four of the lysosomal storage disorders - Fabry disease, Gaucher disease, Hunter disease and Maroteaux-Lamy syndrome - and will shortly introduce a fifth home treatment programme for Hurler/Hurler-Scheie/Scheie patients.

With almost 200 Fabry patients currently registered on treatment, Healthcare at Home works very closely with the hospital team, the patient and their family to provide a package of care specific to the individual requirements of each patient. Therapy can be administered safely and effectively at home by either nurse infusion, or by training patients, or their carer, to self-infuse at home. The option for self-infusion allows the restoration of an independent lifestyle whereby infusions are integrated into the patient's normal routine, with an associated enhancement of quality of life for the patient and their family.

To enable safe infusion administration by the patient or carer, a personalised training plan is agreed between the nurse, patient and carer ensuring that adequate time is allocated to each element of the training plan to ensure patient/carer competence is achieved, regardless of the length of time taken to achieve this. 'Competence' is defined as the knowledge and ability to perform the action and also the confidence with which to perform it. At all times, the patient continues to be supported by the Healthcare at Home team, allowing complete flexibility of access to all service elements, thus ensuring the changing needs of the patient are always met.

To date, approximately 33% of all Fabry referrals to Healthcare at Home have been successfully trained to self-infuse. An essential element of this independence is the provision and availability of a fully comprehensive support service incorporating customer services, logistics, pharmacy and nurses - both during normal working hours, but more importantly 24 hours a day, 7 days a week. Over the past few years Healthcare at Home has invested significant resources in order to improve services for patients and their families. Further service development and innovation is planned and will focus principally on improving communication channels (e-mail, web access etc) and enhancing the convenience of home delivery (e.g. text messaging of expected time of delivery, extended delivery time options etc). Whilst patient surveys reveal high levels of satisfaction with homecare services, patient feedback and suggestions are always welcome to help guide future developments.

In conclusion, the potential commitment required from both the patient and their family when receiving ERT can be somewhat daunting. By working closely with the hospital clinical teams, the patients and their families, Healthcare at Home has developed a homecare service which allows safe and effective management of therapy in the community, allowing the patient to retain control of their disease and minimise the potential impact it may have on their lives.

For further information on the services provided by Healthcare at Home, please contact:

Jill Stephenson — ERT Operations Manager
0870 600 1540 — jills@hah.co.uk

Dr Gareth Jones — PharmaService Director
0870 600 1540 — garethj@hah.co.uk

My Fight to get Enzyme Replacement Therapy

My name is Vibeke; I live 30 km from Trondheim in Norway. I'm 26 years old.

In Autumn 1991 I was diagnosed with MPS VI, Maroteaux Lamy Syndrome. I was then around 10 years old.

The reason my parents were concerned about my health was my hands, which were different than other kids' hands, I had a heart problem, something to do with a leaking heart valve and frequent ear infections. Other than that I was like other kids my age.

In March 1992 we were sent to Frambu for a two week stay. For those who don't know much about Frambu, it is a centre for rare diseases. Families go there to get information, to get help to cope with their child's (or their own) disease and to meet other families that know how it is to have a child with a disease. There are also summer camps throughout the summer (two camps for kids from the age of 10 to 16 and two camps for adults from 17 to 30) - a wonderful place! At this stay my parents talked to a doctor who said to them that in 10 to 15 years there would be a treatment for their daughter.

I didn't accept the disease... I wanted to be treated as normal. I wasn't sick… I also did all the things that my classmates did.

It wasn't until 2000 I became more aware of my disease. After the New Year celebrating 2000 I started to lose feeling in my feet and arms… I was sent to the hospital, and took some tests but they didn't show that anything was wrong. But the final test was an MRI and from this they decided to do surgery on my neck. The surgery was planned on 7th February 2000 (I was 18 years old). Although the doctor told my parents that the surgery would take only 2 - 3 hours, I was in surgery from 8:00am until 6:00 pm! After the surgery my feeling came back and I was so happy. I didn't know at the time that this surgery could actually have paralyzed me… Luckily I didn't know.

Slowly I was back to normal again. After my return home from hospital we also got access to the internet, so I started to read more about my disease.

In 2003 I was at Frambu again. There was an MPS meeting, and I met other people with an MPS disease for the first time. It wasn't quite the same type of MPS because I know I'm the only one in Norway with MPS VI. So I'm very rare here in Norway.

Whilst at Frambu, I talked to Dr Ed Wraith (a wonderful doctor) for the first time; he was then talking about a new medicine for which they were starting trials. Exciting news for me. (This was 11 years after my parents talked to a doctor about treatment for this disease.) Dr Ed Wraith also said to me that I had a less severe type of MPS VI. More exciting news for me.

I don't regret that I attended this stay, it was the start of my journey to fight for ERT. You probably think that it would be easy… well, it wasn't. So here comes my journey:

In May/June 2005 the enzyme was approved by the FDA in USA, and I thought that soon it will be approved in Europe too. Well, I had to wait around half a year more. On 30 January 2006 it was finally approved in Europe by the EMEA… I was happy, but also sad. It was only a short time after my mother earned her angel wings… She earned her angel wings Friday, 13 January.

I thought that after European approval it should be a piece of cake to start the treatment but this was not to be the case. Money is a big problem; it shouldn't be a problem in Norway. When you think of what they say about Norway: that Norway is one of the richest countries in the world. Well, only outside, not from the inside.

ERT has to be given at the hospital, which means that in Norway hospitals have to carry the cost of the medicine. (My opinion is that the state should pay, but I'm not a politician…). So nothing happened and my case laid idle!

In February 2006 we got a new family member, a little puppy. His name is Kermit (like the green frog in The Muppet Show). He is a Bichon Frisé, the world's most beautiful dog. He has helped me a lot.

In November 2006 I once again attended a seminar at Frambu, and also this time I spoke with Dr Ed Wraith. He was curious to hear if I had finally started ERT. Well no, I said.

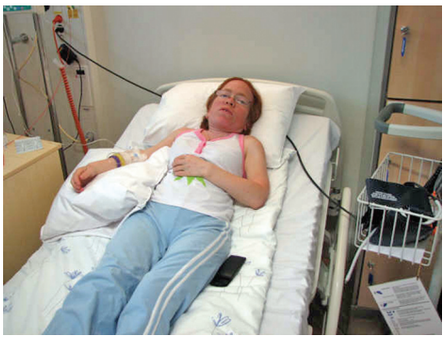
In December 2006 I got a call to say that the treatment was approved and that I was going to get it. Hooray! Finally! But I didn't hear anything more. I thought it was because of Christmas and that I would wait until after the holidays. However, I still didn't hear anything. I emailed my doctor, but he hadn't heard anything either.

Then my good friend Rita Hausken Barkhodaee (mother to a child with MPS I), asked me to send her a letter with my thoughts about the situation. Rita sent this letter to Frambu, and after some weeks I got a call from Frambu. They said that there had been some misunderstanding and that it wasn't approved to give me treatment after all. Once again I was left in limbo, just waiting. How can they torture people like this? Anyway I'm getting good at waiting.

In February I got sick, with what I thought was a cold, but actually pneumonia and I was almost hospitalised. This was my first pneumonia and I thought "It's time for the enzyme to be approved for me!"

In March I received a phone call from the hospital as my doctor wanted to see me. He wanted to talk about ERT. So off I went to the hospital and my doctor said he would send one more letter to the Director of the Hospital and if this didn't help, I would need to go to the media. I really hoped that the letter would help. But no, we didn't receive any answers, only that they won't pay for it because it was too expensive… But hey… three other patients who have MPS I get ERT in the same area, why not me?

So my Dad called several TV channels and newspapers. In May I received a phone call



from NRK (a Norwegian National TV-channel), as they wanted to interview me.

Only one hour before the interview was supposed to go on air; my doctor called me, the hospital director had said YES! The interview wasn't put out on the news - maybe it should have been so that people could see how patients in Norway are threatened.

I got my first ERT on 13 June 2007 and everything went well! I didn't get any bad reactions, only tiredness.

After five treatments I started to notice some changes. Maybe small changes for you, but big to me. My hair is softer and I sleep longer in the morning, I eat more and have gained some weight.

Some of my friends ask me why I'm willing to be hospitalised one day a week when I'm not that sick. One also said to me when I was fighting to get the treatment: "You are too healthy to get the treatment, that's why you don't get it…" But that isn't a reason; I needed this treatment to try to prevent the disease doing more harm to my body.

Now I've had 19 treatments, and everything is going well with no allergic reactions.

I'd like to thank Rita H. Barkhodaee. She helped me a lot, and if it wasn't for her and her pushing me to fight, I wouldn't have started yet. I also want to thank Frambu; they've helped me a lot in these last few years. I am forever thankful to you all. **Vibeke Hovd**

10th International Symposium
on MPS and Related Diseases

26-29 June 2008

Vancouver, British Columbia, Canada

Forward Planning in Canada

Christine and I recently visited Vancouver, Canada to meet with the organisers and discuss all the finer details of the International Symposium being held in June 2008.

After a 10-hour flight I was ready to get into bed. I was surprised how much a 10-hour flight takes it out of you (I consider myself a little bit of a professional jet-setter after flying back and forth from Australia so many times). The Sheraton Wall Hotel didn't disappoint and before too long we were checked in and enjoying a comfortable space as well as investigating all there was to know about the hotel.

The next day we shared our experience of running conferences and looking after MPS children and siblings whilst parents enjoyed the conference, with the Pinnacle Pursuit Group who will run the childrens' activities next June in Vancouver. They have put together a brilliant programme for the children attending the conference called 'Camp Canada'. I was wishing I was still young enough to attend and we briefed them too on MPS and what to expect from some of the more challenging situations they might find themselves in.

Over the 25 years of running the conferences here in the UK we have been able to establish good systems that keep our members' children safe and sound as well as having a great time! I can say without boasting (as I started work here at MPS in January 2007) that we do things quite well and it was a proud and exciting process to explain what works and what doesn't and all the things that go on behind the scenes to pull off our National Conferences and other events. So once all wisdom was handed over we set about having a tour of the hotel to see where and how the conference for next year was going to flow and talked (what I find very exciting) logistics!

That night we were taken out for dinner to Grouse Mountain. I was told we were going on a gondola (which I thought was a boat) but it wasn't a boat! I did wonder how a boat was going to get us up the mountain! It was a huge cable car which stayed flat but carried us up a huge mountain to the top for dinner. I was terrified but was very brave and conquered all my fears by breathing a lot and not talking about the fact that we were way too far off the ground for my liking! Dr Lorne Clarke, Consultant Paediatrician at the British Columbia Children's Hospital, didn't help matters on the way down the mountain which I think was worse than going up by making comments and telling stories about it falling once upon a time! Thanks Dr Clarke, you were not one of my favourite people that night!

During the following couple of days we hired a car and went off to explore sight-seeing options to take the families on before the conference starts. Another terrifying experience was crossing Capilano Suspension bridge with my Boss Christine Lavery giggling behind me telling me how brave I was and I could do it. This bridge is so far off the ground and it wobbles a lot. I must have looked ridiculous! But all in the matter of forward planning I braved it, there and back, and was even able to smile for some photos.

By the time we were due to depart a very cold and wet Vancouver I was exhausted but pleased that we had come to see whether we have wheelchair access and what will be easy and what we need to start sorting out for the visit for next June.

Vancouver is a beautiful city with friendly people and lots to see and explore. The airport alone is worth visiting with a beautiful aquarium! We had the pleasure of being hosted most of the time by Kirsten Harkins, the Executive Director of the Canadian MPS Society. She is wonderful and working very hard organising the Conference for next June.

I have already started booking the itinerary and working out costs. Nothing like being ahead with organising logistics!

Miriam Blowers - Events Coordinator

Visit to BioMarin

Having travelled to the East Coast of the USA, California, to attend the American Society of Human Genetics Meeting it seemed good use of time to visit Dr Emil Kakkis and his team at BioMarin in Novato.

The Society and Dr Kakkis go back to the mid 1990's when Dr Kakkis was working single-handed to develop an Enzyme Replacement Therapy for MPS I. Post marketing ERT for MPS I became the responsibility of Genzyme. However, BioMarin has grown over the last ten years and has since developed and brought to market ERT for MPS VI. In discussion I learnt that BioMarin has a number of other products in development for orphan diseases. Watch this space.

During my visit on 31 October 2007, I was shown round the manufacturing plant that produces Aldurazyme and Naglazyme and it really makes one appreciate the enormity of the task. As the manufacturing plant supplies drugs worldwide every country sends in its own inspectors to check health and safety as well as regulatory compliance. Therefore BioMarin is rarely without visitors.

I would like to take this opportunity to thank everyone at BioMarin for making my visit so worthwhile and for all they have achieved over the last ten years.
Christine Lavery

Dr Emil Kakkis and Christine Lavery

Clinical Trial Update, Lyon 26 - 27 July 2007

Six months had passed since the start of the Miglostat trials in Lyon began and it was time for the two UK families to travel to Lyon to undergo a repeat of the baseline tests in order to monitor progress. The results came through in September which unfortunately showed no significant improvement and as such the children will continue on the double blind placebo controlled trial until January 2008. **Steve Cotterell**

Eighth workshop on partnering for rare disease therapy development: The reality of orphan medicines

Copenhagen, 18-19 October 2007

The starting point of this workshop was simple; children and adults with rare diseases have no choice over their disease but are as entitled to an equitable treatment as children and adults with more common conditions.

It was an enormous privilege to be invited to EPPOSI to make the key note speech 'A patient's view on health technology assessment for orphan drugs: are there models?' in the Danish Parliament in front of her Royal Highness Crown Princess Mary of Denmark.

I presented the scenario of how MPS achieved National Commissioning in England and the situation in Scotland, Wales and Northern Ireland. Thank you to the families who allowed me to use their stories. Over 120 stakeholders from patient groups, industry, research, hospitals and authorities from both sides of the Atlantic participated with the aim of seeing where consensus lies, what the facts are, how we estimate the value of the innovative medicines now (and in the future), what further research is needed and how we might go about finding data.

Briefly the conclusions which emerged from the EPPOSI workshop sessions were:

Despite uncertainty about exact numbers, there is no likelihood of an avalanche of expensive new orphan medical products within the next five or even ten years – not such good news for patients, but payers need not be alarmed about setting precedents that would lead to an imminent exponential rise in costs.

The real cost of orphan medicines may be much lower than thought, once account is taken of reduced costs in other areas, plus the contribution that some patients may be able to make to society through employment.

Equitable and timely access to orphan medicines varies not just between countries but also within them. That is a problem not just for patients but for Europe as a whole. Health technology assessment is a useful, indeed vital, tool for decision-makers if adapted to the scarcity of available data, but it can only be one element of the decision-making process. But patients need to be both won over to the concept and included in the process as partners; and politicians should recognise that the final decision is theirs, a political rather than solely an economic one.

More research can and should be done to establish the real value of orphan medicines; the number of patients in Europe with orphan conditions; the likely number of those conditions for which treatments (and perhaps cures) will be found; and the timescale over which those will be developed.

Good databases and patient registries, preferably international, are essential to establish how orphan medicines actually work in real life. In all these areas, there are examples of good practice that can be followed and developed. Dialogue, partnership and transparency, nationally and internationally, will hold the keys to progress.

These conclusions should make worrying reading for policy-makers but, unexpectedly, perhaps reassuring reading for the payers. **Christine Lavery**

Left to right: A Kent, C Lavery, W Goettsch, E Jessop, E Tambuyzer, T Gronnebaek, F Borlum Kristensen, F Meyer

New Clinical Trial Initiatives

This article on new clinical trial initiatives is taken from a presentation given by Barbara Wedehase, Executive Director of the National MPS Society in the USA, at the International Network Meeting in Brussels, Belgium on 16 November 2007.

MPS I

A cognition study is taking place at the University of California in Los Angeles (UCLA), USA. The lead doctor is Dr Patti Dickson, in collaboration with Dr Chen, Dr Charnas and Dr Shapiro. The criteria is that patients must be aged 6 years or over. This is a one year randomised controlled study with no placebo and with the option for a one year treatment extension.

A Haemopoetic Stem Cell Transplantation study is underway at the University of Minnesota in Minneaopolis, USA led by Dr Paul Orchard. This study is for children under the age of 5 years and is for one year's duration following transplant.

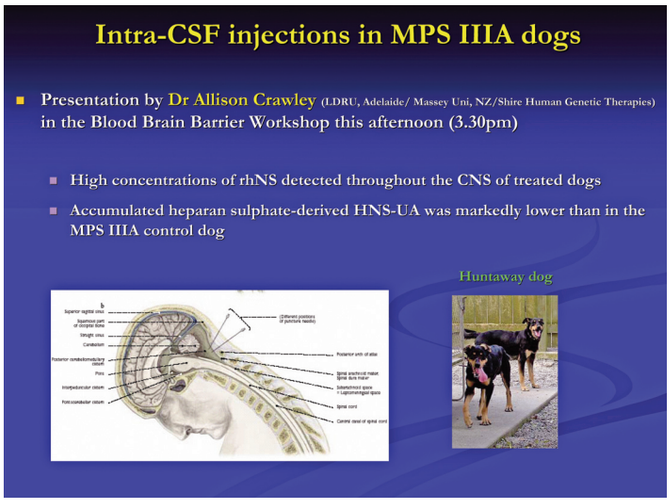
Dr Patti Dickson is leading a Spinal Cord Compression Study at UCLA, Los Angeles, USA. This is a one year extension study for children aged 8 years and older (by December 2007).

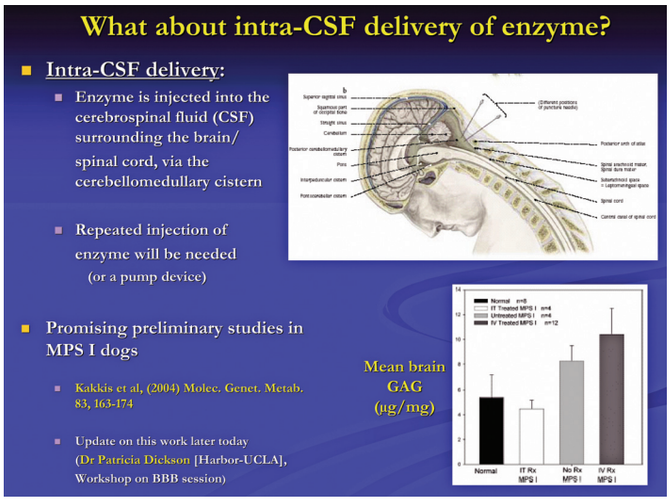
MPS II

A Shire MPS II Intrathecal Study is taking place at the University of North Carolina in Chapel Hill, North Carolina, USA, led by Dr Joseph Muenzer and Dr Maria Escolar. This is a phase I and II trial measuring safety and dosage with the goal of preventing CNS involvement.

MPS III

A Shire-sponsored MPS III natural history study is underway in preparation for an MPS III-A intrathecal clinical trial. Previous intrathecal studies have been conducted in dogs with MPS III-A.





New DVD resource: Communication and Challenging Behaviour

Following the very positive feedback received from earlier DVD resources, the Challenging Behaviour Foundation has now released a new DVD: 'Communication and Challenging Behaviour'.

Communication is one of the most important ways in which we control our environment and influence other people. However, many people with learning disabilities may have difficulties with communication, and these difficulties may contribute to challenging behaviour.

Focussing on individuals with severe learning disabilities, in this 45 minute DVD we meet Rhys, Laura, Sean and Isobel to examine the link between communication and behaviour.

Interviews with family carers and a speech and language therapist highlight some common communication difficulties, illustrate different communication approaches available and demonstrate the importance of all carers working together to develop communication skills and to minimise challenging behaviour.

Cost: £30.00 plus p&p; free to parent carers. (Reduced rates for registered charities).

Challenging Behaviour Foundation - T: **01634 838739,** E: info@thecbf.org.uk, www.challengingbehaviour.org.uk

Mucolipidosis - Where are we?

Pamidronate
There are now approximately 30 children and young adults around the world using Pamidronate for Secondary Metabolic bone disease. All those being treated have shown a marked improvement in the bone pain and bone density. In New Zealand we have one patient with MPS VI also on Pamidronate. We have two Alpha Mannosidosis patients, one in New Zealand and one in the USA on a Bisphosphonate.

As we have gone through this uncharted water and come up against problems such as dental care and surgeries we have had to address these issues and decide what would be the best way to deal with them.

Children facing major bone surgeries have stopped Pamidronate about 3 - 4 months prior to surgery to allow the bones to settle down. The reason for doing this is to stop bone fractures.

An example is taken from Hayden who had hip surgery five years after starting treatment and Pamidronate was not stopped. He ended up with a bone fracture of his femur which took quite some time to heal. He has recently had spinal surgery and stopped Pamidronate four months prior to surgery and has not yet gone back onto treatment as he is not showing any signs of bone pain.

Children needing dental care have also stopped treatment several months prior to treatment as a precautionary method to prevent cronosis of the jaw which is seen in people who are on extremely aggressive treatment.

I still continue to receive e-mails from doctors around the world who are treating ML patients and stumble across Pamidronate treatment in ML. It is highly likely that there are many more children being treated than we actually know about.

Natural History Study for Mucolipidosis
The following article was written by Denise Crompton for the ISMRD and LDNZ newsletters. We are very privileged to have such experienced doctors such as Dr Sara Cathey from the USA and Dr Jules Leroy from Belgium working on this very important project that has led to more than 50 ML families world-wide knowing their gene mutations and understanding how this affects their children.

Families from the USA, Canada, New Zealand, Australia and the United Kingdom took part in this study. Dr. Sara Cathey called it serendipity. We called it an answer to many prayers.

A variety of efforts made by dedicated members of the medical community and dedicated parent advocates created the circumstances that led eight individuals affected with Mucolipidosis II and III and their families to the Greenwood Genetic Centre (GGC) in South Carolina.
Since 1974, the Greenwood Genetic Centre has been a leader in the delivery of genetic services, diagnosing genetic conditions; counselling individuals and families; researching the causes of birth defects and genetic diseases; and educating health care providers, teachers and the public about genetics in the United States.
The lab at the Centre has started to compile the first ever Natural History Study for Mucolipidosis, having completed extensive testing on gene mutations of ML II and III. This will help the GGC, families, and future researchers to better understand the manifestations of this condition. Now, they want to analyse the information more closely to determine if specific gene changes are associated with specific clinical features.

Since the gene for ML II and III, known as GNPTA, had recently been isolated, this Centre believed that an in-depth study with as many affected individuals as possible was important to undertake. We were all happy to participate. Once our lab work was done, and they established the markers in each participant within the GNPTA, they sent us questionnaires to be completed. The next step was for them to see Mucolipidosis II and III individuals in order to take photos, x-rays, and family histories.

The GGC staff were most gracious and accommodating to all of us. Everything was well planned, although we did run into a glitch with X-rays. We laughingly assured the doctors that we were accustomed to running into such problems.

We were treated to lunch and presentations by doctors explaining about the concept, origin, and growth of the Centre, as well as plans for expansion. GGC works with the South Carolina Department of Disabilities and Special Needs to

provide diagnostic services, treatment, and prevention programs to reduce the risk and severity of disabling conditions. They are breaking new ground in clinical service, laboratory and diagnostic testing, and providing hope for every family at risk of genetic disease.

Dr. Mike Friez, Director of the Molecular Diagnostic Laboratory, used charts and analogies to assist us in understanding his overview of the concept of DNA. Some of us took notes (the human genome contains 30,000 to 40,000 genes), and later joked with each other about our hopes that we wouldn't be tested. A tour of the labs helped us to more fully comprehend the way in which the genes were mapped. We were assured that they don't want to compete with the popular TV show CSI. It really does take much more than an hour to complete all the lab work!

This study of GNPTA is a genotype (genetic makeup of an individual), and phenotype (external appearance of an individual and their characteristics) produced by that individual's genotype interacting with the environment. In our private meetings with staff, we learned how very complex this condition is. This is the reason that we see a variety of ways in which it is manifested in different individuals.

It was difficult to say goodbye at the end of our gathering, particularly so for our precious children, who have quickly bonded with others like themselves. Sincere promises to meet again in the future were exchanged, along with warm hugs. We left knowing that a second group like ours would be meeting the following week to have an unforgettable experience in Greenwood, and that our world of ML II and III holds so much more promise for the future than it once did.

Denise Crompton
Author of Kelley's Journey: Facing a Rare Disease With Courage

Natural History Update
While in Brussels on 16 November Wendy Boon and Jenny Noble met with Dr Jules Leroy over their evening meal. It was a real privilege to hear how from the very grass roots of the Natural History study Dr Cathey and Dr Leroy are now moving forward with the study and are looking at two groups of ML children who have unusual mutations. These are seven ML II children who are more MLII/III, (five of these seven children come from Australia), and five ML III children who have stop codons in their DNA.

My two young adults Hayden and Sarah have received a stop codon from their mum and we wait with interest to see what is unravelled in this very complex disorder. Dr Cathey and Dr Leroy are most interested in the bone conditions that this group suffer from and would very much like samples from children when they are having surgery.

Knock out Mouse models
One more area that is really exciting is the development of knock out mouse models which have been developed by Dr Stephan Tiede from Germany. These models are the Alpha Bata subunit model and the Gamma subunit model.

What we do understand from all the work that has been happening in Mucolipidosis is that most of the children tested are all on the Alpha Bata subunit, and this both is in ML II and ML III. However the children with ML III in Israel come in on the Gamma subunit. We look forward to seeing the research that Dr Tiede will be doing with the knock out mouse models.

Hayden and Sarah have been diagnosed for 20 years now and I remember the Paediatrician saying to us way back then, that there will be a cure for your children in their life time. I think we are still a long way off in this respect but to see and be a part of what is happening now for this very rare and complex condition is incredibly exciting.

To have some understanding of their very significant bone conditions is a huge step forward and now, add to that the Natural History study and two knock out animal models that have been developed, I believe we will start to see some huge leaps forward in understanding Mucolipidosis.

Jenny Noble
Secretary
Lysosomal Diseases New Zealand

genzyme

Genzyme and NHS National Commissioning Group form a £7 million Partnership to support world class commissioning for rare diseases

The NHS National Commissioning Group and the biotechnology company, Genzyme, today announced that they have joined together in a £7m partnership to support a specialised system of care for patients with Lysosomal Storage Disorders (LSDs).

LSDs are rare, progressive, and often severe metabolic diseases that require specialised multidisciplinary expertise. The National Commissioning Group (NCG) has designated seven hospitals in England for the care of these patients: Royal Free Hospital, Great Ormond Street Hospital, The National Hospital for Neurology and Neurosurgery in London, Addenbrooke's Hospital in Cambridge, Birmingham Children's Hospital, Royal Manchester Children's Hospital and Hope Hospital in Salford. The Partnership will support the ongoing development of these centres to meet the needs of patients with LSDs such as Gaucher disease, Fabry disease, Pompe disease, and the MPS group of disorders.

Announcing the Partnership, Dawn Primarolo, Minister of State for Public Health said:

"We are delighted that we have been able to join in this Partnership with Genzyme for patients with lysosomal storage disorders and that we can jointly support patients living with these debilitating diseases. The Partnership will aid in sustaining our 'world class' commissioning of services for rare diseases through the NCG."

Professor Tim Cox, Professor of Medicine at the University of Cambridge, who was one of the first European physicians to treat patients with these diseases at Addenbrooke's Hospital, warmly welcomed the partnership. He said:

"These are not simply orphan diseases; they are equally marked by the pain and alienation associated with every longstanding medical condition. Without partnerships like this one, such rare diseases would remain marginalised in medical textbooks and patients suffering from them, forever neglected."

Paul Drohan, general Manager of Genzyme UK and Ireland said:

"Genzyme believes that the NHS has developed a highly effective model of care for patients with lysosomal storage disorders (LSDs) and that this is one of the best examples in the world of a universal healthcare system managing rare diseases. As enzyme replacement therapies are a key element of successful treatment of patients with LSDs, Genzyme is excited to have an opportunity to work closely with the NHS to support this service now and into the future."

LSDs are categorised as orphan disease since they are rare: they affect a small number of individuals, approximately less than 1 in 2000 population. In consequence they have had few options developed to treat them in the past. They are also difficult to recognise and diagnose.

To ensure equitable access the NHS in England has, since 2005, developed this 'world class' NCG commissioning structure, a commissioning model that is attracting interest internationally. Through this commissioning structure, the NHS has been able to meet the needs of patients with LSDs in England. Genzyme recognises the success of the LSDs Service and is committed to providing support for this model now and in the coming years.

Metabolic Networks

I want to thank all our members who participated in the ballot and wrote to their Members of Parliament on this matter.

On 7 September 2007 I wrote to Dr Keiran Morgan, Chairman of the Strategic Advisory Group for Metabolic Networks. The letter is shown below:

When we hadn't received even an acknowledgement to our letter on 11 October 2007 I wrote again resending a copy of the 7 September 2007 letter. To date, neither of these letters has been acknowledged, let alone responded to in depth.

I believe it has been suggested that the Metabolic Clinicians are now looking to set up their own mechanism for networking in England and Wales and this has to be welcomed. If this is the case, the defining issues should not be down to geography and laboratory budgets but to a patient-centred clinical approach to the management of all those affected by metabolic diseases where clinicians can learn from each other for the benefit of themselves and their patients.

I hope to be able to feedback further to you in the months to come. **Christine Lavery**

Dr Kieran Morgan
Bath and North East Somerset PCT Trust
St Martin's Hospital
Midford Road
Bath BA2 5RP

7 September 2007

Dear Dr Morgan

IMD Strategic Advisory Group
I am writing to you to further Adrian Pollitt's letter to you of 26 July 2007, following the BIMDG's decision to ballot all its members in respect of the geography of clinical networks for Metabolic Diseases, and the ineffective representation on the IMD Strategic Advisory Group of the metabolic groups and their stakeholders.

Stakeholders were finally emailed by CLIMB's representative on the IMD Strategic Advisory Board on Friday 8 June requiring responses by the 15 June attaching the three IMD documents. As I was on compassionate leave at that time the date had passed by several weeks when I saw this communication but in any case this was not sufficient time to begin to understand the issues and consult with our stakeholders.

When we saw the BIMDG briefing and ballot paper that had no consideration for stakeholders in respect of the patients and their families issued at the beginning of June we realised there was time to consult with our members and following consultation and then a ballot to our members we wish to advise you that there is no support for Option 1 or 2. In consultation our members suggested the following option was put forward:-

OPTION 4 - follow Specialist Commissioning Group (SCG) Specialist Health Authority Areas (SHA) with no changes to the proposed North and Central Network and with the following changes to the proposed South Network:-

South - not to take East of England and a separate Eastern Network established

South West patients to be accommodated in the South Network for administrative purposes with a commitment to continuing the MPS outreach clinics in Bristol and expanding these to Devon and Cornwall and establishing a South West Network Region at the earliest opportunity.

This option along with the three put forward by the BIMDG were put to our members and overwhelmingly Option 4 was the preferred option. The second choice was Option 3 as per the BIMDG ballot.

I should be most grateful if you would ensure that this feedback is taken into consideration when you make your recommendations. This is very important from our members' point of view as whilst samples can be posted children and adults with metabolic disease can not.

I fully appreciate the statements in Adrian Pollitt's letter but need to be sure that over time there is no scope for these networks to over rule natural or historical patient flows and because at a local level our families are not finding that Government policy giving choice of provider to patients prevails. Clearly within the Lysosomal Storage Diseases we are currently cushioned by NCG but this is a major concern.

Yours sincerely
Christine Lavery MBE
Chief Executive

FOR SALE!

Otto Bock Eco-buggy

As new, hardly used, a few minor scratches, instructions included, cost over £300, it comes with lap belt and net basket under the seat as standard.

Suitable for up to 50 kg

Also suitable for larger children

Lightweight and stable

Easy to fold to a compact size

Easy to navigate on all surfaces

Individually adaptable - foot rest height adjustable and reclining seat

£200 ono

Contact Julie or Paul on **0113 2614445**

The only limit to a dream is a child's imagination!

Making Dreams Come True...

Established in 1988 Dreams Come True is a national Charity with a simple aim - to lift the spirits of terminally and seriously ill children by fulfilling their most treasured dream.

We help children with both severe and life threatening illnesses, such as cancer, cystic fibrosis, cerebral palsy and MPS.

We want to hear from you…

A dream coming true can provide a positive focus for the children and their families and a release from the pressures and traumas of their illness, leaving them with precious memories.

The Magic of Disney

Swimming with Dolphins

Having a book published

Riding in a Bugatti car

Attending a film premiere

Going on Safari

Having a communication aid

Attending a sporting world cup

Having a photograph taken by a famous photographer

Each dream is unique and we always do our very best to fulfil each and every one.

We help children and young adults aged from 2 to 21 years old inclusive. If you know a child who may benefit from our help, please get in touch today. Initially, we require a letter (or e-mail) telling us about your child, their dream and medical condition.

Martin Plowman
Dreams Manager
Dreams Come True Charity
Knockhundred House
Knockhundred Row
Midhurst
West Sussex, GU29 9DQ
Tel: 01730 815000
martin.plowman@dctc.org.co.uk

dreams come true

React - Rapid Effective Assistance for Children with potentially Terminal illness is a dynamic charity working to improve the quality of life for children with life-limiting illnesses across the UK.

React exists to ensure that families who care for a life-limited child at home can provide whatever items are essential to make their lives as easy as possible, from replacing a broken washing machine to buying specialist equipment.

Funding is available for a wide variety of items, including domestic items, costly medical equipment, travel expenses, clothing, bedding, and much more. Requests are considered for any items of a 'basic or essential need' which will help you care for your child or improve their quality of life in some way. In order to qualify, you must be able to show that you cannot comfortably afford the item yourself and that (where relevant) funding is unlikely to be available through any other source.

Applying to React is designed to be fast and simple. Awards are made every day, and the charity replies immediately to each application.

If you would like an application form and more information, you can either:

Download and print one at **www.reactcharity.org/applications.php**
Ask your MPS Advocacy Officer or your nurse, social worker, etc. for a form
Call React on **020 8940 2575** or email react@reactcharity.org to receive one by post

React
St Luke's House
270 Sandycombe Road
Kew
Richmond
Surrey
TW9 3NP
react@reactcharity.org
Tel: 020 8940 2575

The **Regional Reform Project of Wheelchair Services** has been set up to examine how the Wheelchair Services can become more person-centred, responsive and accessible to wheelchair users.

For further information and/or comments, please contact the Project Manager, on or before the end of February.
Tel: 078 25 146511 or E-mail: lockhart@btinternet.com

The MPS Society would like to express their thanks and appreciation to the charity REACT. The advocacy team have approached this charity for funding for various items for our families, ranging from equipment for a soft play area to essential equipment, for example, height adjustable baths and washing machines. REACT have been fantastic in the speed of their response and the efficiency of supplying the cheque for the item(s) required.

We are sure the families who have benefited from the generous grants from REACT, are extremely grateful. Without charities such as REACT an enormous number of our families would struggle to obtain the necessary items/equipment.

If there are any families who are in need of funding for equipment, clothing or other essential items, and feel they would benefit from receiving a grant from REACT, please contact the advocacy team and ask for an application form. Once again, a big thank you to REACT.

Do you have a buggy which you could donate to a boy in Turkey suffering from MPS II, Hunter Disease?
He is 1.3 metres tall. We are looking for a buggy that is fully reclinable as this little boy is unable to sit up. We can arrange delivery. If you think you can help, please phone the MPS Office on **0845 389 9901**.

POSTAGE

Please ensure that you use the correct postage. As a charity we do not collect undelivered items due to the expense incurred.

All figures shown here reflect the new pricing system, which is based on size as well as weight.

LETTER FORMAT
C5+ or under. Up to 100g.
Less than 5mm thick.
Max weight 100g
Max dimensions 240 x 165 x 5mm
For example: Most letters, bills, statements, greetings cards, some brochures and catalogues

LARGE LETTER FORMAT
B4 or under. Up 750g.
Less than 25mm thick.
Max weight 750g
Max dimensions 353 x 250 x 25mm
For example: Letters containing unfolded A4 paper, most brochures, CDs and DVDs

Any questions please visit
www.royalmail.com

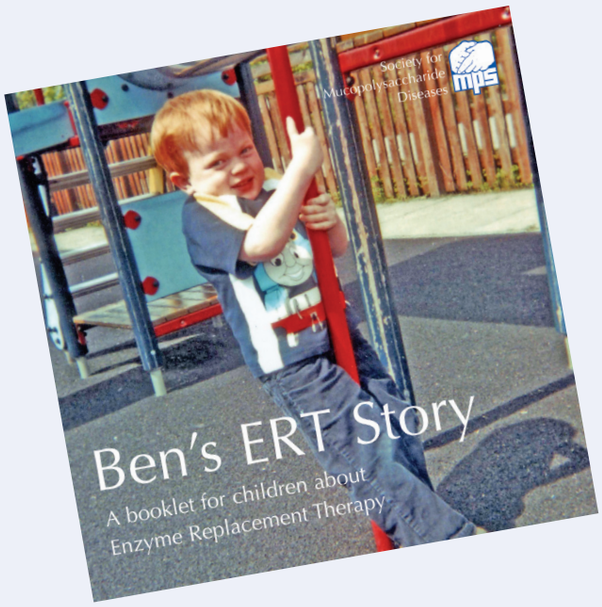
'Donate As You Spend' MasterCard

Donate As You Spend is an organisation founded to help smaller charities raise funds in ways only previously available to the largest charities. MPS are one of the first charities to take advantage of their new MasterCard.

Donate As You Spend Donations to MPS
£10 (plus Gift Aid £2.80) to MPS when you first use your card, 25p (plus 7p Gift Aid) to MPS for every £100 you spend on your card (N.B. balance transfers and cash withdrawals do not generate donations).
plus
A **voucher** for a FREE HOTEL BREAK FOR 2 with a choice of 200 hotels (subject to availability) once you activate and spend on your card three times!
Exclusive offers and discounts with your account statements
- Accepted in over 24 million outlets worldwide
- No interest to pay for up to 50 days
- 0% interest on Balance Transfers for 6 months
- 24/7 access to cash at over 56,000 ATMs in Britain alone
- Typical APR 16.9% variable

Ask us for an application leaflet now.
Subject to status. Applicants for credit must be aged 18 or over and UK resident. The DAYS MasterCard is issued by Sygma Bank UK, Equipoint, Coventry Road, Birmingham B25 8FE

New children's book out now!



Ben's ERT Story

Ben's ERT Story is a new publication from the MPS Society. It is a booklet for children about Enzyme Replacement Therapy and is written by Peter Conlin about his family's experience of ERT for his son, Ben, who has MPS IHS.

The booklet costs **£2.00**
(UK price inc. postage and packaging).
To order please phone 0845 389 9901
or visit **www.mpssociety.co.uk**

Carers Legal Rights in Employment

The work and families Act 2006 gives certain rights to families to request flexible working such as changing hours or working from home. From April 2007 the act extended the right for carers of a disabled adult to also request flexible working. This applies to all parents, carers, guardians, foster parents and holders of a residence order.

This means that employees who have worked for their employer for at least 26 weeks can apply to make a permanent change to their terms and conditions. However only one request can be made per year and although employers must consider any requests for flexible working they do not have to agree them if there is a good business reason why an application is refused. In these circumstances employees have the right to appeal. Your employer should have a policy regarding flexible working and may have their own application form to complete if you are wanting to make a request.

The act defines a carer as someone who cares for, or expects to care for, a husband, wife or partner, a relative such as a child, uncle, sister, parent in law, son in law or grandparent, or someone who falls into neither category but lives at the same address.

Under the Employment Relations Act 1999 employees gained the right to 'reasonable time off' to deal with any unexpected situations that arise in relation to their caring or parental roles. There are a number of different ways in which leave can be granted these are; carers leave, compassionate leave, borrowing or buying leave or taking a career break. At the discretion of the employer, time off can be paid. Situations where leave might be taken include:

A disruption or breakdown in care arrangements

If a dependent falls ill or has been assaulted or in an accident including when the victim is hurt or upset rather than physically injured.

To deal with an incident involving a child during school hours.

To make longer term arrangements for a dependent who is ill or injured.

PARENTAL LEAVE

If you are a parent with parental responsibility who is named on your child's birth certificate or adoption certificate you have a statutory right to take parental leave if you have a child under five years of age or a disabled child up to 18 years of age.

If you have one year's continuous service and you're an employee with a contract of employment you are entitled to 13 weeks of parental leave for each child under five years or 18 weeks for each disabled child up to 18 years.

Most employers will have their own set up for employees wishing to take parental leave, however if this is not the case the following applies:

Leave can only be taken in blocks of full weeks. Odd days should be taken as holiday or requests should be made to your employer to work flexibly. However, if your child has a disability, days rather than weeks can be used to take into account the possible need for hospital appointments.

Only four weeks leave can be taken for any one child and you must give your employer 21 day's notice. Your employer can also postpone any leave for up to six months if it will cause a disruption to the business. You are also able to carry over any unused leave to a new job but you cannot take this leave until you have been in their employment for one year.

CARERS ALLOWANCE

If you care for or look after someone who is disabled you may be able to claim Carers Allowance. You do not have to be related to the person.

To qualify you must be over 16 years of age and spend 35 hours or more a week caring for a disabled person. The disabled person must be in receipt of Attendance Allowance, Disability Living Allowance or Constant Attendance Allowance. However, you will not be entitled to receive Carers Allowance if you are in full time education with over 21 hours of supervised study or earn £95 or over a week, less reductions. It is also important to note that if you are in receipt of other benefits and make a claim for Carers Allowance, this may affect them or the total amount you are awarded.

For more information or to apply online you can either look on the website **www.directgov.co.uk** or apply to Carers Allowance Unit, Palatine House, Lancaster Road, Preston, PR1 1HB, Tel: 01253 856123

You may also wish to contact Carers Line
Tel: 0808 808 7777

Carers Line is Carers UK free advice line for carers. It is open on Wednesday and Thursday each week between 10-12 pm and 2-4 pm. **www.carers.gov.uk**

We now have an extended deadline for the Early Bird Rate.
Only £199 if you book before 1 February 2008!
Send in your booking forms now or phone us if you need a replacement!

Society for Mucopolysaccharide Diseases

Fun Weekend

Saturday 3 May and Sunday 4 May 2008