MPS

Society for Mucopolysaccharide Diseases

Winter 2006

Care Today, Hope Tomorrow

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

What is the Society for Mucopolysaccharide Diseases?

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

What are the aims of the MPS Society?

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

To bring about more public awareness of MPS and Related Diseases

To promote and support research into MPS and Related Diseases

How does the Society achieve these aims?

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

Telephone Helpline
Includes out of hours listening service

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

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

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

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

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

Information Resources
Publishes specialist disease booklets and other resources

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

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

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

MPS Awareness Day
Tuesday 15 May 2007

CARE TODAY HOPE, TOMORROW

Cover photograph:
MPS Adult Weekend - on the London Eye

MPS Magazine

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Newsletter Deadlines

Spring	1 Mar 2007
Summer	1 Jun 2007
Autumn	1 Sep 2007
Winter	1 Dec 2007

Become a Friend of MPS

CONTENTS

BBC Lifeline Appeal for the MPS Society

Sunday 19 November 2006

The MPS Society was very excited to have had the opportunity to take part in the BBC's monthly charity appeal programme 'Lifeline'. Running for 20 years, Lifeline has helped raise money for, and increased the profile of, hundreds of charities across the UK and abroad. There have been over 250 appeals so far and over £6 million has been raised. **www.bbc.co.uk**

The Society's appeal was broadcast on a number of BBC channels in November 2006 and so far has raised over **£10,000**. The MPS Society would like to thank everybody involved in the making of the programme and particularly Anthea Turner for agreeing to front our Appeal, Aisha Seedat (MPS IV) and her family, the Otway family and the Stevens family. We couldn't have done it without you!

CHIEF EXECUTIVE'S REPORT

Another year has passed and the MPS Society is fast approaching the start of its 25th Anniversary year. Amazingly, there are members reading this MPS Magazine that will remember the MPS Society's first attempt at a newsletter in autumn 1982. Over the coming year we will be bringing some of these stories alive, looking down memory lane and reflecting on how things have changed for those affected by MPS, their families, carers and professionals over the years.

Talking of time flying, it is a year since the Society moved into MPS House and what a difference it has made. Clearly the MPS staff team love their new working environment but what is quite amazing is how we have been able to use MPS House to widen the awareness of mucopolysaccharide diseases and the services and opportunities the Society offers. In the last twelve months MPS House has been the venue for major events including four high profile international events, the launch of Naglazyme for MPS VI, a European Workshop on MPS II, a European Bone Marrow Group meeting and a review of the research conducted to obtain a deeper understanding of experience of MPS I on ERT. In March 2007 we look forward to hosting the launch of Elaprase ERT for MPS II following its expected marketing approval in January.

As we enter 2007, the Society has a number of exciting events planned for our members. The bookings for the MPS Weekend Conference have been rapid so now that Christmas is over don't forget to book if you haven't already done so. Early Bird booking finishes on 28 February 2007 and no bookings for families will be accepted after 4 June 2007. For those 9 - 18 year old siblings who have or had an MPS brother or sister there is a fun-filled sibling weekend at Centre Parcs in Nottingham on 31 August - 2 September 2007. Please book now and give children the opportunity of a lifetime, sharing experiences with other brothers and sisters in similar circumstances, whilst taking part in activities that every teenager looks forward to.

As many of you know, the 'Ollie G Ball' which took place in September is heading for a record sum raised. The first £60,000 is to fund a PhD post in the MPS Stem Cell Biology Group at the University of Manchester, but wait for the exciting news to come... It is the wishes of the 'Ollie G Ball' organisers that the money raised also benefits families caring for MPS children and adults. Enclosed with this magazine is a form to apply to be one of the MPS families selected to go to Disneyland Paris in May or June 2007, so if you would like to enjoy the magic of Disney with the MPS Society with the cost of travel and accommodation funded, please complete the entry form in the MPS Magazine.

Returning to the present, we are reminded that Christmas and the New Year may come with mixed blessings for our members. On behalf of the Trustees and MPS staff team we send our best wishes for 2007 and thank you for all your support throughout 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 6-7 October 2006.

Governance

The Chairman confirmed that Professor Bryan Winchester would be attending the meeting on Saturday 7 October. The Board of Trustees, having agreed to make Professor Winchester an Honorary Member of the MPS Society, expressed their appreciation of Professor Winchester's decision to join the MPS Board as a Trustee with a particular remit of advising on scientific issues facing the Management Committee.

The Society's Risk Register was reviewed and it was agreed no changes were required at this time. Eleven policies were reviewed and agreed without amendment.

Personnel

The Chief Executive reported to Trustees that Ashley Siberini and Maureen Cummins were leaving their employment with the MPS Society for personal reasons and that Cheryl Pitt remains on maternity leave. Having had prior Trustee approval, the Chief Executive confirmed that Neisha Hall will start her Social Work Training course in February 2007. Approval was also given for Steve Cotterell to be offered the same course to start in October 2007.

Overseas Collaboration

The Chief Executive reported to Trustees on the Society's interaction with overseas organisations and in particular the Inborn Errors of Metabolism meeting in Japan and the Australian MPS Society meeting in Brisbane in September.

Generating Funds

Trustees fed back on the second 'Ollie G Ball' and how well it had gone. Trustees were advised that the final figure is likely to be in the range of an amazing £164,000. The Management Committee asked for their thanks and appreciation to be passed to the organisers. The Trustees agreed that there is a need to continue to widen the scope of where the Society generates funds and particularly welcome the MPS BBC Lifeline Appeal that went out on 19 November presented by Anthea Turner.

MPS Research Grants

Trustees received feedback on the progress being made by the six major research grant recipients and were advised that the MPS Society should be in a position to award a further grant(s) in February 2007.

Ollie G Ball

The second Ollie G Ball was held on Saturday 16 September 2006 at Whithorn Farm in Surrey. The Ball was organised by David Gosling (father of Oliver Gosling, MPS I) and Countrywide Special Events in aid of the MPS Society. The night was an amazing success with nearly 600 guests attending the event, 100-strong crew, 20 miles of cable, 200 lights, 4696 pieces of cutlery, 3750 pieces of crockery, 5120 glasses, 725lbs of food and over 2000 square metres of marquee.

On the night it was announced that they had raised £164,000 but the final figure has yet to be confirmed. The money raised will go towards research into MPS and Related Diseases, and in taking MPS families to Disneyland in Paris. If you are interested in taking part in this trip please complete the enclosed form and return it to the Society.

The MPS Society would like to thank everyone involved for their support.
Photo top: David Gosling, Photo below: Anthea Turner and Grant Bovey

ANNOUNCEMENTS

New Members

Mr and Mrs Robinson have recently been in contact with the Society. Their son Harry has a diagnosis of Hunter Disease. Harry is five years old. The family live in Hampshire.

Your letters - We are always pleased to receive letters from all readers of the MPS Magazine and especially our members. We welcome letters on any subject and your views and comments would be very welcome.

Many congratulations to the family of **Jack Atkinson**, MPS II. Jack has a new baby sister called Isobel.

Deaths

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

Thomas Flaig who suffered from Sanfilippo Disease and who died on 23 September 2006 aged 12 years.

William Ferrier who suffered from Sanfilippo Disease and who died on 26 September 2006 aged 17 years.

Kerry Parker who suffered from Sanfilippo Disease and who died on 21 October 2006 aged 14 years.

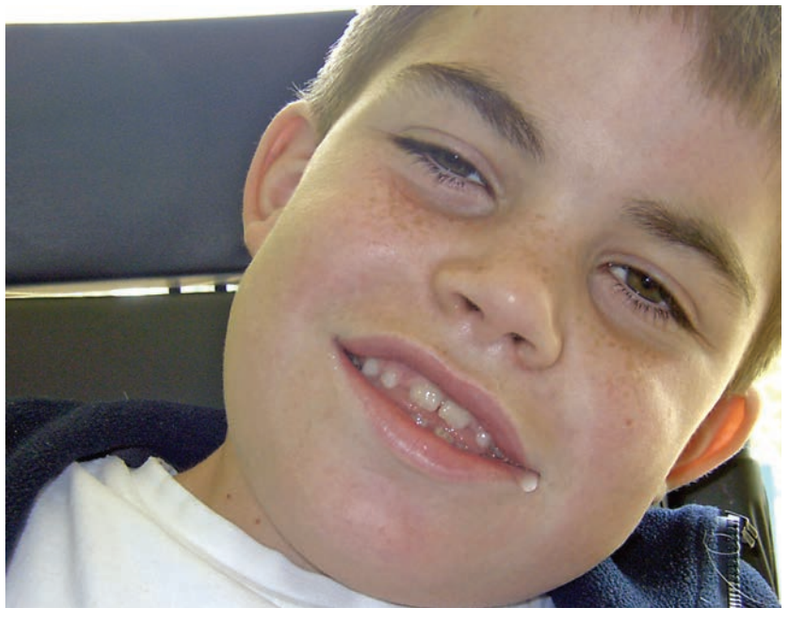
Kyle Shields who suffered from Sanfilippo Disease and who died on 13 October 2006 aged 15 years.

Jack Onion who suffered from Hunter Disease and who died on 26 November 2006 aged 11 years.

Lorren Damen who suffered from Sanfilippo Disease and who died on 2 December 2006 aged 13 years.

2007 Dates for your diaries!

9 January	Bristol MPS Clinic
18 January	MPS III Clinic, GOSH
19 January	BMT Clinic, Manchester
8 February	Newcastle MPS Clinic
17 March	Cadbury World
3 April	Bristol MPS Clinic
20 April	BMT Clinic, Manchester
28 April	South East Get Together
May - June (tbc)	Disneyland, Paris
29 June - 1 July	MPS National Conference
10 July	Bristol MPS Clinic
27 July	BMT Clinic, Manchester
31 Aug - 2 Sept	Sibling Weekend
2 October	Bristol MPS Clinic
19 October	BMT Clinic, Manchester



In Memory of Thomas Flaig

Our little boy Thomas died aged 12 at the end of September. Thomas had MPS IIIA and Duchenne Muscular Dystrophy. We were told that he was the only child known with this combination of conditions.

The last years of Thomas's life were difficult but looking back we know that his short life was a happy one. We all have lovely memories of a beautiful smiley boy - always on the go. Thomas gave so much happiness to his family and friends, and enriched our lives by bringing us into contact with many special people. Andy and I, and Thomas's brothers, Joseph and Oliver, feel privileged to have been part of his life and wouldn't have missed it for the world.

Cathy Flaig

Professor Bryan Winchester retires

Professor Bryan Winchester celebrated his retirement with a retirement symposium 'The Lysosomal Storage Diseases from Farms to Pharmaceuticals' at the Institute of Child Health on Friday 29 September 2006.

Bryan was educated at Sutton Grammar School in South London and studied chemistry at Queen's College Cambridge. In 1963 he returned to London to study for a PhD in the Biochemistry Department of University College. Bryan's research was directed mainly towards the glycoproteinoses, in particular Mannosidosis and Fucosidosis. Bryan spent a year in New Zealand studying bovine Mannosidosis and returning to London continued to pursue his interest in Lysosomal Storage Diseases. He set up a screening programme in connection with the English Springer Spaniel Society for heterozygotes with Fucosidosis, even being invited to Crufts one year!

In 1988 Bryan was appointed a senior lecturer in clinical biochemistry at the Institute of Child Health and Great Ormond Street Hospital with part of his remit to introduce molecular analysis for Lysosomal Storage Diseases (LSDs). This he successfully did working on the x-linked Fabry and Hunter Diseases and then on other autosomal LSDs. Bryan was appointed Reader in Biochemistry in 1991 and Professor in 1996 but most importantly he was committed biochemically and molecularly to doing his best to make a difference for MPS families. No one could or can today take away the pain of an MPS diagnosis but Bryan and his team developed a first class biochemical service that enabled families to get a rapid diagnosis once an MPS disease was suspected and enabled hundreds of MPS families to have a pre-natal diagnosis on successive pregnancies.

Bryan retires having been the grant holder for six MPS research grants that started with developing gene mutation analysis back in the 1990's and finished with a three year project carried out by Clare Beesley on Biomarkers.

Whilst in the scientific community for LSD's and rare diseases Bryan will be sorely missed, it is the Society's good fortune that Bryan has agreed to become a Trustee and Scientific Advisor. We are all delighted to have Bryan on board at a time when the research applications for MPS Grants are increasingly more complex than ever.

Following Bryan Winchester's retirement symposium, a reception was held in his honour. During the reception tributes were paid and the MPS Society's Chairman, Barry Wilson, made a presentation of a glass decanter engraved with the Society's logo to Bryan.

Tribute by Lucy Lavery

I would like to think that I am representing all of the members at the MPS Society who if it were possible would like to have been here in person to celebrate Professor Bryan Winchester's retirement and therefore on behalf of all the MPS families you have helped over the years, a huge thank you.

On a more personal level, my brother Simon was diagnosed with MPS II, Hunter disease in 1976 at the age of 18 months. Although I was not born until 1986, I know that I, my two brothers and hundreds of other siblings of MPS sufferers have entered this world after Professor Winchester's team declared us free of MPS. One of my brothers even did work experience with Professor Winchester but would you believe it - he ended up pursuing a career in the media!

However as a student of Human Genetics, I know that Bryan and others like him have greatly influenced my life before birth and after.

MPS Awareness Day
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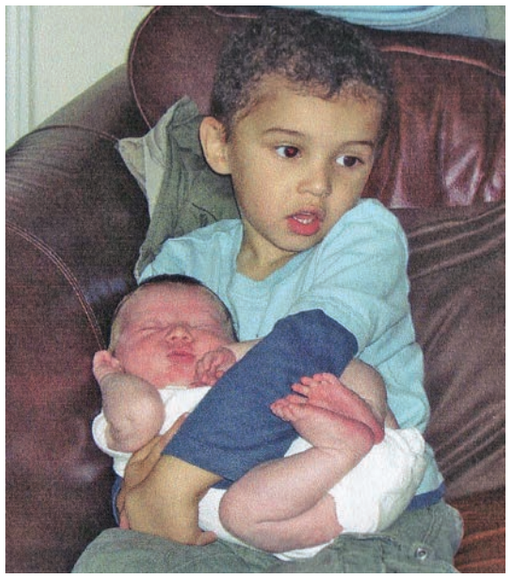
Congratulations to...

Pam Hughes recently wrote to tell us that Louise had her first baby on 8 October 2006, a little boy called Thomas Christopher. He weighed in at 7lb 4oz and is a lovely baby. We are all very proud of him. Special mention was made of Dad, Chris, who was brilliant through quite a difficult labour. The photo below shows Thomas being well looked after by his cousin, Jozef. Louise is sister to Daniel (MPS II), who sadly passed away on 29 December 2001.



Dear MPS Society

We are writing to thank you for the help you gave us in applying for and getting Isaac's tricycle, which arrived recently. The trike is fantastic and Isaac loves it more than anything else he owns. It has given him so much joy and independence.

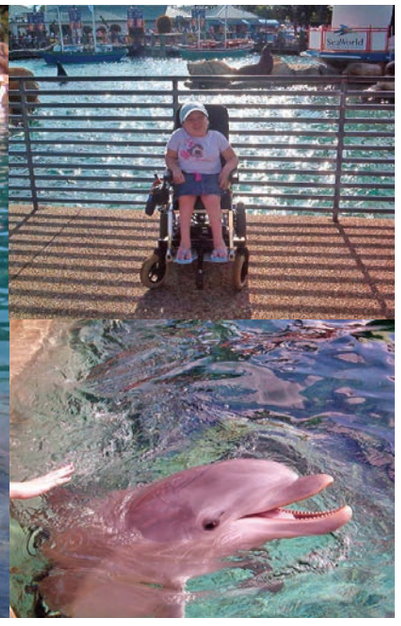
Here is a photograph of Isaac on the trike. Thank you again for the help you gave us. Think of us as we spend the winter standing in the street shivering whilst Isaac practices reversing, three point turns and going around the block!

Yours sincerely

Adam, Louise,
Isaac and Eliza Turner

Sinead swims with the dolphins

Here are just a few of the pictures that were taken at Discovery Cove and Seaworld in Florida. Sinead won the category of Children of Courage in the local newspaper's New Year Awards last February. Her sponsors Extec which have a factory local to the family in Swadlincote paid for Sinead, her mother and her mother's partner, Andy, to go to Florida on a dolphin swim as this has been Sinead's dream for years. Sinead was over the moon and loved the experience. They would recommend it to anyone. Strangely enough Sinead found the humidity in Florida made her breathing easier and didn't need to use her inhaler at all. The family went in October and it was very, very hot then.



Congratulations to Sophie and Phil!

Many congratulations to Sophie Denham, now Sophie Thomas, Senior Advocacy Officer at the MPS Society. Sophie was married on 30 September 2006. With best wishes to Sophie and Phil for a long and happy future together, from all at the MPS Society.

Congratulations to...

The MPS Society would like to wish Byron and Lynn Wibberley all future happiness. Byron is Forest Ranger at Sherwood Pines and works closely with the Society to oversee the Childhood Wood. Byron and Lynn got married this summer and spent their honeymoon in Australia. Many congratulations to them both.

Samuel Stevens Starts School!

On 11 September 2006, Samuel (MPS II) made his first journey to 'Big School'.

He was a very proud boy as he donned the same smart uniform as his brother, Oliver. With his Power Rangers lunchbox and red school book bag, he happily marched alongside his Daddy with Mummy capturing the moment on camera (bravely running backwards to try and get the right shot!).

Samuel was turned down in a pre-statementing assessment and has joined the mainstream children, so he is very happy to be with the friends he had at nursery, although he would dearly love to do horse riding and swimming like Oliver does in the Special Needs Unit. You can't win them all!

The teachers are very happy with him and to my amazement when I visited the classroom a few weeks into the term, I found he had 7 stickers on his sticker chart! I asked the teacher if the children put the stickers on themselves, only to be informed that Samuel had got a sticker that very day for missing his whole playtime because he was tidying up… That might have been the point when I asked if they took boarders!

Something tells me that Samuel, a self confessed 'cool dude' who has just turned 5, will get along at Big School just fine. While he is oblivious to Hunter's, he is looking forward to having a magic medicine which will help him run as fast as his friends, but in the meantime, his sunny little face with the cheeky smile will make sure his friends are always close by.
Claire Stevens

MPS Wins Fight for Hayleigh's Treatment

Robert and Heather Reynolds, supported by the MPS Society, won a three-year battle to get ERT for their daughter, Hayleigh, who has MPS I Hurler Scheie Disease. Although Hayleigh met the Department of Health Guidelines for accessing ERT for MPS I in England in 2004, and even though another similarly affected child in Scotland was on treatment, Hayleigh was made to wait for her condition to deteriorate considerably before, following a judicial review, the Greater Glasgow Health Board were forced to reconsider their decision.

On learning of the decision Mr Reynolds said "As time has gone by Hayleigh has got a lot worse. She has had to have an operation a couple of weeks ago to drain fluid from behind her eyes, to ease the pressure. You can't help wondering if that would have been necessary if Hayleigh had been on ERT much earlier."

Editor's Note: This was a very important case to bring as Hayleigh was the only child in the UK with MPS I Hurler Scheie Disease who wanted to be on treatment, but denied ERT. We are delighted for Hayleigh and her family that the court found in their favour. This case was also important in that there are at least six MPS II members in Scotland who are also looking to access treatment when Elaprase is licenced this month.

Statement by Greater Glasgow Health Board

In keeping with the commitment given by Greater Glasgow Health Board at the recent Judicial Review involving Hayleigh Reynolds, the Clinical Review Group has now met to consider the case for prescribing laronidase to this patient.

The Group notes that Hayleigh now meets four of the five inclusion criteria used by the Department of Health in England to assess eligibility for treatment with laronidase. In 2004 she met only two. Although we feel that the overall evidenced health benefits of laronidase are modest, in keeping with the Scottish Medicines Consortium's view that laronidase is not a cost-effective treatment, we believe that there are particular circumstances associated with this case that lead us, exceptionally, to recommend that Hayleigh be offered treatment. This recommendation is made on the understanding that Hayleigh's response is monitored closely using the minimum schedule of assessment for treatment with laronidase and that her treatment will be discontinued if any one of the exit criteria outlined in the National Specialist Commissioning Advisory Group document 'Guidelines for the Investigation and Management of Mucopolysaccharidosis Type I' are met.

Press Statement on the decision of The Greater Glasgow Health Board in respect of Hayleigh Reynolds on behalf of the MPS Society

We are delighted following the recent judicial review brought by the family and supported by the MPS Society that The Greater Glasgow Health Board has today granted funding for Hayleigh Reynolds to receive Enzyme Replacement Therapy (ERT), Laronidase at The Hospital for Sick Children, Yorkhill. This decision clearly comes as a huge relief to Hayleigh's family and after nearly a three year wait for a treatment that is routinely available to children with Hurler Scheie, who meet the clinical criteria in England.

At last Hayleigh Reynolds has a chance of fighting this degenerative and life limiting disease and preventing further deterioration that would have resulted in a very early death. **However, the MPS Society is deeply concerned that the statement made by Greater Glasgow Health Board stating that 'Hayleigh now meets 4 of the 5 inclusion criteria used by the Department of Health in England to assess eligibility for treatment with Laronidase' is at best misleading to the public at large and suggests that Hayleigh needed to meet the 5 criteria before treatment should start when in fact only one of the five is required for funded treatment in England.**

The Department of Health guidelines clearly state that 'patients with Hurler Scheie, who have little or no cognitive impairment, as in the case of Hayleigh, with any of the five symptoms should be considered for immediate treatment.'

Therefore Hayleigh qualified for treatment under the Department of Health Guidelines at the time of her diagnosis over three years ago and it should be little comfort to anyone that she has had to wait nearly three years for her condition to deteriorate to such an extent that she has lost skills and undergone major surgery that may have been preventable had she been started on treatment much sooner.

Laronidase was approved by the EMEA in June 2003 and is the only medical treatment available that has shown efficacy in the treatment of Hurler Scheie Disease. It has been routinely available and funded for all children affected by MPS I Hurler Scheie and Scheie Disease, meeting the clinical criteria in England and Northern Ireland since this time. In Wales patients are treated on a named patient basis.

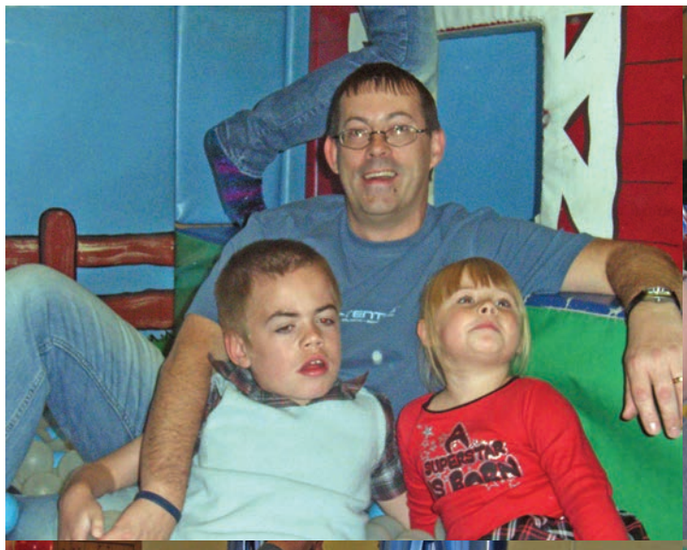
Over 40 children and young adults in the MPS Society are on this treatment and showing considerable benefit to the extent that they can now fully participate in every day activities that we all take for granted.

Scottish Christmas Party at Almond Valley

9 December 2006, the day was off to a good start, the sun was shining although quite cold. All families congregated in the soft play area and I thought it would be a great photo opportunity to get some action photos of the children, and grown up children, playing around. Little did I expect to get pelted with the balls from the ball pool, some I think were thrown by the parents but blamed on their children (a likely story)!

Lunch was in the next room and the children tucked into their party food, pulled their crackers and made a bee-line for the balloons. This was shortly followed by the ringing of a familiar sounding bell, and in the next instant Father Christmas made an appearance, and gave out the presents he had carefully chosen for the children. After all this excitement we all made our way outside for a leisurely trip on the tractor, some ventured onto the train, and we rounded the day off with a run around in the Adventure Zone.

I hope everyone had a great time, and a huge thank you to the staff at Almond Valley for yet another great Christmas party. I would also like to thank Santa's little helper for ensuring the children got the presents they deserved. Here are a selection of photos from the day. **Neisha Hall**



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**

MPS Awareness Day
Tuesday 15 May 2007

MPS Adult Weekend in London

The London trip was a brilliant experience for me as it proved to myself that I could go on a plane to fly to London and be very independent e.g. sort out assistance for myself at the airport, sort out my room at the hotel and get up in the morning, which normally I don't do by myself. Now I know that I can cope on my own and fend for myself. It was a great and brilliant weekend that I needed to do.

I arrived at London City airport on Saturday 11th November, where I was picked up by Ben Lavery who I haven't seen in a while. As I got to the airport the 2-minute silence was just about to start, so we stopped to wait until it had finished. When it had finished we went for a taxi and we were off to the Hilton Tower Bridge Hotel. We arrived at the hotel where we met up with Steve and Neisha who were in charge of the weekend. Everyone arrived and we all were discussing what to do next so we went for lunch.

After we had lunch we went to investigate the bus tour to see if we could go on it and over the road was the London Dungeon. When I saw the London Dungeon I wanted to go in and see what it was all about. So Ben and I went in and the others went to Harrods.

Whilst we were in the London Dungeon we had a photograph taken of us, Ben in the Stocks and me with an axe trying to chop his head off. Then we went round and we found that we had to wait in a queue for a while before continuing into the Dungeon. Well, I think if I type everything that happened in the London Dungeon you would get bored so I will just tell you my best bits. My first bit was going into a room, which was full of mirrors, but it was a maze, it was so confusing. The funny thing was that Ben was using me as a battering ram into the mirrors so he wouldn't bash into them but I was. My second bit was going on the boat ride, it was pitch black and we couldn't see anything, it went forwards slowly but when it went backwards it went fast, it was so exciting. The third bit was when there was a court bit which was good because the women picked on me and put me in a jury trial, it was so funny.

We went back to the hotel after the London Dungeon to have a rest before we met up with everyone to head for the Hard Rock Café.

We got to the Hard Rock Café but on the way we went down the mall and went past the Palace; guess what, the Queen was in, I wanted to go and have a nice old cup of tea with her but we didn't because we were going to the Hard Rock Café! We went in to the Hard Rock Café and it was so loud I couldn't hear my own voice, the waitress showed us to our table and we discussed what we wanted for starters. In the end we all decided to have a mix of food so we can have a bit of everything. The starters were gorgeous and I was a bit full but we all still hadn't had our main meal yet.

I could just about hear the conversations we were having, but I still had a great time. Instead of telling you what I had to eat I will tell you the most embarrassing moment of the trip was, remember this is on a Saturday night and my birthday was a week later on 19th November... We could see people with balloons and we wanted some so Neisha asked the waiter for some balloons and he said whose birthday is it and guess what, they all made out it was my birthday, so he went to the kitchen and he came back with a small bowl of ice cream with a candle in it. I was so embarrassed, but they got the balloons they wanted but apart from that it was a great night.

On the Sunday I had to get up early - unlucky me! Everyone met up at 8.30am to go and have breakfast; now that was really nice. When we all finished we went to our rooms to get ready to go to the London Eye and the River Cruise with a 3-course meal. We all met up in reception, so we could get taxis to take us to the London Eye. We got to the London Eye and all of us were so excited with anticipation. Eventually it was our turn to get on, we went on and it was so fantastic to see the view of London as we went higher and higher. When the ride finished we got off and went down to look at the photograph that was taken,

as we looked at the photo, Big Ben rang and that was the start of the 2-minute silence. We had about half an hour to spare until we went on the River Cruise.

We got on the River Cruise and it was like you were going into a restaurant, it was that posh. The Cruise started and it was so nice because it was like you weren't even moving. The funniest thing that happened on the Cruise was when it was time for our pudding and on the menu was apple pie and custard, but I don't like the apple pie so I thought I would ask for just custard. So I did and the waitress was looking at me like I was stupid, I don't think she ever heard of anyone having just custard before. The waitress got me custard and as she brought me the bowl of custard she went away giggling. It was so funny! I really enjoyed myself, as it was so nice to be with people that I really liked. It was a brilliant experience.

When the River Cruise finished we all went our separate ways as the others were going home. My flight was at 8.40pm so Ben and I had plenty of time to do whatever we wanted to do - we just went sight-seeing and eventually ended up at Hamley's.

The day ended so I had to go to the London City Airport to get my flight home. It was nice to get away from home to spend time by myself and to socialise with new people. I would like to say a BIG THANK YOU to the MPS Society for making the London trip happen.

David Oulton (MPS II)

Editor's Note: The Society is most grateful to the Barchester Healthcare Foundation for giving the Society a grant for the MPS Adult Weekend.

Family Day at Twycross Zoo

On a cold, but dry October day Steve and I travelled up to Twycross Zoo in Staffordshire to meet up with six MPS families for a family day at the world primate centre.

As we arrived all wrapped up warm we were met by the screeching playful sounds of the monkeys swinging in their cages and playing with their friends. As the families arrived we greeted the families with a map of the zoo and money for lunch. Allowing the families to wander around the zoo at their own leisure we arranged to meet up at 12:30 for lunch.

The zoo was great; it was fully wheelchair accessible with a wide range of animals to see and activities to take part in. The main attraction at Twycross are the monkeys, however they have so much more to see and do such as Giraffes, Elephants, Lions, Parrots and more besides.

At 12.30 we met with the families in the café, it was very busy so we sat outside. Although the weather was still very fresh it gave the opportunity for us all to mingle giving both the families and staff the opportunity to catch up from previous meetings or form new friendships. Most of the families had said that the monkeys were their favourite and how could they be resisted, they were so funny to watch and their behaviour is so similar to ours! After lunch and a chat families then went off to do their own thing and see the animals that the others had seen and talked about.

At 3pm we then met again for a drink. It was a really good day, families were given the opportunity to meet up and both adults and children were entertained by the animals of all different shapes and sizes. Those families local to Twycross were discussing the next time they would return. Steve and I then said our farewells and headed back. The day was a great success and although it was cold at least the rain held off. **Charmaine Scott**

Photo page 14: Aaron Craig (MPS III) and family. Photos this page clockwise from top right: Charmaine Scott and Aaron Craig; MPS families at the Zoo; Bethany Allen (MPS III); Thomas Coney (MPS VI) and family.

MPS Regional Clinics

Birmingham Clinic

On 26 October 2006, Birmingham held its second MPS clinic for this year at the Victoria Special School. The weather as always was very wet, something that seems to be a regular trait whenever I visit Birmingham for a clinic. We saw a number of families at this clinic and the hospital staff team are very good ensuring that there is hospital transport for those who have difficulty accessing the clinics. As always the school provided us with the space and parking needed but being offsite it is difficult to arrange any follow up medical care that is sometimes needed.

The clinic ran smoothly and it was great to catch up with the families who attended their appointments. Our thanks go to Dr Chris Hendriksz and Joy Hardy for all their hard work and to all the other professionals who were a part of the clinic.

We have recently heard through Dr Hendriksz that they have been afforded two clinic rooms within the hospital. This is great news as the clinics can now re-locate back to the hospital and follow up care will be a lot easier. It is also hoped that the clinics being back at the hospital will also allow other medical professionals to be present, which will hopefully help reduce the number of appointments a family have to attend. They have currently secured representation from genetics and neurosurgery and are in the process of confirming cardiology, orthopaedics, dental and ENT. **Sophie Thomas**

Photos clockwise from top right: Birmingham Clinic - Tariq Mahmood (MPS IV), Bristol Clinic - Louis Quant (MPS III), Archie and Isaac Newton (MPS IV), Cardiff Clinic - Gavin and Sarah Hyde (ML III)

Bristol Clinic

Tuesday 31 October, and Steve and Neisha were on the road, well on the M4, heading towards Bristol. We made it in good time and managed to get a cup of tea before the clinic started. The appointments ran to time and it was lovely to meet everyone.

We laid out an array of biscuits and cakes, and it was all I could do to keep Steve from eating them all! Luckily the children did get a look in and the chocolate fingers were a great hit.

All in all the clinic went extremely well, and Steve and I would like to thank all the families who attended, the staff at the Children's Development Centre at Frenchay Hospital for their hard work in setting up the clinic, and of course a special thank you to Dr Jardine and Dr Ed Wraith who without their continuing commitment and support, this clinic would not be possible.

Northern Ireland

On 16 November 2006 Dr Fiona Stewart, Dr Ed Wraith and the MPS Society held the MPS clinic at Antrim Hospital Northern Ireland. The clinic was well attended and it was a good opportunity to meet with both new and existing members, we have certainly picked up a lot of advocacy work to take forward! We would like to thank Dr Stewart, Dr Wraith and all those involved in helping to co-ordinate the clinic. The next clinic will be in 2007, dates to be confirmed.

BMT Clinic Manchester

On 17 November 2006 Dr Ed Wraith and the MPS Society hosted a BMT clinic at the Royal Children's Hospital in Manchester. The clinic was very busy with lots of children running around and playing together, they certainly kept their parents, Steve and the Willink staff on their toes! We would like to thank all those involved in organising this clinic in particular Dr Wraith, Jean and Dot.

Cardiff Clinic

The clinic at Cardiff was held on 24 November 2006 at the University Hospital in Cardiff. Unfortunately Dr Ed Wraith was unable to attend on this occasion and sent his apologies (he had to jet off to Norway). Dr Shortland had a nearly full day of appointments, which ran pretty much to schedule. It was a pleasure to meet with all the families in attendance and I hope that you found the opportunity to meet with the doctors and MPS Advocacy Staff worthwhile. I would like to thank all those involved in organising the clinic particularly Dr Shortland.

Photos clockwise from top right: BMT Clinic - Rachel Rothwell (MPS I BMT) and Charlie Escalonilla (MPS I BMT), Melissa McKie (MPS I) and family, Northern Ireland Clinic - Aaron and Dean Doherty (MPS III), Brigid McDonagh (ML II) and family

Annual Planting

On Friday 20 October 2006 nine families attended our annual tree planting at the Childhood Wood, Nottingham to plant trees in memory of those who had lost their lives to an MPS or related disease.

The day started off very wet and we all hoped for a break in the clouds when it came to the tree planting. While we all gathered at the Clumber Park Hotel for a drink and the buffet lunch, the weather did just that and we had blue skies and sunshine. Christine Lavery and Judith Swann, who represented the Lord Lieutenant of Nottingham, gave a welcome speech to all the families who were participating in the day and invited them all to follow in convoy to the Wood.

We were met at the Memory Board by Paddy Tipping MP, who welcomed everyone to the Wood and shared with us all his reasons why the Wood is so special to him. Judith Swann then read out the names of the children and adults who were being remembered at the Wood on the day. Wilma Robins, one of our trustees then read the poem 'Remember' before the families each collected their oak saplings, planted them and released a balloon in memory of their loved one.

Judith Swann and Paddy Tipping MP planted the trees for those families who were unable to attend the Wood and Byron Wibberley, Forest Ranger, and his team were on hand to help with the planting as needed.

As this was my first event in my post with the MPS Society I was very nervous and hoped that the day would run smoothly, but my nerves eased once the families arrived and the sunshine appeared. It was an emotional but enjoyable day, an event that I will never forget. The Childhood Wood is a lovely place, within beautiful settings and a pleasure to visit. **Charmaine Scott**

Photos clockwise from top left: the Shah Family; the McLaren-Hall Family, Paddy Tipping MP welcoming everyone to the Wood; the Shaw Family

Pervivo Park and the Childhood Wood

Pervivo Park is the metabolic patient organisation (BOKS) in Belgium's answer to the MPS Childhood Wood. On the top of a hill not too far from Belgium's capital, Brussels, is a special memorial to BOKS children who have lost their lives to a metabolic disease. The focal point is a wooden structure and the carvings of two children. Around the monument are planted young trees.

I must admit I had never heard of Pervivo Park until contacted by Michel, Josiane, Erica, Jens and Marc because they wanted to visit the Childhood Wood. On 28 October 2006 we met up with the Belgians and gave them a tour walking from the car park on the white route and doing a circle of the Wood. We lingered long at the Memory Boards and this impromptu visit enabled us to check on the trees planted a week earlier by the MPS families.

Thankfully Marc, Josiane, Erica, Jens and Michel spoke perfect English and were able to discuss the aims of Pervivo Park and the Childhood Wood. There are many things we do similarly like the annual planting and balloons and after a most fruitful coming together we all had food for thought. **Christine Lavery**

We are delighted to announce that the Memory Boards at the Childhood Wood are now in place!

Pervivo Park

Memories of Kyle

When our third son, Kyle, was born on 18 October 1992 we were thankful that we had another healthy son to complete our family. It transpired that our illusions were to be shattered four years later when Kyle was diagnosed with MPS III Sanfilippo Disease. It was a relief to know that there was a reason for his delayed speech and failure to 'potty train' never mind the severe behavioural problems. However, we were also devastated to learn that our extremely active, boisterous four year old would only have a life span of about fourteen years.

Those early days now evoke many happy and funny memories although at the time they were often a source of extreme embarrassment. This included Kyle eating the contents of ash trays in hotels, capsizing plant pots in waiting rooms, attempting to empty the shopping trolley as quickly as I filled it and lobbing items in all directions regardless of who was in the firing line.

As the disease progressed Kyle was no longer the uncontrollable child he once was. He lost his speech and the ability to run, jump and walk. He started to have seizures, his swallowing deteriorated and following a videofluoroscopy he had a gastrostomy in August 2004. Throughout this period Kyle still remained a happy little boy who enjoyed and loved attention and would often smile and laugh as his brothers entertained him. Kyle retained his love of music although his taste mellowed over the years and he greatly enjoyed his multi-sensory station, as the musical vibrations and watching the fibre optics relaxed him.

Over the last few months Kyle's nursing needs increased as he became more prone to chest infections and was too ill to attend school. We are grateful for the constant stream of carers who were a great support to us and gave skilled professional care to Kyle. Many of these went beyond the call of duty and were willing to be contacted at any time of the day or night and to those we will be forever grateful.

Kyle was admitted to Horizon House on Tuesday 10 October for planned respite and passed away peacefully with his family present in the early hours of Friday. The funeral took place on Sunday 15 October with approximately 500 attending, an indication of how much Kyle was loved by all who knew him.

We feel tremendously privileged to have been Kyle's parents. Although his life has been so short it has impacted on the lives of so many others. His brothers and us have so many recollections and momentos gathered over the years which are deeply valued and will remain as treasured possessions and memories.

We recognise that we will encounter difficult days ahead as we attempt to come to terms with the huge void that Kyle's passing has left in our lives. We are thankful that our trust is in the Lord who has sustained us throughout Kyle's life and in our recent loss. We are assured of His faithfulness and know He will give us the strength to face each new day.

Kyle touched our hearts, changed our lives and will always be a part of us. **Alison Shields**

God said that you were getting tired
And a cure was not to be,
So he wrapped His arms around you
And whispered "Come with Me"
Your weakened heart stopped beating,
Pain free and now at rest
Although our hearts are breaking,
We know God only does what's best

The value of respite care

I am writing this piece in the hope that all MPS families are aware what respite is and hope they are all getting what they are entitled to allow them to function as normally as possible with a child affected by MPS in their family.

I recently found a letter in the loft whilst busy clearing it out. The letter made me laugh, but at the time I was close to the end of my tether. My husband, Andy, was in the Royal Navy and spent a lot of time at sea, so we used to correspond regularly by letter (in the days before mobile phones were popular). I have included several paragraphs from one of these letters below. This was before my son, David, was diagnosed with MPS. I challenge you to recognise which condition he has.

The letter began with an apology to Andy for my inhospitality on his weekend leave, but life just seemed so difficult at the time. It goes like this...

“My mood then was nothing compared to the last ten days. I’ll start at the beginning. On Monday, after you had gone back, David finally got back to school only to come home poorly at 3.30pm. As the evening went on, he got worse. At 9pm I called the doctor. At 9.45pm the doctor arrived and announced it was an infection, but not the usual right ear, it was his left one. More medicine. As it was gone 10pm before I got him to bed, I decided to keep him off school, give the medicine a chance to work and send him back on Wednesday. I couldn’t believe it, the school rang to say the heating had broken down and that the school would be shut on Wednesday. So I sent him back on the Thursday and broke up for half term the same day.”

“Well, that’s when he started to drive me mad. He has put everything in his mouth, has actually bitten the doors off the matchbox cars, chewed the wheels beyond recognition, eaten loads of paper, thrown anything he picks up, broken a vase whilst holding it and banging it with a toy in the other hand, tipped two bottles of sauce on the new carpet, emptied a box of rice crispies on the dining room floor, tipped Horlicks in the cupboard, emptied salt in the kitchen and poured kitchen cleaner round his bedroom and in his desk.”

“By Monday I just couldn’t cope with another week of the holiday to go, so I rung Mike (the nurse with the learning disability team). He suggested respite. I asked him what it was and, after a brief explanation, was sold on the idea. David went off to the local centre for children and despite me feeling very guilty, I cried all the way home. I left David there

for three days. After a long chat with them when I went to pick him up three days later, it was decided it would be beneficial for us as a family to have some regular respite. The respite relieved the strain that much, that I feel like I have been on holiday! Before I left, I booked David in for more respite in three week’s time and if you let me know when you are home next I can book for future dates.”

For those of you that haven’t guessed, David had Sanfilippo disease, MPS III, diagnosed later that year. After reading stories in the MPS Magazine about other families and the problems they faced, I think I could have probably recogised his condition from their stories myself.

I do urge you to try respite if you have not yet, and know that the MPS Society is very good at helping families to access what is available in their area. So, if you think you are entitled and don’t know how to proceed, get in touch with the MPS office.

Sadly, David died almost three years ago, but letters like this help me to remember him with a smile, although at the time it was very rough.

Angela Seymour
angie@seymour.freeserve.co.uk

A Year in Aix

When Joanne began her degree at Glasgow University, I knew there had been some vague mention of her spending a year abroad as part of the course, but I pushed it firmly to the back of my mind because it seemed an impossibility. I convinced myself that she would probably change courses and not have to go and, anyway, it wasn't until the third year of the course which was so far in the future I didn't need to contemplate it.

Well, guess how quickly those first two years went and, although she did change part of the course after the first year, it wasn't the French! So, about this time last year, the 'year abroad' working as a language assistant at a Lycee, was very much a reality. True to form, Joanne made it quite plain that she was making all the arrangements and that I didn't need to worry. Well, she did almost all the preparations herself, although I was entrusted with a few of the minor arrangements (just so I didn't feel totally redundant!) such as arranging a somewhat complicated train journey from home in Scotland to Aix en Provence (we couldn't risk flying with her powered wheelchair as it always gets damaged in flights!), hotels, shipping out her manual wheelchair and bulky items she couldn't live without, sorting out insurance cover - all reasonably straightforward if you are able-bodied but mention 'disabled' and all hell breaks loose! To make matters worse, in the midst of all this, Joanne also learnt that she would need to have cervical fusion surgery within the next year. However, at the moment it is scheduled for next spring after she returns from her stint in France. For me, this was a real double-whammy but having recovered from the initial shock, Joanne coped in her usual pragmatic way.

At this point I must mention the invaluable help we received from a family Joanne and I met during the International Symposium in Venice in the summer. Monika and Alain live in Marseilles (about 25 kilometres from Aix) and have two young sons, one of whom, like Joanne, has MPS IV. Now, whether meeting them was fate or coincidence I don't know, but I do know that the advice and practical help they provided prior to and after our arrival in France made everything so much easier. Although the 'year abroad' is arranged through the British Council and its equivalent body in France, it is very difficult to get any information and we were worried that they hadn't properly understood how having Morquio affects Joanne (and, as it turned out, we were right to be worried!). However, on Joanne's behalf, Monika liaised with the school where Joanne's placement is, checked out the accommodation which had been arranged for her, and arranged transport from the TGV station into Aix (public transport wasn't wheelchair accessible!). They were absolutely wonderful and I have peace of mind, knowing that Joanne can call them if she needs any help.

So, on 19th September, we set off on the first stage of our journey from Central Station in Glasgow down to London, to stay overnight before catching the Eurostar early the next morning (even getting to Waterloo on time was almost scuppered by an unco-operative black-cab driver - a complaint to the DRC is pending!) but we made it with a few minutes to spare. The Eurostar was fantastic. We knew we were to be upgraded to first class (one of the advantages of travelling with Joanne!) and we settled back to enjoy a sumptuous breakfast while speeding through the English countryside towards the Channel Tunnel. We were through the tunnel in about twenty minutes and then continued onto Lille where we transferred to the TGV train down to Aix which took about five hours. Travelling down through France was really interesting but we decided that the British countryside we'd seen the previous day was prettier.

We arrived at Aix TGV station which is 13 kilometres outside the city late in the afternoon. We were so relieved to see Monika and her sons waiting on the platform for us, together with an ambi-bus and driver (which she had had to bully the school into arranging!) to take us to Joanne's accommodation at the University. She was allocated the wheelchair accessible accommodation there as a special dispensation; this was a huge advantage since language assistants usually have to arrange their own accommodation and it's apparently not easy to come by. Joanne's room is the only one with an en-suite bathroom so she is the envy of all the other students! I had booked her a room at the hotel Graham and I were staying at for the first few days, but she insisted on staying in her own

MPS Awareness Day
Tuesday 15 May 2007

room at the university residence that night and it was with a heavy heart I left her. But having checked into our hotel, had dinner and a few glasses of wine (well, we were in France!) I soon got over it!

Despite Monika's help, the first few days were a bit traumatic to say the least. Trying to battle our way through French bureaucracy was a real eye-opener and we'll never moan about accessibility in Britain again. It's much better than it appears to be in France. However, by the beginning of the second week, Joanne was making some progress with all the red tape, and lots of progress with her social life! Living in the student residence is proving to be a real advantage as it's easy to make new friends. By the second night, she had a party arranged in her room and hasn't looked back. Several mornings I arrived to find the detritus of the previous night's revelries!

On the second day, we accompanied Joanne to the Lycee Paul Cezanne, where she will be working, to meet the staff; imagine our relief to discover that the one bus in Aix which is wheelchair accessible not only runs past the end of the road where she lives but also goes past the Lycee - at least something was working out for us!

Like everyone else we met in Aix, the staff at the Lycee were friendly and eager to help Joanne settle into her first paid employment. Although she wasn't due to start work till the middle of October, they invited her to go to the school to sit in on classes and get to know some of the pupils, all of whom are delightful and, understandably, curious about Joanne. She has already given most of them a master class about MPS diseases and the Society! Joanne works with five different classes and their respective teachers; she is contracted to work for twelve hours each week, which has been organised into two six-hour days to give her the maximum amount of free time (ie for retail therapy - Aix has more than its fair share of designer shops!).

Her first official day at work came as a bit of a shock after student life and, having worked a six-hour day, she was exhausted and couldn't imagine how anyone manages to work 9-5, five days a week! However, she is really enjoying her job and all pupils seem to enjoy having her in their classes - naturally her role is to help improve their English - and she's hoping they will help her to improve her French, although since most of the students at the residence are international, the common language is French, so she gets plenty of practice. The boys at the residence are also becoming adept at running repairs to her wheelchair!

For my part, it's really strange not having her around but it's no different for me than it is for any parents when their children go off to university. But, how typical of my daughter that she not only moved away from home but also moved to another country!

Joanne is relishing her first taste of real independence, and it is proving to be another wonderful experience for her. I am enormously proud of all Joanne has achieved against quite difficult odds, but she just takes it all in her stride. So far, so good and watch this space!
Judith Evans

Chelsea TV send recording of Barca game to boy with rare illness

Ben Conlin (MPS I) and his sister, Sarah

On Halloween this year, exactly three years to the day since Ben was diagnosed with MPS, Sarah, Ben and Tracey were preparing to go out 'Trick or Treating'. After that, the whole family were going to settle down and watch the first half of the Chelsea/Barcelona match on television. I was then going to record the second half for Ben to watch over the weekend.

Unfortunately, Ben, as usual had other ideas. Ben had been complaining of headaches for nearly two weeks. He had spent most of half-term in bed; we had been told by one doctor that Ben simply had a virus.

However, on Halloween, Tracey decided that Ben did not simply have a virus and so she took him back to North Tees Hospital for further tests. Luckily this time Dr. Verber, Paediatrician was on duty, this was the doctor who had virtually diagnosed Ben in 2003, so he knew the family well. He examined Ben, phoned Manchester (National Centre for MPS) and before we knew it the whole family were rushing home to pack clothes to drive to Manchester for the first of Ben's two 'Shunt' operations, to relieve the pressure in Ben's head in order to prevent brain damage.

Unfortunately, we missed the whole of the Chelsea game; we listened to the radio in the car and got to Manchester just in time to miss Drogba's last minute equaliser for Chelsea but we did see Jose slide down the touchline on his knees. Ben stayed in Manchester for nearly two weeks so I missed every opportunity to record the game with being in Manchester for so long.

In desperation I wrote to Chelsea TV and asked for a copy of the game. Last Tuesday 28 November we received not 1 but 2 DVD's from Chelsea TV of both Barcelona games. **Peter Conlin**

MPS Sibling Weekend

Centre Parcs, Nottingham, 31 August - 2 September 2007

The Society is delighted to offer this opportunity for MPS Siblings to participate in a fun-filled weekend at Centre Parcs in Nottingham. This is sponsored by the Newby Trust and the D'oyly Carte Charitable Trust. We would love as many siblings to come as possible so please complete the form enclosed with this magazine. If your child or children would like to attend but you can't get them to Centre Parcs please don't be put off. Contact the MPS Office immediately to discuss how we can help. Please note that if there is insufficient interest in this event the Society will not be offering a siblings event in the foreseeable future.

International Congress on Inborn Errors of Metabolism (ICIEM)

Chiba, Japan, 11 - 16 September 2006

The Conference addressed how recent research developments have revolutionised the understanding of disease mechanisms, genetic diagnosis, DNA diagnosis, stem cell biology, neuroscience, screening methods and new therapies. The Congress provided a wonderful opportunity to meet up with well known experts in the field and make new acquaintancies with doctors from Asia and the new world.

This important meeting brought me back to Japan, my home for nearly four years between 1976 and 1980, for the first time in almost 30 years. There was some apprehension returning knowing that the visit would bring back vivid memories of our time with Simon, our eldest son who spent more than half his life in Japan and died in 1982 less than eighteen months after we returned to the UK. The visit was not as painful as I thought it might be, not least because my husband Robin and two of our three surviving children, Ben and Lucy treated themselves to a holiday and joined me in Japan. Our visit started with a private lunch with the British Ambassador and his wife, Sir Graham and Toyoko Fry and finished with His Imperial Majesty Emperor Akihito of Japan speaking at the closing ceremony of the conference. Time was also spent meeting with the organisers of the Japanese MPS Society at which we discussed possibilities of greater collaboration. Tetsuya Motomura who is 20 years old and has ML II, and his mum Sally who are frequent contributors to the MPS Magazine live in the suburbs of Tokyo. Although Tetsuya keeps his spirits high, sadly his Dad, Yuki died recently and Tetsuya finds it very difficult to leave his home. It was lovely to visit Tetsuya and Sally at home and catch up on all their news. Sally and Yuki were our friends when we lived in Japan and had Tetsuya sometime after we left.

During the Congress Dr John Hopwood spoke on New Born Screening and the ongoing efforts of his team at the Women's and Children's Hospital in Adelaide to find a way to get enzyme into the brain and open the door for therapeutic treatment for all those diseases with CNS involvement in particular MPS III, Sanfilippo disease.

Drs Ed Wraith, Jo Muenzer and Paul Harmatz spoke on the outcomes of Enzyme Replacement Therapy for MPS I, II and VI whilst Dr Shunji Tomatsu described the rationale for developing an Enzyme Replacement Therapy targeted at the bones for MPS IV. Dr Robert Desnick gave a plenary presentation 'Treatment Options - what do we know and where are we going in the future?' This presentation set out The Principles Learned from Enzyme Replacement Therapy: Enzyme Delivery is Receptor Mediated and Dose Dependent; Substrate Clearance is Dose Dependent; Early Treatment is Critical for Maximal Effectiveness and to prevent Pathological Accumulations; ERT is Safe and Well Tolerated with Infusion Associated Reactions Mild and Manageable. There was also discussion on New Emerging Treatments including: Substrate Reduction Therapy; Pharmacological Chaperone Treatment; and Stem Cell Therapy. Discussion on What is New? covered: Enzyme Replacement Therapy with NEO-rh GAA; new generation of enzymes.

Gene Therapy

A lot of Gene Therapy has been carried out in Lysosomal Storage disease mice but where are we now? Are we lost in translation? STOP CODON READ-THROUGH THERAPY is a new concept that reads trends in molecular medicine and we need to watch this space.

Stem Cell Therapy

There is considerable debate around the ethical issues relating to the use of fetal stem cells. Adult stem cells are offered as an alternative but there are major issues and challenges ahead. A principle question to be asked is 'Can adult stem cells be isolated?' A paper published in Nature August 23, 2006 : Cell 126; 663 - 676, 2006 Takahashi and Yamanaka 'Induction of Pluripotent Stems Cells from Mouse Embryonic and Adult Fibroblast Cultures by Defined Factors' suggests they can. **Christine Lavery**

Photos above: UK - Japan MPS Society Meeting, 13 September 2006, Hotel New Otani, Chiba, Japan

11th National Australian MPS Conference, Brisbane

28 September – 1 October 2006

After the heavy schedule of Japan, as a family we had a relaxing nine days' holiday before Robin and I arrived in Brisbane to be welcomed by a terrific rainstorm. We may tend to think it is only parts of England that are suffering drought conditions but this was the first substantial rain Brisbane has had in many months.

Walking across the car park on the morning of the first full day of the conference, the scene could have come from any of our UK MPS Conferences. Children were eagerly boarding buses for Movieland with their volunteer carers whilst anxious parents looked on. We were quickly reminded that it couldn't be England by the sun beating down on us, the abundance of palm trees and, yes, the parents were dressed in shorts! After waving off the children and chatting to a Mum whose son is 31 years old and has Sanfilippo disease, the conference was opened by the President of the Australian MPS Society, Wendy Boon. With sadness Wendy also announced that she would be stepping down as President after the conference but staying on the Board of Trustees.

Dr Ed Wraith then gave an excellent overview of the Lysosmal Storage Diseases, the first of three presentations he would make during the conference. Ed described the clinical variability in infantile, juvenile and adult onset variants seen in most conditions. We then moved on to hear two family stories. Firstly, Rebecca Johnston, a year 9 student at St Margaret's Anglican Girls School in Brisbane, showed a 'virtual' presentation depicting the life of her sister Nicola who died from MPS III, Sanfilippo Disease, a year ago. Rebecca, who created her presentation for her Technology Assessment has achieved academically appearing on Australia's brainiest kid TV programme, and has recently been on tour in the USA playing her cello and singing in the choir. However, Rebecca said that the most important lessons she has learnt, as yet, are about laughter, compassion, bravery and unconditional love taught to her by her beautiful sister, Nikki.

The second family speaker was Nicole Millis. Nicole and her husband, David, are the parents of Sean aged 5 years. Sean was born 7 weeks premature via an emergency caesarean section. After such a dramatic beginning Nicole and David looked forward to things settling down as they enjoyed being first time parents. However, things never did settle down and after a number of confusing medical problems Sean was diagnosed with MPS II, Hunter Disease, at the age of 9 months. Subsequent tests showed that one of Sean's cousins also had the disease and it is now believed that Nicole's uncle also had MPS II which was never diagnosed. Nicole went on to share her family's journey with Sean and described three partnerships important to Sean's diagnosis: The family and healthcare professionals; Nicole and her husband, David; Nicole and Sean. Lectures on recognising the Symptoms of Neurological Problems, Benefit of Registries, Spine, Hips and Knees and Palliative Care preceded workshops by disease groups.

Next day, after seeing the children off for a half day visit to Lone Pine Koala Sanctuary and a cuddle with a koala bear, the conference started with Dr Ed Wraith giving an update and results of MPS clinical trials. Professor John Hopwood described the advancement of Newborn Screening for Lysosomal Storage Diseases and Dr Martin Delatscki shared the results of his newborn screen survey. This survey took place nearly two years ago and included the USA and UK as well as Australia. Unfortunately, so few responses were received from our UK members to the questionnaire that they were significantly insignificant and not included in the feedback of

Catherine Hartcher (MPS IV) and her parents showing off her painting by Evan Standish (MPS VI). Brisbane, Australia

results. Dr Kim Hemsley, who works with Professor Hopwood at the Adelaide Women's and Children's Hospital, gave an excellent overview of the obstacles to crossing the Blood Brain Barrier to Provide Therapy for the MPS Diseases that affect the brain. Clearly there is significant work in progress but more time is needed.

Dr Jim McGill and I ran an MPS I workshop which included six MPS I families. All but one had had a bone marrow transplant with the oldest child aged 8 years. The families were very keen to learn of Cheryl Pitt's research on the Psychosocial Outcomes of MPS I post BMT and I was able to share the information from the poster which we had displayed at the ICIEM meeting in Japan.

There was no conference on Saturday afternoon due to the important Cup Final – Aussie Rules Soccer between Sydney and Melbourne. However, as the Guest Speaker at the Gala Dinner, I recollected the 22 years of shared collaboration and managed to raise a few laughs.

The final day of the conference focused on future directions with Ed Wraith speaking on the Limitations of Current Therapies and John Hopwood explained the challenges of Gene Therapy. John explained that we are each made up of: A hundred trillion cells; Every cell has a nucleus which is very small but still visible under the microscope; The nucleus contain an even smaller set of 46 chromosomes; Every chromosome contains DNA; Along our DNA lie over 24,000 genes.

John described this as the 'Small Large Universe' and explained the obstacles this presents in developing a gene therapy that will target the defective causative genes in a particular disease.

The topic of the weekend for a majority of MPS families at the conference was access to Enzyme Replacement Therapy. In Australia ERT for Gaucher and Fabry Disease is funded but not MPS I or MPS VI and it is not expected for MPS II. Dr Jim McGill explained the funding process for new drugs and explained to families the actions they could take in lobbying. Australia is a huge country with many layers of bureaucracy (even more than the UK) and I was shocked to learn that the final decision on funding of ERT for MPS I, MPS VI and MPS II in the future will be made at Cabinet level by people with no direct experience of MPS diseases and who need to balance the books! Many Australians have descended from relatives that held UK passports and two desperate families told me they are seriously

thinking of moving to England so their children can be treated. It is a very sad state of affairs but no different from one that our members face in Scotland and Wales.

The conference concluded with me presenting an update on the International MPS Network, John Hopwood on GOLD, Wendy Boon on the Australian MPS Society and John Forman on the Lysosomal Storage Disease Society in New Zealand (LSDNZ).

I am deeply grateful to everyone at the MPS conference for such a warm welcome and particularly to Wendy Boon who worked so hard to make the conference the success it was. I am also grateful to Shire Pharmaceuticals for an educational grant that enabled me to attend to both the ICIEM meeting in Japan and the Australian MPS conference. **Christine Lavery**

Photo above: Christine Lavery, David Lewis and Dr Ed Wraith
Photo below: Christine with the Stewart family who lived in Berkshire until 3 years ago when they moved with Lisa (MPS III) to the Australian Gold Coast.

VENICE

Judy Holroyd, a Trustee of the MPS Society, attended the **9th International Symposium in Venice** in July 2006. Judy has summarised two of the topics covered in the sessions for readers of the MPS Magazine...


Judy Holroyd

Update on haematopoietic cell transplantation (HCT)

Since 1980 worldwide more than 400 patients with MPS I Hurler Disease have undergone HCT. Although long term follow up of successfully transplanted children is very encouraging, the engraftment and survival rates are very variable between studies.

Dr J Boelens from The Netherlands reported on a retrospective study carried out with other clinicians in Europe. They analysed the results of 146 patients transplanted in Europe from 1994 - 2004. The focus of the study was:
1) to evaluate the preparatory and ongoing treatment given to patients to prevent rejection of the graft (conditioning regime) and the stem cell source for the graft (bone marrow, cord blood or peripheral blood) and 2) to evaluate the complications (morbidity)/survival rates (mortality) related to the transplantation.

In summary:

No stem cell source was found to be superior.
- Patients that received a T-cell depleted graft and a reduced intensity conditioning regime showed less good engraftment rates. Engraftment of HCT for Hurler syndrome can be optimised by avoiding these.
- Second HCT's were successful in more than 80%.
- Full engraftment was more often seen in patients receiving cord blood.
- Relatively low morbidity rates were seen.

Dr P Gupta from Germany followed with a presentation about the potential use of stem cells from sources other than bone marrow or blood (non-haematopoietic) that may be used to ameliorate some of the lysosomal storage diseases.

Stem cells have the potential to develop into many different cell types in the body. They can theoretically divide without limit and each new cell has the potential to either remain a stem cell or become another type of cell with a more specialised function, such as a muscle cell, a red blood cell, or a brain cell.

It is hoped that these non-haematopoietic stem cells may prove to be powerful tools for the future to overcome the limitations of current available treatments. Dr Gupta concluded by saying that despite these exciting advances, a number of questions remain to be answered and major hurdles remain to be overcome before these types of stem cells can be used in clinical trials.

The third presentation in this session was given by Ms N Wager who outlined the findings of the study carried out by Cheryl Pitt on the Psychosocial Outcomes of Bone Marrow Transplantation (BMT) for MPS IH. The conclusions were:

'The findings illustrate how aspects of the MPS condition in terms of adaptive skills, and physical and cognitive function contribute to the personal adjustment of children and young people affected by MPS IH post BMT. However, they also implicate aspects of the mother and the family environment, particularly in terms of mothers' coping and the recreational activities of the family. Aspects of parenting and the family, as well as aspects of the MPS disease, therefore require attention when providing support to these patients and their families. Appropriate and consistent classroom support for this patient group also requires attention, as does the question of whether psychosocial support should be considered within the school environment.'

Blood brain barrier (BBB)

This workshop on the blood brain barrier was given by scientists who are working at the cutting edge in this highly specialist and technical field.

The blood brain barrier is both a physical barrier and a system of cellular transport mechanisms. It restricts the entrance of potentially harmful chemicals from the blood entering the brain but does allow the entrance of essential nutrients.

Although enzyme replacement therapy (ERT) has proven to be an effective treatment for non-neurological symptoms of lysosomal storage disorders (LSD's) it has not been an effective strategy to treat the neurological impact of many LSD's. This is because the BBB acts as an insurmountable barrier to these substances.

Overcoming the BBB is now a challenge facing researchers attempting to treat LSD's by delivering therapeutic enzymes to the lysosomes within the brain.

Dr Wolburg from Germany, Dr Begley from Kings College, London and Dr Leon from Italy all put forward scientific hypotheses about how the BBB may work at the cellular level.

Some late breaking news at the conference was announced from Dr P Dickson et al. from the USA who presented results of their research that demonstrated 'High-dose intravenous ERT treats the brains storage in tolerant MPS I dogs'.

A tale of three children - the latest…

Radhika, 15 years, and her brother Marimuthu, 14 years, with Manikandan, 12 years (the youngest of another family) have been resident at The Cheshire Home, Chennai for the past four years. They have Morquio. It is thanks to Elizabeth Herridge, wife of the British Deputy High Commissioner for South India, who ensured their future when she was living there and gave much of her spare time to the over 100 disabled children at Andhra Mahila Sabha. At the end of her tenure with her husband Michael, four years ago, she arranged for them to be moved to The Cheshire Home from AMS - an orthopaedic centre for poor children.

The emphasis at the Cheshire Home is on education and rehabilitation, and at the outset we were able to place the children in a school for special needs children with provision for children in the mainstream system. Radhika and Marimuthu were placed in the mainstream system and showed particular aptitude in their studies. Manikandan's background had not prepared him for the mainstream system and he was placed in the special needs system.

The physical condition of the children is fairly weak. They are able to walk on their knees but mostly move around on small tricycles. We do have to check ourselves these days to accept that, despite their fragile and small frames, they are now young adults with the same demands of young teenagers and the same fears and expectations. Marimuthu is very inquisitive and very reasonably independent. Radhika is not so confident but is a very sweet young girl. Manikandan charms everyone and should a camera be in sight he is the first to have his photo taken, and is probably the most photographed child in the Home.

Their daily routine is supported by a Home Carer who brings them morning tea at 6.30am, after which they have their bath and are then dressed in their school uniforms. The children do need help with their dressing but are independent in their daily habits. Breakfast is at 8.00am and they leave for school at 9am with their lunch (tiffin) packs. They are taken to school by the Home vehicle and collected again at 4pm. On their return from school, they are given a hot drink and snack, change out of their school uniform into casual clothes and then relax for an hour.

They are supported by a physiotherapist/occupational therapist and during weekdays they spend time either on the trampoline, slide or swing, after which a tutor comes to help them with their homework and study from 5.30 pm. Radhika, in Standard 9, and Marimuthu, in Standard 8, have a heavy homework load but are very enthusiastic and doing well. They have their dinner at 8pm and are in bed at 9pm.

It is pleasing to note that the children keep in good health and are rarely ill. The heaviest cost is their hearing aids which are either misplaced, lost or sometimes snatched from their ears by the other children. This hearing defect also deters them from swimming therapy although Manikandan enjoys the pool activities.

Radhika and Marimuthu's parents live over 200 miles from Madras which is an overnight train or bus ride, but the children receive regular calls from the parents. They go back to their families for term holidays and the occasional festival holiday or, alternatively, their mother will come and visit them. More recently, the elder sister of Radhika and Marimuthu was married and the children attended the wedding.

Manikandan's mother lives near the Home and visits on alternate weekends. His sister suffers from night blindness and is being supported by our Cheshire Home Community Outreach Project which assesses and treats the condition of those persons with disability in the community.

Members of Manikandan's family were victims of the tsunami which hit the coast of Tamil Nadu in December 2004. They are now housed in a new home thanks to the sponsorship and support of Elizabeth and her friends, including members of the MPS Society which enabled the purchase of land and other donors constructed a building for them and they now have a home for life.

Prepared by Maureen Hudson Murari
Hon. Sec., The Cheshire Home, 23 Third Seaward Road, Valmiki Nagar, Thiruvanmiyur, Chennai 600-041, Tamil Nadu, South India.

International LSD Meeting, Budapest

November 10 - 11, 2006

Christine and I were fortunate to be invited to attend the International Workshop on Lysosomal Storage Disorders in Budapest, Hungary. The workshop was being held in the Intercontinental Budapest located on the river Danube.

The workshop was opened on Friday 10th November with updates from HOS (Hunter Outcome Survey). This is something that has recently been set up to monitor and assess individuals with Hunter disease in order that they can learn more about their condition. Countries from around the world can contribute to the study and it will help physicians analyse and evaluate data to look at outcomes. It was good to hear that the UK has been very proactive in registering patients onto the study and other countries were following suit.

Dr J Muenzer gave an update on the natural history of MPS II which was followed by our very own Jane Roberts who shared an inspiring presentation on their clinical experience of caring for Hunter patients in the UK. The day was concluded with some very in-depth presentations on the central nervous system in Fabry disease.

On the Friday evening we were taken to a restaurant on an island. The evening started off with us being entertained by some traditional Hungarian folk music followed by a true Hungarian buffet of soups (which they are famous for) stews and very sinful strudels.

The day on Saturday started early with more presentations on Fabry disease. The morning focused on cardiac problems and after lunch the discussion was around the benefits of early diagnosis and treatment in Fabry disease. These presentations covered topics ranging from renal, children and ERT. During the poster session Christine and I were invited to attend the International MPS Network meeting. Countries from across Europe had attended the conference and the organisers had been kind enough to allow us time and space to meet. This is something that is not always possible and to have so many countries (approx 12 in total) in the one location is a rarity and something that we were fortunate to take full advantage of. It was good to hear about the other countries' experiences of running a patient organisation and to get updates on ERT.

On the Saturday evening we were taken to an old fashioned Turkish bath and although we did not partake in a dip we were able to view the hot bath from afar. After yet again another huge feast we returned to the hotel to have one last drink with colleagues to bid farewell as many were leaving the following morning.

The weekend was very informative and it was great to meet up with so many friends and colleagues. **Sophie Thomas**

Photo top: Bettina Wildi (German MPS Society) Martin Weigl (Austrian MPS Society, Flavio Bertoglio (Italian MPS Society), Fer Pidden (for Turkish MPS Families). Photo bottom: Christine Lavery, Sophie Thomas and Rita Hausken Barkhodaee

Norwegian MPS Meeting

20-24 November 2006

Frambu

The Norwegian MPS Patient Meetings are always a privilege to participate in and the one held recently in Frambu was no exception.

Frambu is a national centre for rare disorders and disabilities. Frambu's mission is to improve the quality of life for those with rare disorders and their families, at home, at school, at work and in the community so that they can better cope with daily life.

Frambu has a multidisciplinary staff, covering the medical and social care aspects of life for families affected by rare diseases.

The MPS week at Frambu is always very busy. The children go to school at Frambu for most of the week coming out for clinical and social care sessions. Whole families come so the place is alive with not only those with MPS but all the healthy brothers and sisters of all ages.

During the clinic sessions Dr Ed Wraith saw over 20 families. The adult MPS workshop I ran brought different challenges. There were several young adults with Morquio, four with MPS I (2 MPS I post BMT and 2 with Hurler Scheie on ERT) and one young lady with MPS VI.

We covered areas of further education, employment, independent living, access to treatment and issues around sexuality and reproduction. The age range of the young mixed group was 16-33 years.

Towards the end of the week all the parents, adults with MPS, doctors, Frambu staff and professionals from the community came together for a one day conference at which Dr Wraith was the main speaker.

Thank you to everyone at Frambu for organising another most successful MPS week. **Christine Lavery**

Frambu is about 15 miles outside Oslo in a deserted area where, in the summer months, the lake and woodland are fully utilised by children with rare diseases and their brothers and sisters. This was the scene from Christine's bedroom window (a room that would usually be occupied year round by a number of families).

5th International Fabry Patients' Meeting in Brussels

October 19-22, 2006

We have all just returned from a wonderful meeting at the Sheraton Hotel in Brussels. This was my first time at an International Fabry meeting and to start with I felt overwhelmed to suddenly be amongst so many Fabry patients. As time went on this feeling turned into a sense of being part of a huge family. There were many nationalities there, Belgian, British, Danish, Finnish, French, German, Italian, Norwegian, Portuguese and Spanish. The simultaneous translators were fabulous. Many of us looked the same of course with our polymorphic features which made us feel even more like a family. The feelings of isolation that are part of daily life were melting away.

There were several lectures and question and answer sessions. Many doctors, professors, nurses and Fabry support groups were there. It was extremely interesting to hear how different countries deal with the treatment and care of Fabry patients. Some countries were still having huge problems with bureaucracy and therefore had relatively few patients on ERT as a result. It was a big surprise to find that in Italy, where all babies are tested for genetic diseases, the number of Fabry babies born is 1 in 3100. If this amazing figure is true for other countries, we could be way out in our current estimation of 1 in 40,000. 1 in 2000 counts as a rare disease.

It was wonderful to hear people talking about the improvements in the progression of their disease since starting ERT. Of course the longer people have been suffering, the longer ERT takes to work. There was no doubt however that the progression of the disease was first of all slowed down and then halted over time. It could take years to clear the Gb3 away but no one was in any doubt that so long as you were not in too bad a shape to start with, you would surely improve.

That was the feeling for both Fabrazyme and Replagal. It could take a while for ERT to stop the heart and kidney from deteriorating in the early stages of receiving ERT but there would come a point where the deterioration would stop and an improvement would begin to be seen. One doctor described the build up of Gb3 in the body like bags of uncollected rubbish in a house. First of all it builds up and fills the kitchen, then the rest of the house. Then it spills out of the front door, at this point your internal organs start to show damage, then it moves down the garden and when it reaches the garden gate unfortunately your organs will be permanently damaged and may then go on to fail.

The important thing is to start ERT at the latest when it reaches the front door and before that if you are symptomatic. If ERT starts when it is some way down the garden it might take 2-3 years to get it back to the front door and during that time, further damage will continue to take place in your organs. It is an argument for treatment happening before it gets to the front door but for most of us we only receive treatment when there is evidence of deterioration; when it is some way down the garden.

We learnt the difference between missense and nonsense mutations. They were likened to getting a flat-packed article of furniture home and trying to put it together. In the case of the nonsense mutation, the company have forgotten to put the instructions in the box and in those mutations people have no enzyme. Then you could have a set of instructions but they are so poorly drawn that you cannot make them out at all. These are missense mutations where you may have a little enzyme. The nonsense mutations are usually more severe than the missense mutations although there are some missense mutations that also create very serious cases. Many of the missense mutations may become treatable by the so-called chaperone treatment which refolds the instructions to make them understandable to the body. There were a couple of people at the conference on the current chaperone treatment trial and we look forward to hearing how they are getting on as time goes by.

There was a great deal of talk about symptoms of course and many I had not known about before. I did not know for example that stress increases body temperature which of course increases pain. One patient stood up and described how he had used a thermometer to control his pain. He had

Photo above: British Fabry sufferers and partners at the dinner, Photo page 33: Young people get together

worked out that when he first began treatment with ERT that pain kicked in at 36.9 degrees centigrade and now after some time on ERT pain kicked in at 37.8 to 37.9 degrees centigrade. For him it was just a question of keeping the temperature below this point. If we could find out these precise levels for each individual maybe we could reduce the amount of pain felt.

There was a comment about the body's inability in Fabry disease to adjust the heart rate properly during exercise. There have been some accounts of patients with excessive sweating and also those who suffer from excessive sweating in some areas of their body, for example, in their hands and from not being able to sweat at all in other areas. I talked to a couple of patients who were infusing every week, doing their own infusions. They used half their two-weekly supply of enzyme in the usual amount of saline they would use for their fortnightly infusion. They both said they felt better for the whole two weeks whereas before during the second week, they had seen a worsening of their symptoms. This seemed to be backed up by a doctor who said that sweating levels fell in the second week. I hope to be brave enough to infuse myself one day so I can try out the weekly theory.

There were other symptoms I had not heard about before like pain in the teeth, ulcers in the mouth, recurrent miscarriages from coagulation and mood swings from one day to the next. Also we were told that where people suffer with gastro-intestinal symptoms it is because the food is not absorbed properly in the intestinal tract. Depression in the normal population is 3-15% and in Fabry is 30-50%. They suggested not letting depression last for more than two weeks before seeking help. Signs of depression included disturbances of sleep, tiredness or restlessness, feeling very sad and down, lack of appetite and becoming bored with hobbies. Psychological problems could often be caused by either pushing yourself too hard at work or by being pushed too hard by work colleagues or bosses. Many of us talked to each other about our problems with short-term memory loss like instantly forgetting someone's name or where they came from more or less as soon as you'd been told. Luckily we all had name badges! I imagine it is the lesions in the brain that probably cause this. Many of us also shared our experiences of loss of concentration and mood swings.

In Fabry babies, diarrhoea is a frequent symptom. In girls if they had problems with sweating or had frequent hospital admissions as children, they were more likely to have a more serious form of the disease when they grew up. Two wonderful teenagers stood up and described their experience of Fabry and one of them said that over the four years he had had ERT, he had suffered less and less pain. There were many young people there and separate sessions were organised for them. A few people came on their own but mostly with other family members.

There was a detailed lecture on family planning with some interesting information on PGD (pre-implantation genetic diagnosis) and PND (prenatal diagnosis). The PGD method was also recommended for men so they could have daughters free of the disease using donor sperm.

There were many ideas put forward to help us all cope with our symptoms such as eating light, frequent meals, eating a low-fat diet, avoiding meals that are too hot or too cold,

avoiding aggravating drinks and avoiding getting over-heated especially after eating. Another patient told me he had investigated what he eats with a nutritionist and found a very precise diet to avoid pain, even down to different brands of baked beans and definitely avoiding cheesecake which for him would produce six days of pain. Maybe we should all work out our own eating regimes too. Taking a short walk or maybe a swim each day and only exercising to a level at which you know your body is comfortable.

Several specialist Fabry support groups spoke about their activities and described the feeling of isolation which all Fabry patients feel. This made us all realise how important these meetings are for talking to other sufferers and sharing experiences.

There was a wonderful, uplifting session by someone who called himself a 'change-ologist'. He illustrated his ideas with some jugglers, comedy sketches and slides. He put across the importance of patient power and how to synchronise it. He described how we had all had to be so adaptable to adjust to our difficult situations not only when we first found out about our diagnosis but then as time went on too. He suggested something called 'out of the box thinking' where you use creative thinking to improve your own situation and that of fellow sufferers. I think he was referring to ways to come up with new ideas for dealing with your symptoms or your approach to life and work. Also maybe joining up with others and making a difference by tackling difficult problems like insurance, funding or setting up support groups or online chat rooms etc. His talk was full of emotive words like faith, trust, hope and letting go and we all found some sort of solace in what he had to say. He mentioned several books which could be worth investigating; The Right Brain Manager, Our Inner Ape, Your Brain as Medicine and Visions of the Future.

It is hard to be brave all the time isn't it? But many patients who found the courage to speak were an inspiration to us all.

I was relieved to hear from one of the specialists in the UK that NSCAG have agreed to fund treatment for another year to April 2008.

It was announced that next year's conference will be in Munich and we are all looking forward to it already. I think we have all come back with a renewed sense of hope and full of ideas for the future. We have also found so many new friends. Thanks to the organisers and the sponsors for everything. **Jane Osborne (Fabry Disease)**

The Welfare of Laboratory Animals

Introduction

The MPS Society has supported animal-based research for Lysosomal Storage Diseases, undertaken in properly regulated research centres, where there is no realistic alternative. MPS has also supported efforts to find alternatives to using animals for research. Animal based research in the UK is governed by some of the strictest laws and regulations to be found anywhere in the world. The Society works to a strict Animals in Research policy that is reviewed regularly.

Regulation

The current regulatory framework in the European Union for controlling the use of animals in research and other purposes is the 1986 Directive. This is currently undergoing a review, and a new directive is expected to be introduced to the European Parliament in early 2007. This directive will focus on welfare issues as well as on experiments that are permitted through the introduction of controls and by encouraging the development of alternatives.

The MPS Society welcomes the introduction of appropriate regulation that will prevent abuse if and when it occurs, and also the encouragement of non-animal alternatives in research wherever these will not compromise the safety of any products that may emerge, or retard progress towards much needed novel therapies for serious genetic diseases which include the Mucopolysaccharide and related conditions.

Why are we concerned?

The proper use of animals in medical research has already resulted in substantial improvements for some of our members and emerging research that depends on appropriate animal research offers more of our members hope for the future.

Central to this is a legal framework that is appropriate and proportionate, and which allows essential research and development to proceed safely whilst protecting the welfare of the animals used to the fullest extent reasonably practicable.

The European Commission published a consultation document based on the suggestions of the European Food Safety Agency and the Environmental Directorate-General's Expert Working Group in the summer of 2006. Unfortunately this document was considered seriously flawed by the patient organisation the Genetics Interest Group, of which the MPS Society is a member, both in respect of its scientific rationale and in terms of the processes adopted to seek the views of professionals and lay people.

The MPS Society's concern is that in considering animal welfare issues there was no recognition as to why animal research is undertaken. Without the research being placed in the context of serious (or lethal) diseases and the needs of patients and families for treatments that will meet their currently unmet medical needs, there is a significant risk that the new regulatory framework will prevent much needed research from being undertaken in the European Union. The new regulatory framework needs to be designed with proper consideration as to why the research is needed in the first place.

The MPS Society is keen to stress that it is not the use of animals per se that is the issue rather it is the need to encourage research and development, and the production of safe, effective and available therapies for patients. As things stand animal research is an essential step in this process; without it, much needed medical innovation would be at best significantly impeded or at worst completely prevented in the European Union.

How you can help?

When the Commission's proposals are introduced in Parliament there will be detailed discussion of the technical aspects of the proposed regulatory framework. This is right and proper and whilst most people would expect that animals used for research would be as well treated as possible it is an issue that can be left to technical experts.

However the directive will also seek to put controls on what type of research can be done. The European Parliament is already being strenuously lobbied by those who oppose all forms of animal research with the intent of making the regulatory framework so tough that such research would effectively become impossible in the EU.

With the Commission's proposal being brought to the European Parliament imminently there is urgent need for the members of the European Parliament Committee leading on this to be made aware of your views as patients or patients' family members as to the need for good quality research and development and the role animals play in this for MPS.

Input from you has the potential to improve European medical research opportunities whilst at the same time affording proper consideration to animal welfare issues. **Christine Lavery**

Links to other organisations related to this topic: The People's Petition: www.peoplespetition.org.uk, Coalition for Medical Progress: www.medicalprogress.org.uk, Pro-Test: www.pro-test.org.uk, This article is drafted from a paper written by Alastair Kent, Director, Genetics Interest Group.

Do you want to be on the stage?!

Shed@ThePark was incorporated on 19th December 2005 to provide an exciting new Inclusive Theatre Company for the active creative participation of children, young people and adults in The Chilterns and South Buckinghamshire, regardless of health, social development or ethnic diversity. If your child/children are interested, or you know somebody who is, why not visit **www.shedatthepark.co.uk**

Disability Equality Duty

Disability Equality Duty (DED) came into force on 5th December 2006. The role of the DED is to ensure that public bodies which includes local authorities, health trusts, emergency services, schools (although primary schools have until 2007), colleges and universities have 'due regard' to the promotion of equality for disabled people in all areas of work and life.

It is hoped that DED will deal with peoples' attitudes to disabled people and promote inclusive thinking, ensuring that issues of discrimination and harassment are no longer. Another purpose of the scheme is to ensure greater knowledge is sought to understand what disabled people want from public services and to promote equality for all disabled people.

There is a big responsibility for public bodies to publish a disability equality scheme which should include such things as an action plan and have a strong input from disabled people. They also need to show that the actions have been followed through. Reports need to be submitted yearly and the scheme reviewed every three years.

The aims of the DED is to make public bodies look at the way they do things, so that disabled people have the same rights and access to support as others.

'Marie Pye, head of the DED, from the Disability Rights Commission says that the duty provides "a fantastic framework" to address the institutional discrimination plaguing public authorities with a "specific role" for disabled people to play, as they will have input into the schemes. She also feels that it will help across whole organisations rather than being dealt with on an individual case' (quoted in Disability Now - Nov 2006).

However, it is uncertain how many public bodies will actually have had these schemes in place since 5th December 2006 and whether they have truly consulted and involved disabled people in the setting up of the schemes. Enforcement action is however in place and, for those public bodies who do not comply, action will be taken which could be as early as now, January 2007. If a public body still refuses to comply then they could be subject to a judicial review.

Rotating ELAP car seat and three-wheeled wheelchair available

One of our member families has a three-wheeled wheelchair, plus rain covers that came from America from babyjogger.com. It has been well used, is good over rough ground, but it doesn't fold down very small so is best transported in an estate type vehicle. The family also have a rotating ELAP car seat that can be fitted instead of the front seat in a Vauxhall Zafira. Although it has to be fitted professionally, it is brand new and saved a lot of back strain! For more information please contact the MPS Office.

For Sale

2003 Chrysler Voyager CRD Touring 2.5 Diesel. 30,000 miles on the clock, full service history, air con, tinted windows, alloy wheels, new tyres, 10 months MOT.
Converted by Gowrings, from new, in September 2003.
Lowering rear suspension for easy ramp access, 4-point wheelchair/buggy securing straps and a 3-point seat belt also for the chair user. **£15,000-£16,000**
For more information please contact the MPS Office on 0845 389 9901 or email mps@mpssociety.co.uk

Morquio A Project: over the tragedy and difficulty

We have had great progress through this year in collaboration with all Morquio families, MPS Societies, core medical doctors, Inotech Co, and PFC Pharma Co. and our lab colleagues on the Morquio Project. We are very close to the natural history program in Summer 2007 and will conduct the clinical trial in early 2008.

However, as you may have heard already, we have some very sad news. Our precious lab manager Monica Gutierrez was killed by car accident on 3 November 2006. I heard about this tragedy while I was at Inotech Co. in Switzerland. I am deeply sorry for what happened on our beloved Monica.

I remember I met Monica at the international medical conference in Colombia at 1999. I was introduced to Monica by Prof. Barrera. I remember Monica talked to me about the curiosity of our research even though it is a quite rare disease. Since then, Monica had contact with me and I happily welcomed Monica at St. Louis. Monica and I have worked together for these five years at Biochemistry and Pediatrics. Monica is the first research assistant at my lab. Monica is quite talented and has worked very hard at my lab to develop the drug for children suffering from Morquio disease.

Monica has helped me a lot during my work and given me remarkable achievements in science. Without Monica's dedication to work, it would never happen. We are very close to reaching our goal to develop the drug which we have pursued for a long time. With a big hope, Monica was supposed to apply to graduate school for the coming December. I was so happy that Monica would become a graduate student working together here at my lab. We shall succeed to Monica's hope in medicine and shall surely achieve our goal on behalf of children waiting for the drug. Thanks for all the encouragement we have received following the tragedy. Now our Morquio Dream Team is recovering day by day. We are sure we can work together for this goal.

Shunji Tomatsu MD PhD, Associate Professor, Department of Pediatrics, Saint Louis University

Research Update - MPS IIIA and MPS II

Shire Human Genetic Therapies has completed proof of concept studies on two projects and has advanced them into pre-clinical development; namely Enzyme Replacement Therapies for MPS IIIA and intrathecal delivery of Elaprase for MPS II, Hunter Disease patients with significant central nervous system symptoms.

Announcing studies of new investigational treatments for males and females with Fabry Disease

Females

Study Title: 'A Phase 2, open-label, multiple dose level, 12-week study to evaluate the safety, tolerability and pharmacodynamics of AT1001 in female patients with Fabry Disease'

Who can participate?

This study is open to females 18 years of age and older with Fabry disease who meet certain criteria; including, having a type of genetic mutation called a missense mutation in alpha-galactosidase A. The study investigator can tell you if your mutation meets this requirement.

What is involved?

The study will involve 7 visits to one of the six investigational sites over an 18 week period. Participants in the research study will be eligible to continue in an extension phase of the protocol for an additional 36 weeks if they have a positive response to treatment. The six treatment centres are located in Manchester, UK; Atlanta, Georgia; Montreal, Canada; Melbourne, Australia; Porto Alegre, Brazil; and Paris, France.

Males

Study Title: 'A Phase 2, open-label, single dose level, 24-week study to evaluate the safety, tolerability & pharmacodynamics of AT1001 in patients with Fabry Disease'

Who can participate?

This study is open to males 18 years of age and older with Fabry disease who meet certain criteria; including, having a type of genetic mutation called a missense mutation in alpha-galactosidase A. The study investigator can tell you if your mutation meets this requirement.

What is involved?

The study will involve 9 visits to one of the 4 investigational sites over a 30-week period. Participants in the research study will be eligible to continue in an extension phase of the protocol for an additional 24 weeks if they have a positive response to treatment. The 4 treatment centres are: two centres in London, UK; Montreal, Canada; and Paris, France.

These studies will collect information about the safety and preliminary effect of new, orally administered investigational drugs to treat certain males and females with Fabry disease.

Your participation in this study will be valuable for the continued development of these new investigational drug treatments. These drugs may benefit individuals with Fabry disease in the future.

The reasonable costs of travel necessary for participation will be covered by the sponsor of the study.

The sponsor of the study is Amicus Therapeutics, a biopharmaceutical company developing a novel class of small molecule, orally-administered drugs known as pharmacological chaperones. For more information on Amicus, please visit **www.amicustherapeutics.com**.

The study is currently active. If you would like more information please contact the MPS office.

Statement on the clinical trial with genistein for Sanfilippo disease

In the course of realisation of the project financed by the UK MPS Society, a group led by Prof. Grzegorz Wegrzyn (University of Gdansk) has developed the principle of the method of gene expression-targeted isoflavone therapy for Sanfilippo disease (Piotrowska et al., Eur. J. Hum.Genet, 2006, 14, 846-852). This method is based on the discovery of genistein-mediated inhibition of synthesis of GAGs, due to blockage of expression of specific genes by interference with Epidermal Growth Factor receptor (particularly its tyrosine kinase activity).

It is obvious that further studies on this phenomenon are absolutely necessary to:
(i) elucidate precise mechanism of the genistein action
(ii) determine which particular genes are affected by genistein - this should indicate which subtypes of Sanfilippo disease can be potentially treated efficiently with this isoflavone, and which other MPS types could also be treated by this method
(iii) test derivatives of genistein, which can cross the blood-brain barrier more efficiently than this natural isoflavone, for their potential to inhibit GAG synthesis - this may lead to discovery of an even more effective drug for MPS
(iv) test efficacy of genistein (and its derivatives) in experiments with MPS III mouse model - this will be necessary to evaluate efficacy of this treatment by precise biochemical, histopathological and other tests.

Nevertheless, taking into consideration that: (i) Sanfilippo is a progressive disease, (ii) currently there is no specific treatment of this disease, (iii) life span of MPS III patients is shortened, (iv) results of in vitro studies with genistein are promising, and (v) genistein has been shown previously (by others) to be well-tolerated (with no serious side effects), an application to a bio-ethical committee for agreement to perform a pilot clinical trial with genistein in Sanfilippo disease has been made. An approval from the bio-ethical committee in Gdansk (Poland) was obtained.

Thus, a pilot clinical trial has been started, in which a genistin-rich soy isoflavone extract is used. In this open-label, one-center trial, the soy isoflavone extract is administered orally at the dose corresponding to 5 mg genistein per 1 kg of body weight daily. The following parameters are measured/investigated: urinary heparan sulphate level, hair morphology and cognitive abilities (using the BAE test). A questionnaire is also sent to parents, who should estimate any changes they observed in their children. Though preliminary results of this trial are encouraging, the trial is still ongoing. The results of the one year study should be available in January 2007, and then it will be possible to perform the first evaluation of this method of treatment.

An international team of experts in the field of MPS, who are interested in gene expression-targeted isoflavone therapy for Sanfilippo disease, has been established recently. The current plan of action is that Prof. Grzegorz Wegrzyn will report results of the one year pilot clinical trial to this team (hopefully in January 2007), and based on this report a decision on possible further trials will be made.

Current members of the “Genistein team” are (in alphabetical order): Michael Beck (Germany), Joseph Muenzer (USA), Nicole Muschol (Germany), Merce Pineda (Spain), Maurizio Scarpa (Italy), Shunji Tomatsu (USA), Anna Tylki-Szymanska (Poland), Kurt Ullrich (Germany), Grzegorz Wegrzyn (Poland), Ed Wraith (UK).

Amicus Therapeutics commences Phase 1 Clinical Trials for AT2220 for Pompe Disease

Cranbury, NJ, DECEMBER 14, 2006 – Amicus Therapeutics, a biopharmaceutical company developing small molecule, orally-administered pharmacological chaperones for the treatment of human genetic diseases, today announced that it has commenced Phase 1 clinical trials for AT2220 for the treatment of Pompe disease, following acceptance of an investigational new drug application (IND) by the U.S. Food and Drug Administration (FDA).

Pompe disease, also known as glycogen storage disease type II or acid maltase deficiency, is a relatively rare lysosomal storage disorder caused by an inherited mutation in the lysosomal enzyme á-glucosidase (GAA). GAA is normally made in the endoplasmic reticulum where it is properly folded and subsequently trafficked to the lysosome where it catalyzes the breakdown of glycogen. In many Pompe patients, a genetic mutation alters the structure and stability of GAA which results in reduced levels of enzyme in the lysosome and reduced cellular activity. The deficiency in GAA activity leads to excessive glycogen accumulation in the cells of various tissues, especially in heart and skeletal muscle.

AT2220 is a small molecule designed to act as a pharmacological chaperone that specifically binds, stabilises, and facilitates the proper folding and trafficking of GAA to the lysosome, where it can perform its normal function. AT2220 has been shown to increase GAA activity in cell lines derived from Pompe patients and in transfected cells expressing misfolded forms of GAA.

“We are very pleased to see continued progress in the fight against Pompe disease,” says Dr. Sharon Hesterlee, Vice President of Translational Research at the Muscular Dystrophy Association (MDA). “We look forward to exploring the opportunities to work with Amicus as this new potential treatment option for individuals and families with Pompe disease is evaluated through human clinical trials.”

“AT2220 for Pompe disease is the third Amicus product to enter clinical trials,” says Donald Hayden, Amicus interim President and CEO. “This accomplishment further demonstrates the company’s progress in developing new potential treatments for important diseases using pharmacological chaperone technology.”

The company’s lead compound, Amigal™ (migalastat hydrochloride), is in Phase 2 clinical trials for Fabry disease and AT2101 is in Phase 1 clinical trials for the treatment of Gaucher disease.

About Pompe Disease

Pompe disease affects an estimated 5,000-10,000 patients worldwide and is clinically heterogeneous in the age of onset, the extent of organ involvement, and the rate of progression. The early onset form of the disease is the most severe, progresses most rapidly, and is characterised by musculoskeletal, pulmonary, gastrointestinal, and cardiac symptoms that usually lead to death from cardio-respiratory failure between 1 and 2 years of age.

The late onset form of the disease begins between childhood and adulthood and has a slower rate of progression that is characterised by musculoskeletal and pulmonary symptoms that usually lead to progressive weakness and respiratory insufficiency.

About Amicus Therapeutics

Amicus Therapeutics is a biopharmaceutical company developing novel, oral therapeutics known as pharmacological chaperones for the treatment of a range of human genetic diseases. Pharmacological chaperone technology involves the use of small molecules to restore or improve biological activity in cells by selectively binding to misfolded proteins caused by genetic mutations. Amicus is initially targeting lysosomal storage disorders, which are severe, chronic genetic diseases with unmet medical needs. Amicus is currently conducting Phase 2 clinical trials for its lead compound, Amigal™, for Fabry disease, and is conducting Phase 1 clinical trials of AT2101 for Gaucher disease and AT2220 for Pompe disease.

Genzyme and Healthcare at Home Reach Out-of-Court Settlement

Press Release, 29 December 2006 - Genzyme UK and Healthcare at Home announce that they have reached a final agreement today in settlement of the Competition Appeal Tribunal case relating to Genzyme’s former homecare business in the UK.

Paul Drohan, Vice President and General Manager for Genzyme Therapeutics in the UK and Ireland said: “We are pleased to have reached an amicable settlement with Healthcare at Home. This has been a long and complex case, and throughout we have always acted in the best interests of patients with Gaucher disease. We are looking forward to putting this issue behind us and working with Healthcare at Home to ensure that Gaucher patients continue to receive the best possible care.’

Mike Gordon, Chief Executive of Healthcare at Home said, ‘We very much welcome this settlement. Our homecare service, which involves our highly skilled nursing team training and encouraging patients or carers to self-administer Cerezyme in their own homes greatly increases their autonomy and improves their quality of life. The settlement today ensures we can continue to provide this service to patients at no cost to the NHS while sustaining our business.’

Both parties have agreed that no further comment on the case or settlement will be made beyond this statement. This case was initiated in 2005.

AAV Gene Therapy for MPS IIIB in Mouse Model

Haiyan Fu, PhD., Center for Gene Therapy, Columbus Children's Research Institute, Ohio State University

Mucopolysaccharidosis (MPS) IIIB (Sanfilippo B) is an inherited disease caused by a deficiency in the lysosomal enzyme, -N-acetylglucosaminidase (NaGlu), which is needed to break down cellular waste products. The lack of the enzyme causes accumulation of glycosaminoglycan (GAG) in cells. Patients with MPS IIIB suffer severe progressive neurological disorders, leading to high mortality and premature death. No treatment is currently available for MPS IIIB.

Our research is to develop gene therapy to treat MPS IIIB. We used a harmless, adeno-associated virus (AAV), as a vehicle to carry a normal human NaGlu gene into NaGlu deficient cells and animals. The advantage of AAV gene therapy is that it leads to a long term production of the enzyme within cells so that repeated treatment may not be required. Our goal is to translate these treatments to MPS IIIB patients.

The greatest challenge in our research is that the disease is manifested through the entire central nervous system (CNS), including both the brain and spinal cord. To effectively treat MPS IIIB, we have to be able to deliver the AAV vector to all areas of the CNS, which is made difficult by the presence of the blood-brain barrier. Because this barrier prevents most large molecules, such as the AAV vector and NaGlu enzyme, from entering the CNS, our major focus has been to find ways get around or through the blood-brain barrier to deliver AAV vector to the entire brain and spinal cord.

Using the mouse as model, we have developed two non-surgical procedures, intravenous (IV) infusion and intracisternal (IC) injection, to deliver AAV vector into mouse CNS. We used an IV infusion of mannitol, a routine medical procedure for humans, to temporarily disrupt the BBB. This resulted in widespread distribution of AAV vector in the CNS and offered us an effective vector delivery strategy for MPS IIIB gene therapy.

Over the last few years, we have used these procedures to treat MPS IIIB mice with AAV vector carrying the gene for human NaGlu, which makes the enzyme that is missing in MPS IIIB patients. We were able to significantly increase the survival of MPS IIIB mice to 9.2-15.9 months (IV injected), 10.3-21.5 months (IC injected), and 11.1-19.5 months (IV+IC injected). Untreated MPS IIIB mice only lived 7.9-12.0 months, while normal mice live longer than 2 years. We also improved the behavioral performances of MPS IIIB mice with an IC or IV+IC injection of AAV vector. The production of NaGlu enzyme in cells is long-term, though the mice only received one treatment. These are very exciting research developments. We believe that these AAV gene therapy procedures greatly slowed down the progress of the CNS disease in MPS IIIB mice, though it did not cure the disease completely.

We feel that we have a therapy that provides meaningful benefits in our MPS IIIB animal model, and that we are ready to develop a translational research program leading to clinical trials in MPS IIIB patients. We are currently working on improving the therapeutic efficacy of our procedures and planning to put together an application to FDA for future clinical trials.

In addition, we believe that these procedures may also be used to deliver therapeutic materials for treating other types of MPS III, and other lysosomal storage diseases with CNS disorders, as well as many other neurological diseases.

Can you help with an international exchange?

Maria is a 13 year old girl who has Morquio disease. She lives with her mother, step father and four brothers and sisters in a small village near Linz in Austria. Maria is learning English at school and would like to improve her English by spending some time with an English family who have a son or daughter with Morquio disease.

Maria had a cervical fusion at the Royal Manchester Children's Hospital when she was four years of age and a leg-correction four years ago. Maria is a sociable girl who is keen to do well. Maria has a few needs; she can only walk short distances but does have a specially adapted bike to increase her mobility and she is very able to climb up and downstairs. Maria finds it difficult to reach up in order to comb the back of her head and to squeeze toothpaste onto her brush but is otherwise able to manage her personal care. Maria can go to the toilet alone but does occasionally need assistance. Otherwise Maria is a very independent and happy girl.

Maria's mother, Michaela Weigl, who runs the Austrian MPS Society would be very pleased to reciprocate with a return visit to Austria in 2007/08. If you can help please email Michaela at **office@mps-austria.at** or contact Christine at the MPS Society at **c.lavery@mpssociety.co.uk**.

Maria MPS IV

MPS Conference

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