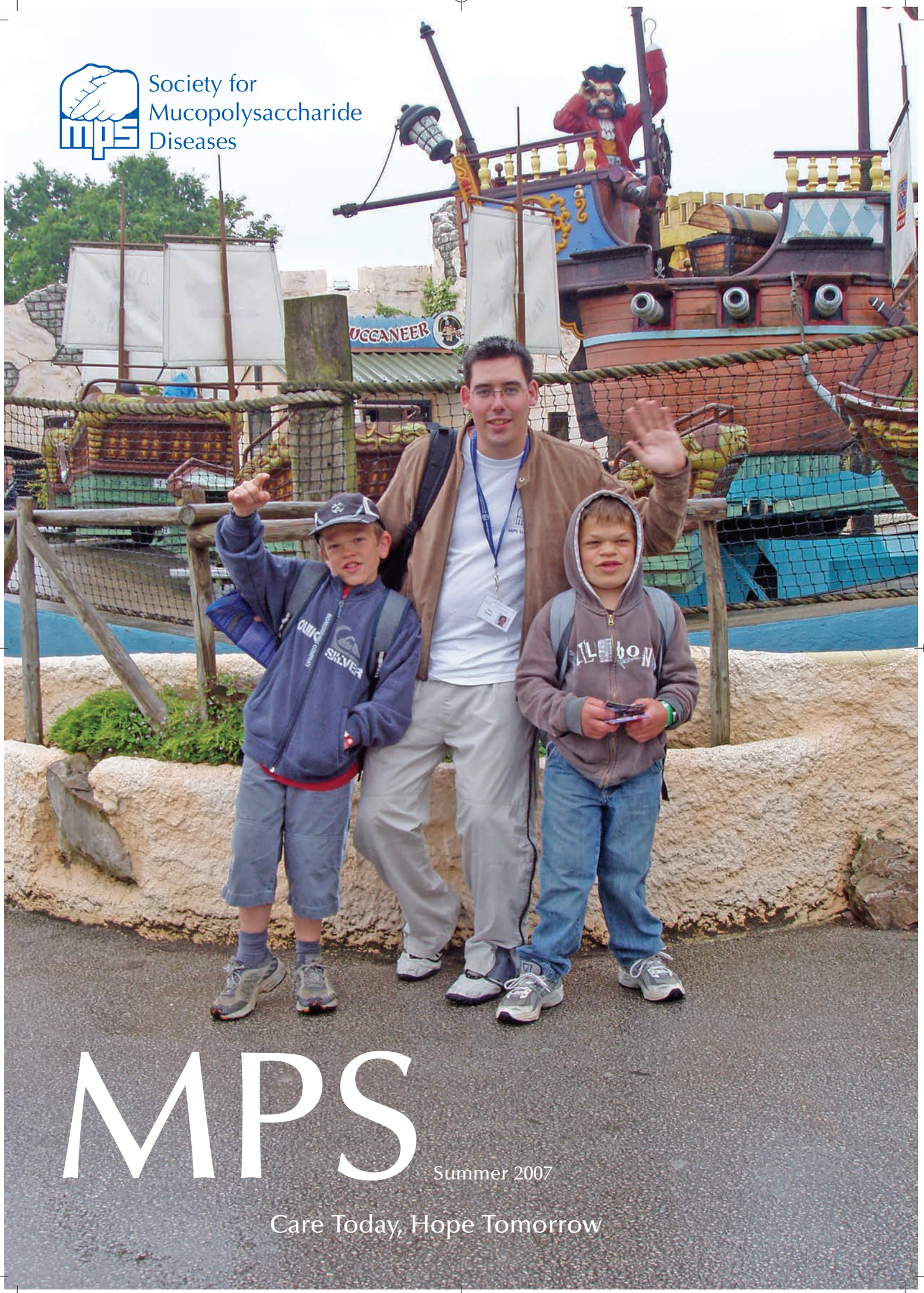
Society for Mucopolysaccharide Diseases



MPS

Summer 2007

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

CARE TODAY HOPE, TOMORROW

Cover photograph: Thomas and Louis Garthwaite, and Alex Smale at the MPS Conference 2007

MPS Magazine

MPS Society

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Newsletter Deadlines

Autumn	1 Sep 2007
Winter	1 Dec 2007
Spring	1 Mar 2008
Summer	1 Jun 2008

Become a Friend of MPS

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

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CHIEF EXECUTIVE'S REPORT

The past three months have been incredibly busy for the MPS Society and in particular the Staff Team as we built up to our 25th Anniversary National Weekend Conference that took place 30th June - 1st July 2007. One hundred families and over a hundred professionals joined us for the weekend along with nearly 70 childcare volunteers. You can read all about the Conference Weekend and the fun everyone had at the Gala Dinner despite the rain later on in this magazine.

The support of so many made this weekend possible and I want to say a huge thank you to the speakers and the volunteers, the Hilton Northampton, the Towersey Morris Men and our sponsors. For personal reasons I was not able to be around much in the two weeks prior to the Conference and offer my deep appreciation to every member of the staff team who worked tirelessly to make the Conference Weekend an amazing success for the Society's members, their children and all the professionals attending.

I wish to say thank you to all those who held fundraising events and raised awareness on MPS Awareness Day, 15th May 2007. This was the first time such a day had been celebrated in the UK and we hope year by year we will be able to gain greatly increased awareness of MPS throughout the United Kingdom.

Those who were at the Conference and the families who have been supported by Sophie Thomas over the past few months will be delighted to know that Sophie who is Senior Advocacy Officer had a baby boy, Harvey James Thomas on 5 July! Sophie went on Maternity Leave on 2 July and we send our best wishes to Sophie and her husband Phil as they become parents for the first time. As part of the Society's Strategic Plan it identified the need to appoint to the advocacy team an individual to work primarily with our members where a child or an adult is in the palliative care phase of their disease. I am delighted to announce that the John Ellerman Foundation have awarded the MPS Society a grant to fund this position over the coming two years and we are now actively seeking to recruit to this important position that will also provide a support service to our members pre and post bereavement.

To the nearly 300 families whose loved ones are remembered in the Childhood Wood I am pleased to announce that the final phase of the Childhood Wood's development plan has taken place. The Society is most grateful to the Geoff and Fiona Squires Foundation who so kindly gave a grant to fund the 2007 updates of the Memory boards, the woodland carved animals, the Remembrance Day and forthcoming planting. The Childhood Wood is an amazing ecological tribute to all the children and adults who have lost their lives to MPS, Fabry and Related Diseases.

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 Meetings held on 23-24 March and 8-9 June 2007.

Governance

The Chairman confirmed that Angela Brown had resigned as a Trustee due to ongoing family commitments and having been unable to attend the last three Trustees meetings. Angela was thanked for all her hard work and support of the MPS Society.

Personnel

Chris Murphy joined the advocacy team on 10 April 2007 and Sue Cotterell joined as Event and Volunteer Co-ordinator on 2 April 2007. Richard Jones continues to volunteer on the MPS Registry.

Access to Clinical Management and Treatment

The Chief Executive advised Trustees that MPS had been successful in helping a Scottish MPS II patient achieve funded ERT and is working to support a second Scottish child to receive ERT for MPS II as soon as possible.

Trustees also learnt that two Welsh members with Fabry Disease have also been awarded funded ERT after legal letters were sent.

MPS Annual General Meeting 2007

The Annual General Meeting of the Society took place at the Hilton Hotel, Northampton on Sunday 1st July 2007 at 9:00am preceding the Research Session of the Society's National Weekend Conference.

The minutes of the Annual General Meeting held on 29th April 2006 were distributed in advance to those members present and were accepted as true and accurate.

The Chairman, Barry Wilson, presented the Trustees report. This is published in the Annual Report and Accounts for the year ending 31st October 2006.

The Treasurer, Judith Evans presented the Statement of Accounts for the financial year ending 31st October 2006, the details of which are also to be found in the Society's latest Annual Report. It was proposed and seconded that the auditors, McLintocks and Partners, Chester be appointed the Society's auditors for the financial year ending 31st October 2007.

The Chief Executive, Christine Lavery presented the Society's report on generating income and thanked all the members and supporters for their efforts raising funds for the Society.

There being four vacant places on the Management Committee, Peter Conlin, Judith Evans and Bryan Winchester were elected without a vote.

There being no other business the Chairman thanked the members and guests for coming to Northampton and making the weekend such a success.

New faces at the MPS Office

Sue Cotterell

My name is Sue Cotterell, and I'm the new Event and Volunteer Co-ordinator at the MPS Society. I started working here in April this year, and on arrival I launched straight into helping to co-ordinate the National Conference which we held in Northampton, 29th June - 1st July. We had an excellent turn-out and hope that all of you who attended enjoyed it. It was good to meet some of you there and I look forward to getting to know more of you in the future.

I come from a teaching background, and decided to take on the role here as I became interested in the work of the MPS Society through my husband Steve. You may recognise his name as he has worked for the Society for just over a year as a member of the Advocacy Team.

When not working I am an assistant leader at a Brownie pack, and a keen quiz team member at our local pub.

I hope to see you at one of our events, some of which are coming up soon! If you would like to contact me my email address is sue.cotterell@mpssociety.co.uk

Chris Murphy

Hi, my name is Chris Murphy. I joined the team in April this year at the MPS Society as an Advocacy Support Officer. I will be covering the East of England including the London area. Prior to this job I worked for Windsor and Maidenhead Council for the past six years, I qualified as a social worker three years ago. I have worked in a very busy hospital team then in mental health, adult disabilities and finally as specialist worker in the older persons mental health team. I became very interested in carers and worked as Carers Champion for Social Services and was heavily involved with carers week and raising awareness and access to carers assessments and respite.

In my spare time my main passion is singing, but definitely not dancing as I have two left feet! Last year I started running to raise awareness and funds for the Alzheimer Society and raised over £4,000 in completing the New York Marathon last November. I got a lot of pleasure and support in trying to get younger musicians involved and we had several music events around the local area. I also love gardening, animals and general sports. Steve Cotterell and I will be running in this year's Great South Run so I hope we can raise some funds for the MPS Society. http://www.justgiving.com/steveandchris

I have had an amazing time already here and have been fortunate to meet so many families at the recent clinics. If you need any advocacy support you can contact me on the main office number or you can e-mail me on c.murphy@mpssociety.co.uk

New Members

Miss Suzanne Mallah has recently been in contact with the Society. Her son Kamal Hoteit has a diagnosis of Morquio Disease Type A. Kamal is two years old. The family live in the South East of England.

Mr and Mrs Houghton have recently been in contact with the Society. Harvey Houghton has a diagnosis of Hurler Disease. Harvey is five months old. The family live in the North West.

Mr and Mrs Vickery have recently been in contact with the Society. Their daughter Olivia has recently been diagnosed with Morquio Disease. Olivia is eight years old. The family live in the South East.

Robert Woodall has recently become a member of the MPS Society. Robert has a diagnosis of Fabry Disease. He lives in the London area.

Ms Drinkwater has recently been in contact with the Society. Her son, Aaron, has been diagnosed with MPS II, Hunter Disease. Aaron is nine years old. He has been receiving ERT since January 2007 and is doing very well.

Emma Morrice and Steven Robertson have recently been in contact with the Society. Their daughter, Jess, has a diagnosis of Hurler Disease. Jess is one and a half years old. The family live in Edinburgh.

Mr and Mrs Afzal have recently been in contact with the Society. Farzooqa has a diagnosis of MPS I Hurler Scheie Disease. Farzooqa is 24 years old. The family live in the South West.

Mr and Mrs Flower have recently been in contact with the Society. Oliver has a diagnosis of Hurler Disease. Oliver is three and a half years old. The family live in the South East. Oliver was diagnosed at five months old. He has two older brothers, Connor aged 7, and Owen aged 4, neither of whom have MPS.

Mr and Mrs Keighley have recently been in contact with the Society. Their son, Isaac, has a diagnosis of Hurler Disease. Isaac is 15 months old and the family live in the South East.

Deaths

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

David Mennim who suffered from Fabry Disease and who died on 25 April 2007 aged 36 years.

Daina Green who suffered from Hurler Disease and who died on 24 May 2007 aged 11 years.

Jack Robinson who suffered from Hunter Disease and who died on 11 June 2007 aged 11 years.

Sophie Richards who suffered from Hurler Disease and who died on 21 June 2007 aged 7 years.

Harold Morgan who suffered from Fabry Disease and who died on 30 April 2007 aged 71 years.

Many of our members will be saddened to learn of the death of Denise Brown. Denise, who died on 15 April 2007 in Wakefield, fought a brave battle against cancer. Denise was the mother of Adam Brown who died from Sanfilippo Disease in 2004. Our thoughts are with Denise's partner Derek Broughton and his son and daughter Myles and Joanne.

Congratulations to **Sue Peach**, MPS Trustee, on becoming Mayoress of Rugby.

Many congratulations to Sophie Thomas, Senior Advocacy Officer, and her husband Phil, on the birth of Harvey James Thomas weighing 7lbs and 1oz.

Daniel's wish comes true

After a long wait and many problems along the way Daniel's wish was finally completed with the help of Neisha Hall from the MPS Society.

We wished for sensory equipment for Dan's room, and Make A Wish used Spacekraft to supply the equipment!

We were off to Manchester the day the equipment was fitted so luckily it was completely hassle-free and was finished by the time we got home. The next day we were taken by limo to Frankie & Benny's for a lovely meal.

Dan liked it inside the limo as there were lots of lights and loud music. Unfortunately we were caught in traffic and Dan was snoring by the time we arrived! Elle (Dan's sister) managed to entertain us there and back and thoroughly enjoyed herself. Dan pulled himself together and managed to put away quite a few courses!

The room has been great for Dan, as he now doesn't have great mobility and not alot grabs his interest. He loves just chilling out with the sensory equipment and watching TV.

Elle also benefits turning the room into a disco/party room, with loud music and singing on the microphone, which seems to highly amuse Dan! He also got a big soft play chair which he loves. Hopefully Dan will continue his enzyme replacement therapy as well as it has been going and we'll have him boom-booming with Elle once again!
Elaine Muers

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

A new playroom for Jordan

I want to take this opportunity to say a big thank you to the MPS Society for helping us with this task. From the very beginning you have been fabulous and we would not of known where to start if it had been for you. Jordan is over the moon with his new playroom and enjoys the ballpond, especially the way he likes to see how many he can throw across the room!

Jordan is five years old and he was diagnosed with MPS III, Sanfilippo Disease, Type A two years ago.

It can be very lonely when you have a child with a disability, but having everyone at the MPS Society makes such a difference. You helped us with finding the funds to equip Jordan with soft play equipment and it is so much safer and a lot more fun for him and his brother and sister.

I would also like to thank REACT as it was them who made our little boy's dream come true by awarding us with the funding. They are a wonderful organisation and are to be cherished. **Wendy Lacey**

2007 Dates for your diaries!

27 July	BMT Clinic Manchester
31 Aug - 2 Sept	Sibling Weekend
15 September	Chessington World of Adventures
2 October	Bristol Clinic
13 October	London Aquarium/ London Eye
19 October	BMT Clinic, Manchester
19 October	Childhood Wood Tree Planting
18-21 October	International Fabry Conference, Munich

The MPS Society would like to congratulate **Pauline and Stephen Harvey** who Chris and Steve recently met at the Cardiff clinic with their daughter Abigail on their new 5 month old daughter Jennifer.

Hi! My name is Mike and I have Morquio's

Michael and his family at London Zoo in 2006

M ike
O yawale
R uns slowly
Q uick reactions
U nique
I ncredibly bright
O utstandingly brave

Hi! My name is Mike. This is a story about me: what I like, what I don't like and my condition that I like to call 'Hippo'. I am eleven and live with my mum, my two brothers (Abe and Joe), my sister Rachel and my granny.
Abe is ten and he is very athletic. Joe is eight and is good at singing. Rachel is only seven, she loves dancing.

I have a condition called Morquio. It is very complicated and I prefer to call it 'Hippo' because it affects my hips and like hippos, I don't run very fast on land, but I can move very fast in water!

'Hippo' slows me down and stops me from being active. It is very painful, but I have got used to it. I have pain in my legs all the time, especially in my hips. It is the most painful in the night time. Unless I am having a very good day, the pain won't go away. I also have carpal tunnel syndrome which means that I sometimes get pins and needles in my hands and they ache when I write.

Some things that I like are …
Spaghetti Bolognese and pounded yam (it's an African food, it's savoury and it's yummy)
Playing on my playstation
Basketball and Football are two things I really enjoy playing. I like them because I can pass the ball instead of having to run.
I like to play 'it' and 'dodge ball' with my friends.

Here are some things I don't like…
Brussel sprouts, because they are green, slimy, putrid, and they make me think of green mush! There's nothing to like about brussel sprouts really!

I also don't like going up the stairs because it hurts. The stabbing pain in my hips is like a hammer with spikes that keep hammering into me. The pain in my hips also slows me down when I run.

A good day!
When I wake up and am able to walk, it's already a good day! If my legs feel fine at the end of the day, that's a very good day! If I can play football and dodge ball, play games with my friends and have fun without pain, it's a great day!

A bad day!
It's a bad day when I can't get out of bed, I can't move my legs and I can't walk. When this happens, I need to push myself out of bed and crawl. My brother Abe or Joe will help me and call mom. On a bad day I can't play with my friends or have fun and I go to bed early. Sometimes mom gives me pills and it helps me.

My wishes...
I wish I could walk and run properly.
I wish all my teachers were aware of my condition.
I wish I could sit inside the classroom when I am having a bad day or there was a bench or chair I could sit on outside.
I wish I could use the lifts at school so I wouldn't have to get pain in my hips from walking up the stairs.
I wish I could eat my lunch at the junior school hall everyday and not have to walk to senior school for lunch.
I wish I didn't have to run for two minutes in gym class.
I wish I didn't have to do extra writing as a punishment and I could have two minute breaks when I write.

One day, I would like to be an Inventor and I would like to invent transport that helps people move more quickly. I have an idea, but it's a secret!

Brain illness boy Jordan dies

A 15 year old boy, believed to be the only person in Cumbria with a rare brain disease, has died.

The family of Jordan Walker, who died in his sleep on 12 March 2007, paid tribute to their loving son who they say will always be with them.

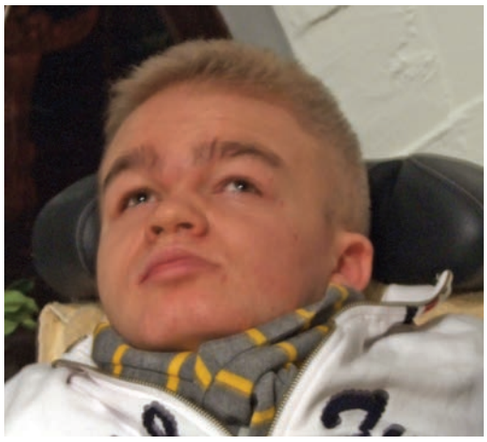
Jordan, from Frizington, was diagnosed with a degenerative condition, Sanfilippo, when he was seven years old. The disease, a brain enzyme imbalance, gradually attacks the brain cells, causing the sufferer to lose memory and motor skills.

Jordan's parents, John and Sharon, said although they knew their son's life would be cut tragically short it had still come as a devastating shock.

John, 55, said 'It's going to be difficult to cope but Jordy's reached out to us that much that even though he has gone he has only physically gone and he will always be here.

'We've had some fantastic times with him and he will be missed terribly. He has given us that much love and affection.' Sharon, 43, said Jordan had touched everyone 'in a magical way'.

'Even though he couldn't speak to you his aura just dragged you in', she said. 'He was only happy when someone was sitting with him to hold his hand. He just loved contact like hand and foot massages. He was an absolute love.'



John added 'I could tell Jordan I loved him and although he couldn't speak he could hear and tell me back just by his eyes'. Everybody he met, whether it be a doctor, a nurse or just people in the street, he could find an affinity with them. He could mentally reach out to people and draw them in because he was so loving. He was a loving child.'

They said many of Jordan's carers, including staff at Eden Valley Hospice and student nurses who visited him at home, had been moved by their loss and sent personal messages about how the teenager had touched their lives.

Jordan's family has appealed for donations or fundraising ideas for the MPS Society to help fund research into the condition.

Article by Daniel Cattanach appears couresy of the News and Star www.newsandstar.co.uk

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**

Charity marks 25 years of supporting families

Before the Society for Mucopolysaccharide (MPS) Diseases was established 25 years ago there was very little advice available for families with children who had the condition.

But now more than 1200 families all over the UK get vital support from the Amersham-based charity.

Since it started in 1982 the Society has also raised more than £6m and has supported the development of five new drugs to aid control of the disease.

The organisation continued its work on its 25th anniversary on Tuesday offering a drop-in centre and a play bus to entertain the children whilst families gained access to information and advice. The charity was started up by Christine Lavery when she was trying to get help for her son who had the condition. She said she had been writing to Mencap when her son died after choking on a piece of lego at school. Mrs Lavery added 'by that time I had heard from 40 families and we felt that we needed to carry on and set up the charity. We started up in Little Chalfont as there were a few families around including my own who had children with MPS and we were told we were the only ones. But we supported 40 families in the UK and today support over 1200 and we are able to help many more families now because there is greater awareness, whereas in the past MPS diseases were often pushed under the carpet because families were so devastated.'

MPS affects 1 in 20,000 births and causes physical disabilities and in many cases degenerative neurological diseases which generally result in childhood deaths. Mrs Lavery explained because of the inability of the person affected with the disease to break down waste products in their cells, the disease affects organs and the skin which physically impairs their appearance and they do not have any resemblance to their families.

The disease is also associated with a lot of discomfort because of the way it affects the joints and the organs. The charity supports families whose children have MPS through advocacy and by working with medical health professionals and social workers assisting famlies to get help with issues such as their child's educational needs and disability benefits. Mrs Lavery added when you are living 24 hours a day and seven days a week with a child who has MPS it is very difficult to deal with the situation especially as the disease is so complex. People can feel very frightened and so having an advocacy team with expert knowledge can help alleviate that anxiety and give practical support as well.

Articles and photos appear courtesy of
The Buckinghamshire Examiner
www.buckinghamshireexaminer.co.uk

MPS Awareness Day
15 May

Each year the Society will celebrate MPS Awareness Day on 15 May. This will be a day devoted to raising awareness for the group of 21 rare, genetic diseases known as MPS and Related Diseases.

Mothers raise awareness for genetic disease

Two mothers whose daughters are surviving an incurable genetic disease against the odds are spreading awareness of the condition to prevent future generations experiencing the pain their families have suffered.

Fer Pidden's 26 year old daughter Natalie was expected to die in her teens from Mucopolysaccharide (MPS), a cruel life-limiting disease which causes physical disability and mental deterioration, often leading to death in childhood.

Jackie Chisling's 14 year old daughter Hannah is a victim of the same disease, and like Natalie, is confined to a wheelchair and has to be fed through a tube.

Mrs Pidden, of Westbury Leigh, said: 'You never accept that this has happened to your child, you just learn to live with it.'

'In a way you tell yourself that you want it all to be over, but when you do that you start to feel guilty for thinking in that way.'

'Natalie has done so well to continue as she has for so long, because in my head I have been planning her funeral for many years.'

'Now I just want her to be as comfortable as possible and I hope for the best for her.'

Every eight days a baby is born in the UK with MPS or a related disease. As there is no cure, the best sufferers can hope for is treatment for symptoms as they arise.

Symptoms, which vary in severity and depend on the type of MPS a person is suffering, range from frequent ear and chest infections in the early stages, through to speech loss, growth restriction and a whole variety of disabilities as it progresses.

However, modern advances are improving the lives of sufferers. Prenatal testing is available to determine if a foetus is affected by the disorder, and in some cases bone marrow transplants can improve a patient's quality of life.

Enzyme replacement therapy is also a treatment in certain cases.

Natalie was diagnosed with MPS III, Sanfilippo disease, at the age of three and a half, while Hannah was two years old.

Mrs Chisling, of Queen's Gardens, Hilperton, said: 'We were told when Hannah was diagnosed that 14 was about the top age children with MPS III could be expected to live to, so she is doing very well.

'At least one of the kindest things about the disease is that they don't know they've got it, but it is very tough for everyone involved, including the families, because people with MPS require 24 hour care.'

'When you find out your child has MPS it certainly puts life into perspective'.

The 15th May has been designated by the MPS Society as MPS Awareness Day, in an attempt to teach people about the disease and encourage them to raise funds.

The Society, which is celebrating its 25th anniversary this year, supports individuals affected by MPS, as well as their families, carers and professionals.

Both Mrs Pidden and Mrs Chisling have been involved with it for a number of years, and would like as many people as possible to find out about the work it does and support them. To find out more about the MPS Society visit www.mpssociety.co.uk

Article by John Ballard appears courtesy of the Wiltshire Times, www.wiltshiretimes.co.uk

MPS Regional Clinics

BMT Clinic

On 20 April 2007 Dr Ed Wraith and the MPS Society hosted a BMT clinic at the Royal Children's Hospital Manchester. Sophie and Chris attended the clinic which was very well attended with lots of children running around and playing together.

The children also got the opportunity of being given by Dr Wraith a selection of coloured flying magnetic pigs! As well as the crisps and chocolate fingers! We would like to thank everyone involved in organising the clinic in particular Dr Wraith, Jean and Dot.

Photos clockwise from top right: BMT Clinic - Melissa McKie (MPS I), Sarah McKnight (MPS I), Northern Ireland Clinic - Aaryanna Lever (MPS IHS), Nathan Worsford (MPS II), BMT Clinic - Rachel Rothwell and Charlie Escalonilla (both MPS I)

Northern Ireland MPS Clinic

The Northern Ireland Clinic was held on 10th May at Antrim Area Hospital. As usual, another busy day was scheduled but thankfully all the families were prompt! The day went very smoothly and I know that all the families appreciated their time with the Dr Fiona Stewart and Dr Ed Wraith.

Cardiff MPS Clinic

The Cardiff Clinic was held on 11th May at Cardiff University Hospital. The day started fairly quietly with families arriving for their appointments with Dr Shortland and Dr Wraith. As the day went on the clinic area became very busy with what seemed like hundreds of children running around the waiting area! All in all it was a successful clinic and our thanks go to Dr Wraith and Dr Shortland for their commitment and time.

Photos clockwise from top right: Northern Ireland Clinic - Aaron and Dean Doherty (both MPS III), Cardiff Clinic - Megan Rennoldson (Mannosidosis), Summer Locke (MPS III), Abigail Harvey (ML III), Northern Ireland Clinic - Jade McAfee (MPS III)

Howletts Zoo

We are an isolated group of families in the South East but on Saturday 28 April 2007 we met up at Howlett's Zoo for a day out. We are the Gremo family, Nurse family and Perfect family.

We woke up to a beautiful day and were really happy to be visiting the Zoo as the last time had been for mine and Nathan's birthdays in November and it had been a really good day where we saw all the animals. True to form, as the weather was warm, lots of animals were asleep or in their houses making it difficult to find them. Nevertheless we still saw some amazing elephants and gorillas. Nathan and I watched the programmes on BBC last year and really enjoyed them so we intended to get the most out of the visit.

We already knew the Nurse family so met them at the entrance but had not met the Perfect family and spent some time trying to locate them which amused alot of people as I kept asking families if they were the 'perfect family'. You can probably guess the answers I got! When we finally met up it was lunch time so we spent some time trying to get to get to know each other.

After lunch the others went off to see the baby gorillas but Nathan and I couldn't because there was a notice requesting people to be quiet and as Nathan can't read we had no chance! Instead we headed for the ice-creams, a good alternative I felt. We all then made our way to the exit reluctantly after a really enjoyable day making new friends. **Janet Gremo**

We just wanted to send you a note to thank you for organising the South East family outing at Howlett's Zoo. The day went really well and it was nice to meet up with other families, such as the Gremo and Perfect families from the MPS Society. We look forward to seeing you at the next venue. **Rosemary and Harry Nurse**

Eurodisney, Paris

31 May - 3 June 2007

Just a quick note to say a huge thankyou for all your hard work. We, the Robinson's, had a fantastic time. It was our best and most relaxing holiday EVER thanks to all the volunteers who were amazing with not only Oli (MPS III) but all of us! It was such a relief to be with people who not only understood what we were going through but even mucked in and assisted at difficult times.

We are now looking forward to the conference so much more now we know a few faces. That said, I will understand if we are blacklisted as the most difficult family what with losing first our tickets to the Disney Park then our child!
Karen Robinson

Kate Murphy, age 12,
sister of Tara (MPS I H)

I'm just one of the many lucky people that were able to go on the trip to Disneyland Paris...

I am probably speaking for many of the participants by saying that the weekend was thoroughly enjoyed by everyone from the kids to the adults. I can remember seeing brilliant smiles all through night and day.

At night with people drinking at the bar (not to mention staying up until ... 3 o'clock … in the morning), the children definitely enjoyed playing pool, I have to admit that I did get beat a few times.

During the day the Park was amazing. The blue pass definitely came in handy, especially missing the long queues. Tara (my little sister who suffers from MPS I) also found the blue pass great as she got a photo and an autograph from the one and only Winnie the Pooh!

Many friendships were renewed, but many more were made. Everyone has so many different stories about their experiences with different MPS Diseases. It all made me think about how lucky we are with Tara's progress. We got very friendly with a lot of families including Phoebe, Lulu and Pat, and Lou, Adam, Elisa, Isaac, and many more absolutely amazing lovely people.

Our weekend at Eurodisney

It all started a week before 31 May 2007. The excitement was building up inside me. I was up very early that morning but it wasn't to pack as I had been packed already for about a week!

My brother Paul and I got a cab at 8:15am to Waterloo. Even though we live in London we still weren't sure what the traffic would be like. However, as you can guess, the roads were empty so we got there in plenty of time and had to wait around for what felt like hours. Waterloo looks like a normal train station but it is not, and you can't just get on the train and wait.

Finally it was time for us to go through to the trains after having our bags put through the x-ray machine. It was then a mad dash to the train, which isn't easy when it felt like 500 people were doing the same as you!

Once on the Eurostar I started to relax a bit and put my ipod on. It was a good journey and as we got nearer the Eurotunnel there was an announcement about how long it would take to go through the tunnel. It only took 20 minutes and it was quite strange to think that in 20 minutes we would be in France. Once in France there was a lot of countryside, which was very beautiful. However, it wasn't long before it was time to get off the Eurostar as we had arrived.

In the Paris station we had to wait around for a bit but then it was time to go and get on the Metro. We all headed to the lift which would take us down to the Metro, but it was out of order and there was no other lift! So we all had to use the escalators and this was a nightmare let me tell you! I found it very strange that no-one that worked at the station offered to help, whereas in the UK there would be at least one person that would offer to help if you are in a wheelchair.

We had to get two tubes to get to Bussy St George, our Metro stop for the hotel. Once we were off the Metro it was a little walk to the hotel and it was nice to be out in the fresh air.

As we got nearer the hotel all the kids were getting more excited as they all wanted to go swimming in the hotel pool. Once we had unpacked our bag Paul and I went and found the hotel bar! I asked for a Malibu and Coke; their singles are like our doubles so it was very nice! We then went and had dinner in the hotel and met the other families which was very nice. After a while Paul and I went for a walk and found another bar and we met some more families.

We got up really early the next morning (didn't know that time of day existed!). We soon got on the coach to take us to Eurodisney. Once we got through the gates we could see Sleeping Beauty's castle which was beautiful. We went to get our fast track passes which were a very good idea as it meant we could avoid the queues.

Then it was time for the rides and there was so much to see we really didn't know which to choose first. We went on the Star Wars stimulator and I was thrown around like a rag doll and held onto Paul for dear life! I did enjoy it, and we did so much but the one ride I really remember was the Buzz Lightyear ride. Ok, so it's for kids but it was a lot of fun and when we got back to the hotel that evening all the fathers were talking about that ride!

We saw the Disney parade which was really good because all the Disney characters were there and some of them came up to the crowds to shake the kids' hands. We went on the 'Honey I shrunk the audience' 3D show and those of you that went on it will know what I mean when I say 'mice'! As we were at the back we could see and hear people down the front screaming and raising their legs in the air! I couldn't feel it but I was told that as the mice run people could feel them run up the backs of their legs. When the snake came out of the screen I did shut my eyes!

When we got back to the hotel we had a bit of a rest then got ready to go out and find somewhere for dinner. We found a local Chinese restaurant and it was funny because there was a crowd of us and we overtook the restaurant! The meal was really nice and afterwards people were talking about going for a drink, but I was so tired I went back to the hotel and went to sleep.

The next day I felt refreshed and ready to go. We went to the Disney studios and saw how the special effects were made in films. We went into the Armageddon set to see how the effects were made in the film. It was done like we were part of the extras in the film and we were right in the heart of the effects. All of a sudden a ball of fire shot up in the air then cold water came down all over us. Then the floor dropped about an inch and a burst of cold wind came rushing in. It was very good and it gave you a good insight into how the effects are made and how real they seem.

We saw a stunt show with cars and motor bikes. First they showed you the stunts, then they showed you how they are done. So now when I watch a film with a car or bike stunt I will know how it is done.

When we got back to the hotel David, Sarah, myself and Paul said we were going to go back to the Disney park as it didn't close until 10pm. We got a taxi to take us.

We went back into the Disney village and we were going to see a Buffalo Bill's wild west show but it was fully booked so we went back into Disneyland. David and Paul went on Thunder Mountain while Sarah and I walked around looking at Sleeping Beauty's castle. It was all lit up and it was very beautiful.

Paul and David went into a shop to buy a Dumbo snow globe as the one I had bought earlier that day had broke. They asked the shopkeeper but she didn't know who Dumbo was, so David was indicating big ears with his hands while Paul was indicating an elephant's trunk with his arm up to his nose. She still didn't know who Dumbo was and she just kept pointing to Stitch! When they came out of the shop and told us what had happened we were all in fits of laughter. Then we all calmed down and found another shop where we managed to get the Dumbo snow globe.

We had something to eat in the rainforest café, surrounded by sound effects of water falls and moving elephants. David jumped every time the elephants moved, even when we gave him warning.

We had a very good evening and when we returned to the hotel we stayed up talking well in to the early hours of the morning.

I really enjoyed myself this weekend and have made some amazing friends that Paul and I will keep in touch with.
Larraine Mullen (MPS IV)

Eurostar and Eurodisney

The disabled way

My name is Tim, I am married to Sally and we have two children Will, 11 and Sophie, 10. During the recent half term break we went to Eurodisney for four days with the MPS Society and approximately 30 other MPS families, kindly funded by some of the proceeds from the Ollie G ball held last year.

Sophie has Sanfilippo disease (MPS III), she is losing her mobility and uses a wheelchair; sadly she has already lost her speech.

We were travelling to Paris by Eurostar from Waterloo. We were upgraded to first class as there is no room to accommodate a wheelchair in the standard section. This was definitely a more relaxed way to travel, and the train staff were extremely helpful.

On arrival in Paris we found our way to the hotel on the Metro with the rest of the group. Reaching the Metro at Gard du Nord was very difficult as each lift we encountered was either out of order or closed for maintenance, when we finally found a lift that worked we reached the required floor, but were stuck the wrong side of the ticket barrier! We finally made it through the barrier with the help of Neisha and some frantic hand gestures!

There was the usual MPS atmosphere at the hotel, the families certainly enjoyed it but I don't think the staff were quite prepared for the many 'pirate battles' that took place in the reception area and the gymnastic displays on the luggage carriers. Events became even livelier on the second day when many of the children had purchased pistols and cutlasses at Eurodisney for added authenticity. The evening meals were a great opportunity to meet other families and exchange stories and experiences. It is amazing how relaxed you can be with so many MPS children running around, perhaps it is not feeling so alone and seeing there are other families with similar challenges.

Our experience of Eurodisney was very positive, it was our first visit and we were uncertain how appropriate it would be for Sophie. The children had a super time and Sophie was made very welcome by all the staff and characters. The Disney characters made a bee line for her, which quite startled us the first time it happened. The chaperones were particularly good at bringing the characters to Sophie, she spent a few pleasurable minutes pulling at Pluto's whiskers! I think the highlight for Sophie was meeting Cinderella after the main parade. Our thanks must again go to Neisha for arranging this with the Disney staff at the last minute.

Many of the rides were wheelchair friendly or had adapted facilities for wheelchair use. Luckily for us, Sophie does not like the white knuckle rides so we were able to stick to the more sedate offerings, the tune from "It's a small World" will be forever in our minds. I am sure there will be a few readers smiling and thinking the same at this point!

Will went off with some of the other children and volunteers, provided by the MPS Society, he also had a wonderful time. Space Mountain was the best roller coaster and he was lucky enough to go on it twice. Indiana Jones was the next favourite, however, it broke down on their first go and they were lead off having only completed three quarters of it. He was delighted that they were given a fast pass for when it was fixed. Personally I'd have just walked away and never gone back! He loved the second ride!

You can obtain a 'blue card' from the information centre which enables you to bypass the queues. To ensure you get the card documentary proof is required of the disability, without such documentation it is very difficult to get the pass, even with a child in a wheelchair.

The medical centre was by far the best find, it was wonderful for changing and feeding Sophie. It was equipped with about a dozen beds with curtains and screens, similar to a casualty department. The staff there were very kind and helpful.

We had a lovely break which the children thoroughly enjoyed and we all came home very tired having had so much to see and do. Many thanks to Gina and the team for arranging and supporting such a wonderful trip.

Tim Summerton

Bonjour from Disney Paris

Small Boy:
Doctor Doctor I have MPS I Hurler-Scheie and I keep thinking abut Mickey Mouse, Donald Duck, Goofy and Pluto

Doctor:
Tell me how long have you been having these Disney Spells (groan and blame his dad)

Small Boy:
Ever since I got a letter from MPS House saying we'd won a competition to go to Eurodisney paid for by the wonderful Ollie G Ball.

The excitement in the house was unreal, we were actually going to Eurodisney. Wow!

Since getting the letter from the MPS Society we'd been working really hard to make sure our passports were up to date. The only real hurdle was travel insurance. Since Ben had had his shunt inserted in November last year (2006) (story to follow once I've written it), travel insurance has become a whole different ball game due to the increased risk of problems and other issues not to be discussed here.

We decided to travel from our home in beautiful Teesside to Manchester to catch the plane to where?…Oh yeah, Eurodisney.

We set off in good time with everything packed, passports, cases, glasses, MP3 players, books, money and of course the kids. Try as they might at MPS House they organised just about everything really well except they forgot to tell the people of Manchester to keep off the M62. Traffic on the M62 blocking the junction we needed to exit from meant the two-hour journey actually took nearly double that. Several phone calls to MPS House meant the airline knew we were coming. Again the brilliant staff at MPS House did everything except move the traffic out of the way to allow us to get through. We checked in with just 5 minutes to spare, wow close call.

We made our way round and met some new faces and some old faces and some very old faces. A brief chat before being boarded onto the plane to where?...
Oh yeah, Eurodisney.

The flight was quick and relaxed before touch down at Charles De Gaulle Airport then the usual chaos of getting your bags, watching the children and all the other fun that goes with airports.

Taxis to the hotel? Well, that's another problem to be overcome when five families arrive at the same airport and need to get to the same hotel. Some obviously needed access for less able-bodied children with wheelchairs. However, everyone seemed to get to the hotel ok and, as far as I am aware, no-one is still in Paris.

Once at the hotel it was a case of straight to the bar, well that's if you are still on an abstinence from alcohol that is, now 18 months and counting. Meeting up with familiar faces (mostly with bodies attached) and the chance to meet new families and share experiences. Also the opportunity to be with a group of people who know what it's like to live every day with MPS. I hope new families with their first experience of this type of gathering got as much out of the experience as I did, meeting and sharing stories of diagnosis and treatments and coming to terms with living with a child with MPS.

Dinner time came which was good because I was starving. The first night meal was hectic as would be expected for 120 quests and it was here that the organisational effort

of MPS staff and particularly Gina Page became apparent. I have worked in catering and organising a big party is an absolute nightmare, now increase that ten-fold by making sure that the people organising and liasing spoke different languages, very “Tower of Babel” stuff. It was also Gina’s 21st birthday. The meal demonstrated the effort that had gone into the enormous task of organising the trip and everything Gina had had to contend with.

After the meal more time to relax and swap stories and experiences. Some of us decided to retire to our rooms whilst others, who shall remain anonymous, kept the bar staff busy until the wee small hours (you know who you are).

Friday morning was packed with excitement and the atmosphere was electric as we waited for the buses to take us to …Oh yeah, what’s the place again… oh Eurodisney. Even the first sighting of a Disney road sign was greeted with enthusiasm by the children, and yes, some adults. And a short ride later and we were there at last.

The children virtually sprinted towards the main entrance and then in front of us we saw it, the main entrance huge in front of us, only one problem: “It’s pink, that’s a girls colour.”

A quick check of our bags and finally, we were in Eurodisney. The whole place was exciting. We spent the whole time rushing from one ride to the next trying to cram as many rides into the first day as possible, moving from one zone to the next. The rides such as Thunder Mountain Mining Company was Ben’s favourite, a fast ride round an old frontier town gold mining. Sadly the Indiana Jones ride was not working by the time we got round.

The rides ranged from the slow Mississippi Steamer round the frontier zone and The Small World, to the fast; Thunder Mountains Mining Company. When Sarah suggested Space Mountain 2 I should have known better, the fact that Ben was too small at 6 was the initial indicator, the huge structure with steam pouring out of the ride was another indicator, but it was only when I neared the front of the queue and saw, shoulder harness...

I advised Sarah, “Look I’m 42 years old, I don’t do shoulder harnesses”, but many accusations of cowardice later and I was sitting down on the ride and shoulder harness in place there was no going back. The ride itself was brilliant.

We experienced as much as we could;
Buzz Lightyear, where I shot as many aliens as possible and won on both occasions, not bad for an old man.
Thunder Mountain, again
Space Mountain again
Phantom Manor, a ride round an old mansion
Pirates of the Carribean, sadly for Tracey my wife no Johnny Depp or Orlando Bloom, she had to make do with me. The Funfair type rides of Teacups and merry-go-rounds.

Then we made our way round to watch the beginning of the big parade through Eurodisney. This was very exciting as the gates opened and out poured, Toy Story, Mickey Mouse and his friends, The Princesses and Princes, Alice in Wonderland. As they left we sprinted off for one or two more rides before heading home. All very tiring.

On the Friday we were given the choice of a coach back at 6pm or make our own way back by train, and we decided to try the train system. The ticket barriers proved a problem but we made it back, meeting up with another family at the station we again shared stories and experiences of MPS.

Back to the hotel for more children running about like idiots, having a great time, more adults drinking and socialising and having an even better time. Then bed.

Saturday
Off we go again, breakfast then the bus, whooppee.
The second day was warm, very warm.

We started at Disney Studio and had a ride on a magic carpet, Ben got to meet Chicken Little as well.

We then headed back to Disneyland proper and more rides. The warm weather and being a weekend meant more people and long waits on all the rides. We tried new rides and back to the old favourites as well. We finally got to go on the Indiana Jones Ride, excellent, I would really recommend it for anyone with a weak heart.

By 2 p.m. we were Theme Park Fatigued from racing from one ride to the next, so gentle walks round the amazing areas, Frontier Town, Discovery Land, Fantasy Zone (although they did not have my particular fantasy, and

I was nearly asked to leave Disneyland for suggesting they have it in for next time), Adventure Zone, the whole place was absolutely amazing.

Then we made our way round the town square to watch the end of the big parade through Eurodisney. We found Eurodisney exciting but tiring.

Once that was over we headed back to the coaches and the hotel. Dinner as before was hectic and we did need to wait, but that's what happens with any large number of people all eating together. One final run around for the children before bed. Some people managed a 4 a.m. finish, you know who you are.

The next morning was breakfast and waiting round for taxis to take us back to reality (sadly). The morning was a series of goodbyes at the hotel, the afternoon was a series of goodbyes at Manchester Airport.

Sadly Charles De Gaulle airport was a farce. Planes were cancelled, two ticket collectors checking hundreds of people in, one policeman on passport control for hundreds of people, all very stressful.

But once we were home we thought about the excitement, the experiences and the people and the airport was quickly forgotten.

I would like to take this opportunity to thank all the MPS staff team especially Gina Page (Happy 21st) for all the hard work and effort and stress they put into organising a brilliant event. Next time can you sort out the M62.

Peter Conlin
Father of Ben MPS I Hurler Scheie

Dear MPS

This message is long overdue but Aisha (MPS IV) has been unwell for a few weeks.

We would like to thank the MPS Society for the magical experience of Eurodisney. Without the Society we would probably not have been able to have this opportunity of visiting Disneyland Paris. Right from the start to end it was brilliant! We met with Gina, Antonia, Steve, Neisha and Sue at Waterloo Station. It was our first time travelling on a train with the girls and we were a bit worried whether we were going to be able to cope but the MPS team and the volunteers were great. They just took over and we felt very reassurred.

When we arrived in Paris we had a siuation that did throw us into a bit of a panic because of the lack of disabled access on the Paris Metro system, but with the terrific team of helpers we made it to our destination.

I have never seen Aisha and Saffiya so happy. They loved every bit of their wonderful trip to Disneyland Paris. Aisha felt so important every time we jumped to the front of the queues and being the centre of attraction with all her favourite Princesses! Thank you for such a wonderful experience.

Asma, Sharif, Aisha and Saffiya Seedat

National MPS Conference 2007

From 29th June - 1st July the MPS Society held its 25th Anniversary Conference at the Hilton, Northampton. Attendees came from around the UK including Northern Ireland, as well as from countries across the globe, such as Poland, Holland, Germany, the USA and Australia. There were 180 adults from MPS member families, over 100 children, 55 speakers, 66 volunteers and over 100 professional delegates.

The conferences

The Friday conference was on ‘Aspects of Fabry Disease’, chaired by Dr Atul Mehta, about aspects of clinical management, psychosocial issues and current and future treatment for those affected by Fabry.

On Saturday there were three symposia running simultaneously.

Conference A, on ‘Palliative Care’, was chaired by Dr Ashok Vellodi. It focused on the challenges of caring for children and adults with MPS conditions which cause degeneration of the central nervous system in childhood, and who become increasingly dependent on others for their care.

Conference B, on ‘Clinical Management and Treatment’, was chaired by Dr Ed Wraith, and focused on children and adults whose main difficulties are related to physical disease progression, and where the disease is usually associated with stability and survival into adult life.

Conference C, on ‘Rarest of Rare’, was a first-of-its-kind conference for those with the rarest of MPS and Related Diseases, and was chaired by Dr Fiona Stewart.

On the Sunday morning the AGM took place, followed by a conference on ‘Hope for the Future’, about developments in research, chaired by Professor Bryan Winchester.

Children's Programme

On Friday afternoon there were two Mad Science workshops, featuring candy floss and slime.
During Saturday the children enjoyed a trip to Drayton Manor Park, despite the rain! On Saturday evening there was a bowling trip for the older children, and entertainment at the hotel for the younger ones. On Sunday morning there was an outing to Wicksteed Park, and a trip to Sno!Zone for siblings over 7.

Other events during the weekend

On Saturday evening children and adults had fun joining in with some Morris dancing, courtesy of the Towersey Morris Men, and there was a funfair enjoyed by many (not just the children!). The 25th Anniversary Balloon Release took place on the Saturday for the children, and on Sunday afternoon for the adults - we shall see whose balloon travelled the furthest. Here are a selection of photos from the weekend.

CONFERENCE

Dear All

Many thanks for another successful conference. We found the whole weekend really useful and also great fun. So different from our first conference, in 2005, when we were so overwhelmed and distressed by it all (although the conference then, too, was of a very high quality).

We enjoyed so many of the talks, particularly the one on Sunday about the blood brain barrier, and felt that there was so much more happening for Sanfilippo than two years ago. We do hope that you realise how much we, and so many other families, value the work that you do.

All the best
Jessica & Tim Hooper

Dear Christine

I would be very grateful if through you I can write and congratulate your staff on the wonderful 25th Anniversary meeting that we enjoyed recently in Northampton.

It is a tribute to your team that they could pull off such a large occasion, attracting such eminent speakers for a 2 and a half day meeting.

Let us look forward to the next 25 years!

Dr Ed Wraith

The MPS Society would like to thank **Pat Pryce** from Cardiff for the two hand-made quilts she donated to the Society. The quilts went to two deserving MPS Member families.

Key note speech by Lady Shauna Gosling

at the MPS Conference Gala Dinner

Today is my grandson's 5th birthday and he's having a party with this friends, which all sounds very ordinary, except that it's not. It's a miracle, because Oliver has MPS I Hurler Disease.

Ollie was first misdiagnosed at 6 months old and when, shortly afterwards, the correct diagnosis was made, the prognosis was so bleak that his parents wondered if treatment was even worthwhile. As I cast my mind back, what stands out most clearly was despair, confusion and not knowing who to turn to or what to do. Ollie's parents, as you can imagine, were distraught. Their saviour was finding the MPS Society, who were able to advise them who to see and what to do, giving them support, help and understanding.

The MPS Society has played an important part in our family's life, and in the lives of all those families who are affected by this condition. When a child is first diagnosed, it is the MPS Society who are there with help, guidance and support and helping to organise practical things, such as adaptations that might be needed in the home, specialist equipment, respite care and fun family days out.

It is their slogan 'Care Today, Hope Tomorrow' that you can see what the MPS Society is really all about. As well as being a lifeline for MPS sufferers and their families, the Society is greatly concerned with the future, working tirelessly to raise public awareness of this devastating condition and to raise money for the research that gives sufferers hope for tomorrow.

So how important is research? The answer to this cannot be overestimated, as research is the one thing that gives MPS sufferers and their families hope for the future. Without research there would be no progress, no new therapies and treatments and no chance of a better quality of life. And it's important that we bear this in mind, particularly at this time because ground-breaking research is being carried out as we speak - all of which, of course, requires constant funding.

As a grandparent, when Ollie was first diagnosed, and during his treatment, there were times when I felt powerless to help. All I could do was be a source of strength and offer my support.

Luckily I come from a medical background myself, and I have a son who suffered from a serious medical condition when he was a child. That, and my experiences with various charitable projects, was a great help, but perhaps the best way a grandparent can offer support is by providing a fresh perspective. Being just one step removed allows you to stand back and view the situation more calmly and objectively, when the parents themselves are perhaps too emotional and upset to think clearly.

Once the decision to commence treatment was taken, Ollie and his family went to America so that he could have a bone marrow transplant. It was a traumatic time for all of us, particularly his parents, and at one point we nearly lost him, but with courage and tenacity, he came through and, almost unbelievably, he's now a boisterous five-year-old, living life to the full.

Regrettably, not every child is so lucky and not every treatment is successful. We still don't know what the future holds for Ollie, as he may well experience problems in the years to come, but we're all unanimous in the belief that it is important to live in the present and enjoy each day as it comes.

The only reminder we have of Ollie's condition are the regular check-ups he has at Manchester and words cannot express the gratitude and debt of thanks we owe the doctors and nurses who have made this possible. I am constantly inspired and reassured by their passion and dedication.

Ollie's illness was a strange experience for all of us - a journey that started out so full of negatives, but out of which we have drawn so many positives. The interesting thing is that we have all learned from the experience.

My youngest son, Ollie's father, was a charming wild child, a carefree soul with a devil-may-care attitude to life. Needless to say, Ollie changed all that, and through caring for him, David has become the most wonderful, devoted, hands-on father. During his treatment he stayed with Ollie night and day, helping and learning how to administer his medications, whilst also playing the role of the ordinary father - encouraging him, making him laugh, having fun and playing with him.

It gives me great pleasure to see David raising an amazing amount of money for other families to go on special outings and adventures, as well as donating to vital research projects.

So, apart from offering my thanks, I would like to congratulate the MPS Society on 25 years spent helping children and families living with MPS to have a better quality of life in so many different ways. The tireless and dedicated work of the Trustees and of Christine Lavery and her team is outstanding, offering us all hope, hope for a better future for our children and grandchildren.

MPS 25th Anniversary Grants

As part of our 25th Anniversary Celebrations the MPS Society, in recognition of the MPS specialist centres who have strived over the years to make a real difference in the lives of those affected by MPS, Fabry and related diseases, each of the centres were invited to submit a proposal of how they would spend £1,000 to benefit our members. Of the top ten centres invited, eight submitted proposals and the following grants were awarded:

Dr Atul Mehta
LSD Unit Royal Free Hospital, London
To purchase a 'Lazy Boy' leather reclining Rolls Royce model infusion chair and side table upon which all the infusion materials could be prepared.

Dr Anupam Chakrapani
Birmingham Children's Hospital
To equip a designated room in the outpatient department including padded walls to make the area safer for children with challenging or destructive behaviour to play or roam whilst the parents concentrate on the conversations taking place.

Dr Fiona Stewart
Belfast City Hospital
To improve awareness of the MPS disorder in the 7,000 members of the Northern Irish traveller community who may be at risk of MPS I (Hurler Disease) and Mucolipidosis II (I - Cell Disease). This community is highly consanguinous and there is a high incidence of these diseases. A leaflet specifically designed for the travelling community will explain the risks of having an affected child if there are affected relatives; options for carriers testing; as well as the early signs and symptoms of these diseases.

Dr Simon Jones and Gill Moss
Royal Manchester Children's Hospital
To purchase a portable pulse oximeter for home assessment of poor respiratory function and / or obstructive airway problems in children with Mucopolysaccharide and related diseases. This may assist the families of children in the latter stages of their disease who find travelling long distances to hospital and avoid admissions to hospital.

Dr Uma Ramaswami
Addenbrookes Hospital, Cambridge
To purchase a Sony PSP value pack and games and a Nintendo DS Lite to provide entertainment to children 3 - 16 years during their infusions and hospital admissions. Half the grant will be used to set up a small discretionary travel fund to assist MPS patients and their families suffering financial difficulty. Three quarters of this grant was donated by Juanita Davenport and family as requested, being the proceeds of a fundraising event.

Robin Lachman
Charles Dent Metabolic Unit, National Hospital, London
To purchase a fully adjustable Riser Chair allowing the seating position to be tailored to the individual patients. This chair also provides assistance in moving from a sitting to standing position and will greatly benefit individuals affected by Lysosomal Storage Diseases.

Judy Holroyd
Dept. of Genetics, University Hospital of Wales, Cardiff
To purchase a lap top computer that has access to the online medical and scientific journals and publications on genetics to the benefit of scientific and clinical staff in the Genetics Department.

The Society was delighted to be able to award these 25th Anniversary Grants raised in memory of Timothy Hope-Gill who lost his life to MPS I Hurler Disease following a Bone Marrow Transplant on Christmas Day 1999.

Photo left: Christine Lavery and Lady Shauna Gosling presenting the 25th Anniversary Grants

Bereaved Programme
at the Childhood Wood

On Saturday 30th June 2007 a number of families gathered at the Childhood Wood for a Remembrance Day as part of the MPS Conference weekend.

The day begun as we met at Rufford Country Park for lunch where we were served a hot meal and some delicious lumpy bumpy chocolate cake! (Some chose that instead of the apple and raspberry pudding just because of its name!). We then drove to Sherwood Pines in convoy to meet at the Childhood Wood.

The weather wasn't brilliant but some of us put on our wellies and others huddled under umbrellas to remember those children and adults who had passed away.

Barry Wilson, Chairman of Trustees, hosted the day and welcomed those gathered by explaining recent developments to the Childhood Wood such as the memory boards and also the latest edition of the beautifully hand crafted animals. The money for these was donated by the Geoff and Fiona Squires Foundation and made by Ben May at Forest Crafts in Devon. The animals are scattered around the path and many enjoyed them as they made their way around the Wood.

Barbara Hopwood from Australia read out the names of those being remembered and Wilma Robins read out the poem 'Remember' by Christina Rosetti. The balloon release went off almost without a hitch thanks to some wonderful dads rescuing the couple that initially got stuck in the trees.

Many thanks to Byron Wibberly at the Forestry Office for all his support and help with getting the woodland animals in the ground safely in time for the Remembrance Day. Also a big thank you to the families who were able to make it in the rainy weather to join us for a special day. I know I found the day very special and from what I have heard so did the families that came. **Miriam Blowers**

An update on Maria's surgery

Maria Wiegl has MPS IV, Morquio Disease and lives in Austria. Her mother Michaela runs the Austrian MPS Society.

Maria underwent surgery on 11 May 2007, and unfortunately it wasn't such a little thing they promised. The doctors made some large incisions, and placed the two plates, each with two screws. The doctor had said that Maria should be able to get up the next day, but no chance. She was in a lot of pain, and she only got up for the first time after four days. We stayed in hospital for one week, went home on the Wednesday and Maria was back in school the following week. We did have to carry her up to the third floor at school, but after that she was able to get around alone.

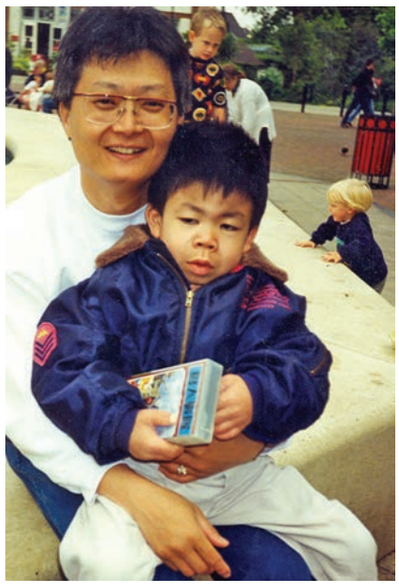
We have recently been back to hospital for an x-ray and the doctors were very pleased. I was a little bit nervous because the knee hasn't returned to its original size yet, and it still feels a little warm. However, the doctor said that this is normal and there is nothing wrong inside the knee. He thinks it is only some blood in the muscle which hasn't yet disappeared but it would go soon.

In the meantime Maria is riding her bike again, swimming, and able to get around with her scooter. No running, no jumping. However, she tried the trampoline in our garden and was very proud that she jumped about 2 centimetres!

I hope so much that the plates will work because the surgery was really painful for Maria and that shouldn't be in vain. She cried a lot in the first three days, and I wonder if this is because she didn't receive sufficient painkillers.

More than one month has passed since the operation, and if she develops well, doing her exercises every day, Maria will hopefully be back to her good condition in summer.

The picture left shows her first step! **Michaela Wiegl**



Eddie Chou

Some of our members will remember Eddie and Virginia Chou when they lived in the UK in the 1990s with their son, David, who had MPS II, Hunter Disease and died on 14 April 1997. When Eddie and Virginia returned to their native Taiwan they set up the Taiwanese MPS Society. It was my privilege to have attended their MPS Society meeting and clinic in Taiwan in 1998.

It is with enormous sadness that we learnt that Eddie Chou died suddenly on 15 May 2007 in his early 50's. Our thoughts are with Virginia and their daughter Julie and son Edward.

7th International Symposium on Lysosomal Storage Diseases

27th - 28th April 2007

Christine, Antonia, Steve and I were fortunate enough to be invited to the Conference held by Shire in Rome. The setting was beautiful and the hotel glorious, but we were not there to take in the sights - there was work to be done! That evening we were all invited to a welcome dinner which was a perfect opportunity for us to meet old friends and make new ones.

The Conference started on the Friday morning with talks from various professionals including a very interesting talk from Dr David Begley on "The Blood Brain Barrier: The Next Target for CNS Therapy".

After a mid morning break we were back into the lecture hall and listening to more informative talks. The day went very smoothly and we all felt that after listening to the professionals speaking, we gained further knowledge and information on MPS Diseases and LSDs. Dr Uma Ramaswami was fantastic and her talk held everyone's attention.

Saturday morning's talks were based on Hunter Disease and again the talks were very informative - not being biased but we felt that Dr Ed Wraith's talk was extremely good!

We would all like to say a special thank you to Shire for inviting us at the MPS Society to such a fabulous conference and for all their organising of flights and accommodation which was second to none. **Neisha Hall**

International MPS Network, 24 – 25 May 2007 Serok, Poland

21 representatives from 15 MPS Societies worldwide came together in Serok, Poland to meet with professionals, share experiences and develop the MPS Network worldwide.

Dr David Begley (Kings College Hospital London) and Dr Maurizio Scarpa (University of Padova Italy), told us of their efforts to establish a European Blood Brain Barrier Group (BBBG) for Lysosomal Storage Diseases called Brains for Brain. The BBBG are initiating discussions in Brussels that we hope will result in the group successfully applying for an EU Grant in the near future. The MPS Societies in Europe are supporting this initiative which may require us to keep lobbying the EU for funding for this important area of scientific research that we hope will translate into new therapies for sufferers around the world.

Dr Grzegorz Wegrzyn (University of Gdansk), informed us of his research.

Dr Ute Vits (Country Manager Biomarin Europe Ltd) updated us on a new survey planned to start this year which aims to assess the effects of having MPS VI on the quality of life of affected individuals and their families and carers. This survey is being co-ordinated by Prof Michael Beck from Mainz, Germany but will be conducted throughout Europe including the UK. Discussion took place around ensuring all affected by MPS VI whether or not they are receiving ERT are encouraged to participate.

Considerable time was spent considering the status of the International NPS Network. It was agreed to establish some areas of governance whilst ensuring the Network is sufficiently flexible to accommodate the fluidity of the MPS Societies around the world, the majority of which are run only by volunteers. Whilst one or two MPS Societies felt the need for a formal constitution, the majority preferred a less formal Memorandum of Understanding that allows the MPS Network to prioritise and address the issues important to members of MPS Societies in Europe and worldwide, rather than being dogged with bureaucracy.

The International MPS Network already has a system of selecting the MPS Society to host the International Symposium on MPS and related diseases. As you will be aware the 2008 International MPS Symposium will be held in Vancouver. At the International MPS Network meeting we heard two proposals to host the International MPS Symposium in 2010. These proposals were received by the closing date of May 2007, and were submitted by the Spanish MPS Society to be held in Barcelona and a joint proposal from the Australian MPS Society and New Zealand LSD Group to be held in Adelaide. Following the presentations questions have been put in relation to the two proposals and a final presentation from each will be heard in the Autumn with each MPS Society having one vote. During the MPS Network meeting the Hungarian MPS Society suggested they might wish to be a candidate to host the 2010 International MPS Symposium, but in a vote it was agreed unanimously that only the proposals received by the deadline could go forward to the final decision in the Autumn.

During the International Network Meeting it was agreed to compile a comprehensive list of all the MPS Societies current literature and information resources giving languages available and cost. This list will be made available on the UK website in the autumn.

Christine Lavery, Chief Executive

Jeans for Genes Day

Friday 5 October 2007

Here is an important date for your diary - put a big ring around Friday 5 October 2007. That is Jeans for Genes Day when we want to get you and millions of people around the United Kingdom to wear their jeans and raise money to help children affected by MPS and other genetic disease.

Go on, get 'denimised' on Jeans for Genes day!

Call the freephone number **0800 980 4800**
or register for your free fundraising pack at **www.jeansforgenes.com**

Age Appropriate Alternatives To Bibs

Are you the parent or carer of a young person/adult with special needs? Can't find an age appropriate alternative to bibs?Then log onto Dribble Bandana's website (www.dribble-bandanas.co.uk) and view the fantastic range of adult bandanas which offer the perfect solution.

Dribble Bandanas produce handmade, funky bandanas that can be used instead of bibs. As youngsters with Special Needs grow up, they no longer wish to wear bibs that are traditionally associated with babies and toddlers. This alternative bib offers them the protection that they need while also acting as a fashion accessory.

Lisa, owner of Dribble Bandanas states, "Originally I made the dribble bandanas for young children. However recently I have been inundated with orders for young people and adults who have special needs. The dribble bandanas that I produce are more aesthetically pleasing than a bib and compliment an outfit rather than distracting from it."

Dribble Bandanas are only available to be ordered online. Bandanas are available in a number of designs and sizes.

www.dribble-bandanas.co.uk

Wheelchairs and Flights

A family have recently made an enquiry to the MPS Society regarding access with wheelchairs on flights. We have looked into this with numerous airlines and organisations and a Social Worker at Heathrow airport. Passengers are unable to board aircraft in their own specialist wheelchairs. The normal procedure is to transfer to the airlines wheelchair on check in, and their personal chair loaded onto the aircraft. This is due to air safety regulations.

All Virgin and BMI flights have onboard wheelchairs to assist to seats and toilets. Virgin also confirmed that they can also provide a travel chair with specialist upper body support which is free of charge. They can also help in locating a hotel that is wheelchair accessible. Telephone Virgin on 0870 990 8350, Monday - Friday 9am - 5pm or email: customercare@virginholidays.co.uk

Electric Wheelchairs
The air access carrier regulation states that spillable batteries must be entirely removed, unless the wheelchair will be stored upright for the duration of the journey and is attached to the chair. Non-fillable batteries do not have to be removed.

More information can be found at **www.direct.gov.uk**

New postage system

When sending post to the Society please ensure you use the new letter, large letter and packet system. For every incorrectly weighed letter this costs the Society £1.05 in charges plus the additional postage.

DISABLED ACCESS HOLIDAYS

All Around the World

Disabled Access Holidays in partnership with **Clydegrove Travel Agents** for the Global Travel Group, (ATOL No. 83973) provide holidays that are suitable for disabled travellers. They also book flights for customers who have sourced their own accommodation.

Clydegrove Travel are a specialist travel agency who assist when booking flights to ensure the appropriate level of assistance is available at the arrival and departure airports.

Wheelchair adapted vehicles are available for transfer to and from the airport on some holidays.

Clydegrove Travel
2352 Dumbarton Road
Yoker
Glasgow
G14 ONN

Tel: 0141 270 7577, Mon-Fri 9am-5pm

Disabled Access Holidays
9 Newton Place
Charing Cross
Glasgow
G3 7PR

Email: flights@disabledaccessholidays.com

ACCESS TRAVEL

Access Travel have apartments covering most of Europe and also Florida. All properties are well known as they have been personally inspected or have been suggested by a wheelchair user. They offer a personal contact to you. Access Travel offer special airfares on scheduled and charter flights. Information on hiring hoists, shower chairs etc is also available. Nursing and Care services are available if travelling to Algarve or Tenerife. Certain resorts have wheelchair adapted vehicles, hoist or ramps.

Access Travel (Lancs.) Ltd
6 The Hillock
Astley
Lancashire
M29 7GW

Tel: 01942 888844

E mail: webeng@access-travel.co.uk

NATIONAL CHARITY HOLIDAYS

for Families with a Disabled Child
This organisation provides information on holidays arranged by Voluntary and Commercial Organisations, accommodation and a variety of useful contacts.
www.holidaycare.org.uk

The Royal British Legion Benevolent Schemes

I wanted to let our members know about the help that is available from the Royal British Legion. I have had experience of them in the past and have found them to be very helpful in providing funding for disability equipment, grants for those in need of assistance with bills etc.

Criteria
The scheme is available to help serving and ex-serving servicemen and women and/or their dependents (up to the end of education), in need. This includes individuals that are divorced, widowed and separated. However, the individual must have served in the Forces for at least seven days.

You can either contact them direct on their general enquiry line Legionline on 08457 725 725 which is open 10-4pm Monday to Friday, via their website www.britishlegion.org.uk or we can contact them on your behalf. **Chris Murphy, Advocacy Officer**

EDUCATION MAINTENANCE ALLOWANCE

Education Maintenance Allowance (EMA) provides financial support for 16-19 year olds who undertake a full time course at school or college.

The schemes are managed by your local council. It is a weekly payment and is intended to assist with the day to day costs of staying on at school, college or training, e.g. travel, books and equipment. Generally the scheme is the same throughout the UK but with a slight difference in Scotland. Full time education means at least 12 hours a week.

To receive the weekly payments the young person must have signed an EMA contract with the educational establishment. This sets out what is expected from them in terms of attendance, coursework and progress.
Bonus payments can also be received if the younger person has achieved their learning goals.
In some circumstances EMA can be received while studying from home, for example, for medical reasons, as long as this entails completing 12 hours of guided education but the young person must be enrolled at a college or school.

The funding is based on household income

Up to £20,817 per year:	£30 per week
£20,818 - £25,521 per year:	£20 per week
£25,522 - £30,810 per year:	£10 per week

Effects on other benefits
EMA is paid on top of, and therefore does not affect other household benefits. Young people can also keep whatever extra income they may earn from a part time job.

A young person is not eligible if they are in receipt of:
An adult learning grant
Jobseekers Allowance
Minimum Training Allowance

Contact details
Application forms can be accessed from schools, colleges, learning providers, Connexions or online at http://www.direct.gov.uk/ema

Wales	http://www.emawales.gov.uk
Northern Ireland	http://www.emani.gov.uk
Scotland	http://.www.emascotland.com
EMA Helpline number	0808 101 6219

Aid for Children with Tracheostomies (ACT).
Lammas Cottage
Stathe
Bridgewater
Somerset
TA7 OJL

Tel: 01823 698 398
Contact: Amanda Saunders
Web: **www.actfortrachykids.com**

Aid for Children with Tracheostomies is a national self help group. It was founded in May 1983 and is run by parents of children with a tracheostomy and by people who sympathise with the needs of such families. It links groups of individual members throughout Great Britain and Northern Ireland.

They offer support and practical advice to parents of a child with a tracheostomy. They also offer a starter pack full of useful information including a parent's handbook and a quarterly newsletter. Membership is free to parents.

Useful website shared by a family

A family have asked us to let our members know about a company who they have found useful when purchasing specialist clothing.

Rackety's is a UK company who specialise in clothing for children with special needs. Their clothes incorporate special fastenings and modifications to aid dressing and to try and make it less stressful.

For more information phone Rackety's on 01538 381430 or visit **www.racketys.com**

Another family have also asked us to let our members know that Debenhams Gold Card members can have their clothing altered at no extra charge.

10th International Symposium on MPS and Related Diseases

26-29 June 2008

Vancouver, British Colombia, Canada

Conquering MPS: Learn Live Cure

The Canadian MPS Society invites MPS families from around the world to take part in the collaboration among families, caregivers, specialists from the scientific and medical communities, and industry, dedicated to improving the lives of those affected. In addition to providing a stimulating three day educational experience for adult delegates, our professionally designed programme CAMP CANADA, for children, youth and teens will provide our young delegates with memories to last a lifetime! See you in Vancouver! **Kirsten Harkins, Canadian MPS Society**

The UK MPS Society is raising funds to provide support to its member families to participate in this important International Symposium. In the Autumn MPS Magazine we will be providing more details but in the meantime, please let us know if you are interested in participating in the International Symposium in Vancouver. Whilst we would hope to meet some of the costs for up to two adults (parents) and two children of 20 MPS families it is most unlikely we will be able to meet all the travel, accommodation and registration costs therefore MPS members need to think about budgeting for this now if they want to apply to be one of the participating UK families to receive support from the UK MPS Society. **Christine Lavery, UK MPS Society**

Your news and views

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

Developments in Understanding the Blood Brain Barrier

As presented by Dr David Begley at the MPS Conference 29 June – 1 July 2007

The blood brain barrier is formed by the smallest blood vessels in the brain, the capillaries. The capillaries of the brain are unlike those of most other body tissues in possessing this barrier. The blood brain barrier prevents the free movement of small and large molecules from blood to brain and vice versa. Molecules that the brain needs are transported across the barrier. The barrier is a physical barrier to free diffusion, is a transport barrier in and out and is a metabolic barrier.

Why a blood brain barrier?
The central nervous system (CNS) needs to maintain an extremely stable internal fluid environment which is an absolute requirement for reliable synaptic communication between nerve cells. The blood brain barrier (BBB) is also a protective barrier which shields the central nervous system from circulating neurotoxic substances in blood which are produced by metabolism or are ingested in the diet or otherwise acquired from the environment. Most fully differentiated neurones are unable to divide and replace themselves and therefore any acceleration in the normal daily rate of attrition of neurones (cell death) will become prematurely debilitating.

According to World Health Organisation (WHO) statistics, neurological diseases including the MPS and Related Diseases constitutes one of the top five causes of disease and suffering and yet most lead drugs under development for CNS disease fail as they do not cross the BBB.

The relevance of the BBB for types of MPS where the CNS system is involved is very important when it comes to developing new therapies. Current substrate reduction therapy (SRT) and Chaperone Therapy needs to cross the BBB to be effective. Current ERT does not appear to cross the BBB in therapeutically significant amounts to protect or repair the BBB.

Why do we need to know more about the BBB?
Damage to the BBB may contribute to the neuropathology of MPS and Related Diseases. At present we do not know what factors determine the entry of SRT and chaperones into the brain. In order to design new small molecule therapies we need to understand the process of brain penetration for them to be effective. The BBB contains a number of transport processes which take large molecules such as proteins into the brain. We need to understand these transporters for macromolecules so that we can adapt ERT to cross the BBB and treat the brain

Current research at Kings College, London into Sanfilippo Disease supported by the UK MPS Society and the Shauna Gosling Trust:

Two mouse models of MPS IIIA and IIIB

The development of a new method for assessing very accurately BBB permeability in situ in the mouse

Investigating changes in the BBB function in the disease models and the transport of metabolites and drugs into the brain and the contribution of the BBB pathology to the disease process

Changes in brain blood volume and blood flow in the disease models have been identified

Also changes in kinetics of glutamic acid transport across the BBB is identified and determined

The MPS Society has granted a further £205,581 over two years to take this important work forward with the appointment of two additional research workers.

Brains for Brain (B4B)

Because Lysosomal Storage Diseases (LSDs) are rare diseases the resources in individual countries in terms of scientific manpower and finance for research is limited. By forming a consortia, B4B, extra resources can generate both scientific manpower and the funding for research.

Brains for Brain is a European Consortia of scientists supported by the European network of MPS Societies and the pharma industry. Discussions have already started with Dr David Begley and Professor Maurizio Scarpa meeting with bureaucrats at the European Commission in Brussels to stimulate interest in funding research on LSDs affecting the central nervous system.

Lobbying is a vital element of this process. Individual MPS families are encouraged to write to their National Representatives telling them how their family is affected and why the EU should introduce further calls to fund vital research for rare diseases involving the nervous system. A new call under Brains and Brain – Related Diseases should specifically include the Blood Brain Barrier and drug delivery to the central nervous system.

UK National Representatives
Dr Mark Palmer, mark.palmer@headoffice.mrc.ac.uk
Dr Jill Jones, jill.jones@headoffice.mrc.ac.uk

EU Commissioners and MEPs

External Trade (UK)	Peter Mandleson
Head of Cabinet	Simon Fraser
Science and Research	Janet Potocnik
Head of Private Office	Peter Droell
	peter.droell@ec.europa.eu

Gene therapy for MPS II

by Joseph Muenzer, MD, PhD

Hunter syndrome is caused by the deficiency of the lysosomal enzyme iduronate sulfatase, which results in the accumulations of glycosaminoglycans (GAG). In individuals with MPS II, the progressive storage of GAG causes tissue and organ damage, and in the severe form premature death due to brain, heart and airway deterioration. My research laboratory at the University of North Carolina (UNC) has focused on developing adeno-associated virus (AAV) gene therapy as a means to replace the missing enzyme in MPS II utlising the MPS II mouse developed at UNC. The goal of MPS II gene therapy is to efficiently transfer a normal copy of the human iduronate sulfatase gene into a sufficient number of cells resulting in the production of enzyme to correct the GAG storage. The success of gene therapy can be measured by many criteria, such as improvement in function and life span, the reduction of GAG storage and how much iduronate sulfatase is produced.

We have previously created an AAV2 vector containing the human MPS II gene (cDNA) that is capable of making iduronate sulfatase in cultured MPS II fibroblasts and the enzyme that is made can correct the GAG storage. In MPS II animal studies, intravenous administration of the AAV2-CMV-IdS vector results in complete correction of the lysosomal storage in liver and only partially correction in other organs. When the AAV2 vector is injected both intravenously and into the spinal fluid around the brain of the MPS II mice, we have demonstrated decreased storage of GAG in the brain, improved function as measured by rotarod test and an increased life span.

Recent AAV gene therapy research has suggested that other forms (serotypes) of AAV may enter cells and are able to make the desired protein/enzyme more efficiently. In the last year, we have made another AAV serotype vector (AAV1) and have injected animals to evaluate the ability of the AAV1 vector to make iduronate sulfatase and correct GAG storage compared to the traditional AAV2 vector previously studied. In addition, the UNC vector core facility is currently making two other AAV serotype vectors (AAV8 and AAV9). Once these vectors are made, we will inject MPS II animals to determine if these vectors are more effective than the traditional AAV2 vector in making iduronate sulfatase and in correcting GAG storage.

We previously had made a new type of AAV vector (self-complementary, scAAV) which was predicted to be more efficient in its ability to make iduronate sulfatase. In the last year, we have studied eight animals that were injected intravenousy and also into the fluid around the brain with the new scAAV2 vector containing the human MPS II gene (cDNA). Our analysis demonstrated complete correction of GAG storage in multiple tissues, including liver, spleen, kidney, heart, lung, intestine and muscle after administration of scAAV2 vector. These results are a significant improvement in the reduction of GAG storage compared to the traditional AAV2 vector. The iduronate sulfatase in liver after injection of the scAAV2 vector was on average 800 percent higher than levels in unaffected mice and the kidney levels of iduronate sulfatase were 10 to 100 percent of normal levels, a previously difficult-to-treat tissue. In contrast, significant amounts of iduronate sulfatase can be detected only in the liver after injection with the traditional AAV2 vector in the MPS II mice. Histological analysis of the tissues after scAAV2 gene therapy and measurement of iduronate sulfatase in other tissues are in progress. In initial studies in two MPS II animals injected with a newly created scAAV9 vector, we have found very elevated iduronate sulfatase in liver (> 50 fold above normal levels).

These preliminary studies suggest that the new scAAV vectors are capable of significantly higher enzyme expression which results in an improved correction of GAG storage in the MPS II mice. In the next year, animals already injected with scAAV vectors will be studied and more animals will be injected to determine if our exciting preliminary findings with scAAV vectors are confirmed. In summary, AAV gene therapy using newer vectors is a very promising approach for treating both somatic and central nervous system diseases in individuals with MPS II.

Dr Muenzer was awarded a two-year grant in February 2006, $60,000 each of the two years, for his work, “AAV Gene Therapy for MPS II”. This is a summary of the first year of his research. This article appears courtesy of the National MPS Society, USA, www.mpssociety.org

Drug Information, Transparency and Access Task Force

On 16th February 2007 the Board of Eurodis created a Drug Information, Transparency Access Task Force. The aim of this Task Force is to adequately deal with the activities of the Patients' and Consumers Working Party at the European Medicine Evaluation Agency (EMEA). There are 12 representatives from 8 European Countries. It is a great privilege that I have been appointed to the DITA - Task Force and I hope the MPS Society's experience with five Orphan Drugs so far will be helpful.

Overall the Drug Information, Transparency and Access Task Force is established to provide recommendations to its representative at the Patients and Consumers Working Party on all matters of direct or indirect interest to patients in relations to medicinal products and to perform the tasks described below.

Training on the respective roles of national and European regulatory agencies and propose actions to train other patient representatives;

Learn the marketing authorisation procedures, their timelines and review the implementation of guidance documents;

Make proposals to improve the provision of medicinal product related information adapted to patients and consumer needs;

Review existing communication tools and evaluate possibilities for new ones;

Contribute to increase awareness of patients in relation to the use of medicines and the rationale for their use;

Contribute to the development and training of a network of patients and consumers' organisations;

Provide general advice in relation to product specific matters;

Identify ways to communicate information related to risk and safety;

Propose ways to improve the transparency of the provision of information released by the EMEA, national competent authorities and the other bodies;

Participate in the collection of information on the access to medicinal products including compassionate use prior to the marketing authorisation.

On 3rd May 2007 the twelve representatives met in Paris immediately prior to the Eurodis Meeting to start working on the tasks described. Key areas already underway are Drug Product information, risk communication, compassionate use of drugs in the EU and reimbursement by member states.

In the future this task force may be asked to contribute to the implementation of new Pharmaceutical Community Legislation as well as high level initiatives such as Pharmaceutical Forum and EMEA Road Map to 2010.

Christine Lavery
Chief Executive

Pharmacological Chaperones:

A Potential Therapy for Fabry Disease

Fabry disease is a lysosomal storage disorder caused by inherited genetic mutations in the GLA gene, which result in deficient activity of the enzyme galactosidase A (GLA). Deficient GLA activity leads to the accumulation of the enzyme's natural substrate, globotriaosylceramide (GL-3) in a part of the cell called the lysosome. The accumulation of GL-3 in the lysosome has historically been thought to cause the various symptoms associated with Fabry disease, including pain, kidney failure, and increased risk of heart attack and stroke. Fabry disease is thought to be significantly under diagnosed, especially in females and in individuals with late onset disease.

Recent evidence suggests that Fabry disease is part of a growing list of conditions that may be classified as disorders of protein misfolding. Each protein, including enzymes such as GLA, is made of a specific sequence of amino acids that assume a 3-dimensional shape by being twisted, bent, and folded. GLA is folded in a part of the cell called the endoplasmic reticulum (ER). Once stably folded, GLA moves out of the ER to the lysosome where it is needed to break down GL-3. Many mutations, or changes, in the GLA gene lead to production of misfolded enzymes, meaning that the enzyme does not assume a stable 3-dimensional shape when being made in the ER. Misfolded GLA is unstable and may not be able to exit the ER. Consequently, GLA may build up and aggregate, or clump together, in the ER. Potential cell damage caused by accumulated GLA in the ER may be added to the damaging effects created by accumulation of GL-3 in the lysosome.

Amicus Therapeutics, Inc. is developing orally administered compounds called pharmacological chaperones for the potential treatment of lysosomal storage disorders, including Fabry disease. These compounds are designed to selectively bind to and stabilise enzymes. Stabilised enzymes are expected to be able to exit the ER and be sent to the lysosomes where they are needed to break down their substrates. Recent studies were conducted by Amicus to characterise how a pharmacological chaperone, designated as AT1001, affects GLA and GL-3 in both humans and animals. The results of these studies are summarised below:

Administration of AT1001 to healthy human volunteers increases GLA levels in white blood cells.

Administration of AT1001 to healthy mice increases GLA levels in the liver, heart, kidney, and spleen of the mice.

Administration of AT1001 to transgenic mice, which have a Fabry gene mutation, increases GLA levels in the liver, heart, kidney, spleen, and skin of the mice.

Administration of AT1001 to transgenic Fabry mice significantly decreases levels of GL-3 in the skin and heart of the mice and shows a trend towards reduction of GL-3 in the kidney of the mice.

Additional studies are being conducted to determine the effect of AT1001 in individuals with Fabry disease.

If you have questions, please contact: patientadvocacy@amicustherapeutics.com.

References:

Yam GH, Bosshard N, Zuber C, Steinmann B, Roth J. Pharmacological chaperone corrects lysosomal storage in Fabry disease caused by trafficking-incompetent variants. Am J Physiol Cell Physiol 290:C1076-C1082, 2006.
Romisch K. A cure for traffic jams: Small molecule chaperones in the endoplasmic reticulum. Traffic 2004; 5:815-820.

Celebrate the fabric of the nation on Jeans for Genes Day

Jeans for Genes Day
Friday 5th October

Friday 5th October is all about putting bums in jeans and making a donation, so make sure you register for a Jeans for Genes fundraising pack before the big day.

Your pack will contain a fundraising guide, celebrity posters, a collection box, stickers and loads of ideas for you to help us raise £3 million on Jeans for Genes Day!

All donations will help fund pioneering research and vital support for children affected by genetic disorders.

For your FREE fundraising pack visit:

www.jeansforgenes.com

or call:

0800 980 4800

JEANS FOR GENES®

Helping children with genetic disorders

Seven national charities working together to help children affected by genetic disorders
The net proceeds from the 2007 Jeans for Genes Campaign will be distributed among the seven charities. Jeans for Genes Campaign. Reg. Charity No. 1062206. Logo and 'Jeans for Genes' ®
The Chronic Granulomatous Disorder Research Trust (CGDRT). Reg. Charity No.1003425.