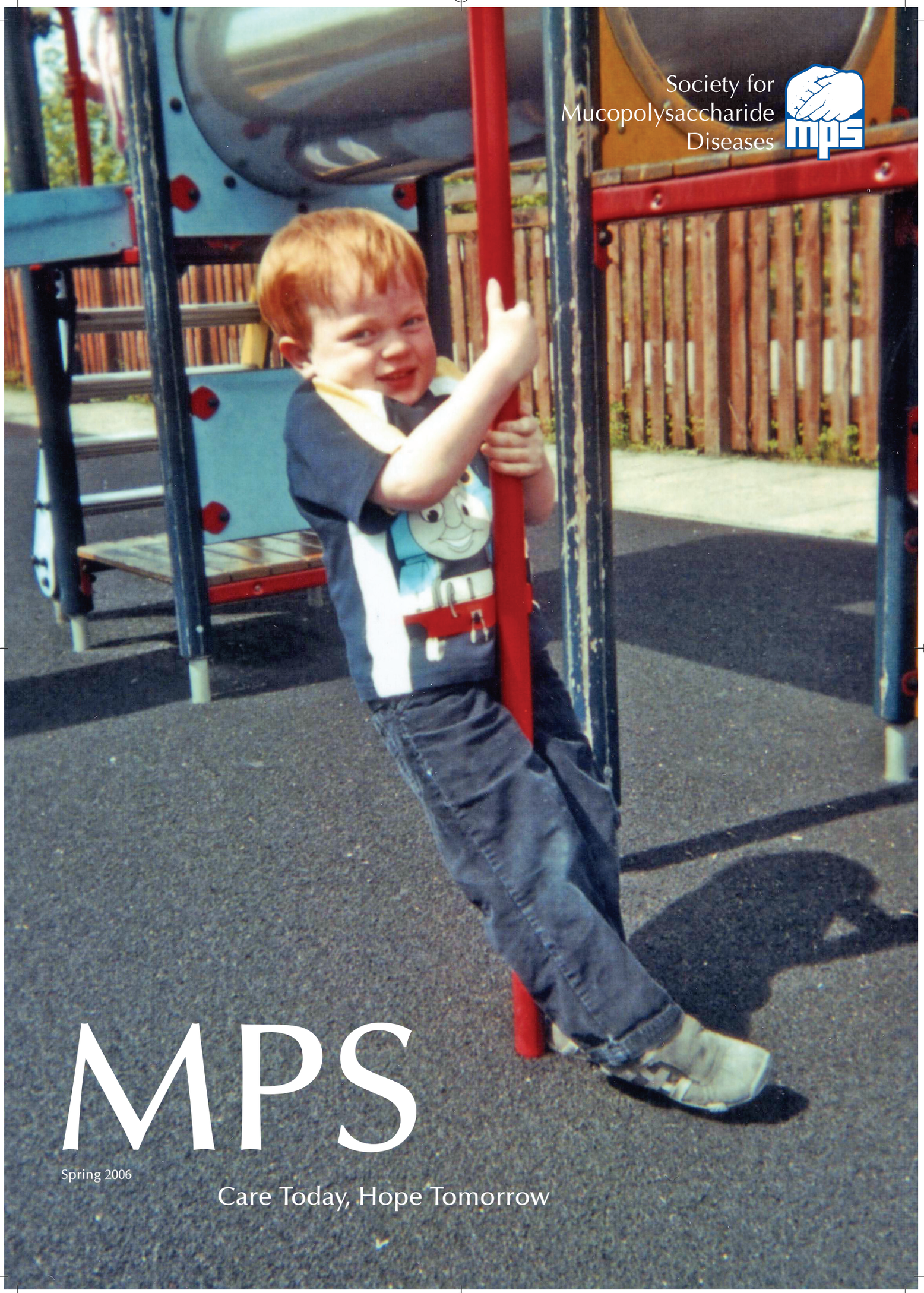

Society for Mucopolysaccharide Diseases

MPS

Spring 2006

Care Today, Hope Tomorrow

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

What is the Society for Mucopolysaccharide Diseases?

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

What are the aims of the MPS Society?

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

To bring about more public awareness of MPS and Related Diseases

To promote and support research into MPS and Related Diseases

How does the Society achieve these aims?

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

Telephone Helpline
Includes out of hours listening service

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

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

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

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

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

Information Resources
Publishes specialist disease booklets and other resources

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

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

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

Cover photograph:
Ben Conlin (Read more about Ben's story on page 14)

CARE TODAY HOPE, TOMORROW

MPS Magazine

MPS Society

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Registered Charity No. 287034

Newsletter Deadlines

Summer	1 Jun 2006
Autumn	1 Sep 2006
Winter	1 Dec 2006
Spring	1 Mar 2007

Become a Friend of MPS

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CHIEF EXECUTIVE'S REPORT

In January the MPS Society finally moved to its new National Information Centre For Mucopolysaccharide and Related Diseases. As well as affording the staff team a significantly improved environment in which to deliver all the MPS Society's services to its members and supporters the building offers a ground floor accessible conference and reception area and library. Already we have made good use of the facilities with visits from members, holding meetings with pharmaceutical companies, training events for the Society's childcare volunteers and hosting the Jeans for Genes Guest Charity Interviews. On 12 April the Society facilitated the Expert Meeting on MPS VI, Maroteaux Lamy Disease, and this was followed by an official opening of MPS House and a celebration of the launch of Naglazyme, and Enzyme Replacement therapy for MPS VI. It has to be said that without the significant financial support of the companies and charitable trusts who supported us to make this move MPS House and the development of a National Centre would not have been possible (MPS House features on page 8).

Rather like the weather, fundraising income is difficult to predict. During the Society's financial year ending 31 October 2005 MPS members and the Society's supporters raised a fantastic amount, 42% up on the previous year. Your efforts are really welcomed and appreciated enabling the Society to continue to build on the support offered to those suffering from MPS, their families and carers. The Trustees are also very conscious of the importance that our members attach to funding research that may lead to a treatment or therapy let alone cure for MPS children and adults now and in the future. With this in mind the Society has continued to strengthen its involvement as a Jeans for Genes partner charity and on 27 March 2006 the four partner charities, signed a long term five year agreement to raise over £20 million for genetic research and support projects. The MPS Society receives 22% of the net income so it is vitally important that as well as continuing your support to the Society you ask family and friends to support Jeans for Genes Day on 6 October 2006.

Finally, a taster of what the Society is planning for MPS families this year. In April we look forward to welcoming nearly 40 families to the Society's AGM and Alton Towers Weekend to be followed in May by MPS Conferences in Northern Ireland and Scotland. Regionally we are planning a South West Members Get-together in Wiltshire on 21 May with events in other areas being planned for the Autumn. The Bereaved Family Remembrance Day at the Childhood Wood will take place on 16 June closely followed by the International MPS Symposium in Venice to which a small number of UK families are attending. The Society has been fortunate to be given two grants, one to fund a Sibling Activity weekend for siblings of MPS brothers and sisters and the other to fund a series of one-day events for adult MPS sufferers. If you are an adult MPS / Fabry sufferer please do tell us how you could best benefit from such an activity and where you would like it to take place. If you are a parent of an MPS sibling(s) or an adult MPS sufferers(s) please encourage them to tell us how they too would like to benefit and where they feel the activity should be located.

Christine Lavery
Chief Executive

News from the MANAGEMENT COMMITTEE

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Management Committee Meeting held 3 - 4 February 2006

Personnel

The Chief Executive advised Trustees of personnel matters and the Trustees sent their congratulations to Clare Cogan, Senior Advocacy Officer, and her husband, Will, on the birth of their baby, Joseph. During the meeting the Trustees undertook the annual staff salary review in the absence of the Chief Executive and HR & Information Officer.

MPS Office

Trustees were informed of progress to date on MPS House following the Society's move from Woodside Road. The Society had been fortunate in achieving substantial grants to meet the cost of transforming the building into a National Resource and Information Centre for Mucopolysaccharide and Related Lysosomal Diseases.

Governance

Prior to the meeting, the Chief Executive had received feedback from Trustees on the draft Strategic Plan for MPS. Amendments were agreed. By law, and for the year end accounts, MPS is required to include a statement regarding mitigating risk. As circumstances have now changed due to the move to MPS House the Trustees agreed to review the MPS Risk Register at the next Management Committee Meeting.

Alton Towers Weekend & Annual General Meeting

Trustees were updated on the plans for the Alton Towers Weekend including numbers of MPS member families attending and arrangements for volunteers.

Policies

The Trustees reviewed and agreed with amendments the Health and Safety Policy and the Internet and email policy. The following policies were reviewed and agreed without amendment: HIV / AIDS Policy; Handling Poor Performance Policy; UK Travel and Subsistence Policy; Use of animals in Research Policy; Financial Assistance Scheme; Policy on use of Electronic Equipment; CRB and Disclosure Privacy Policy; Copyright Policy; Managing Stress in the Workplace; Gift Policy; Sickness Absence Policy; Organising Regional Events; DSE Eyecare; Leave for Domestic and Personal Reasons; No Smoking Policy; Policy Statement on Board Diversity

It was also agreed that a new Paternity Policy would be drafted for agreement by Trustees and that the Reserves Policy be considered further at the next Management Committee Meeting.

MPS Research Grants

A summary of all MPS Research Grants supported with Jeans for Genes funds and other charitable donations was distributed. Trustees agreed to increase the grant to the Senior Research Fellow at the University of Manchester to reflect additional costs. A second year report from Dr Clare Beesley was tabled along with the budget for her third year of the project on Biomarkers. Trustees agreed the costings and confirmed Clare's final year of funding for this project.

Don't forget to make a note of the Society's new address and contact details!

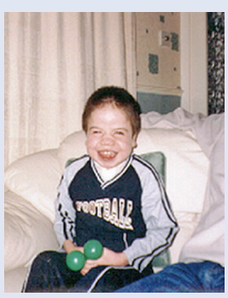
Read more about MPS House on page 8.

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

Tel: 0845 389 9901, Fax: 0845 389 9902

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

Announcements



Michael Thompson

As some of you know, Michael passed away peacefully on 23 December 2005.

We just wanted to take a moment to thank all our MPS friends for the love and support they have shown to us since diagnosis all those years ago. We have many happy memories of MPS family gatherings and they will stay with us always.

Ann, Ron and Gemma

New Members

Mr and Mrs Green have recently been in contact with the Society. Allan has a diagnosis of Fabry Disease. Allan is 56 years old and the family live in the North of Wales.

Mr Mohammadbeigi has recently been in contact with the Society. Amirreza has a diagnosis of Hunter Disease and is 15 years old. The family live in the North East.

John Purdham has recently been in contact with the Society. John has a diagnosis of Morquio Disease. John lives with his family in Scotland.

Mr and Mrs Robinson have recently been in contact with the Society. Oliver has a diagnosis of Sanfilippo Disease and is 7 years old. The family live in the South West.

Deaths

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

Michael Thompson who suffered from Hunter Disease and who died on 23 December 2005 aged 18.

Jack Taylor who suffered from Hunter Disease and who died on 24 December 2005 aged 8.

Siobhan Lowther who suffered from Sanfilippo Disease, passed away peacefully at home on 27 December 2005. This was also the anniversary of her brother Shaun's death. Shaun also suffered from Sanfilippo Disease and died in 2001.

Jake Shaw who suffered from Hurler Disease and who died on 9 January 2006 aged 8.

Ryan Kelsall who suffered from Sanfilippo Disease and who died on 7 February 2006 aged 9.

Matthew McLaren-Hall who suffered from Hunter Disease and who died on 3 March 2006 aged 12.

Carly Barker who suffered from Sanfilippo Disease and who died on 14 March 2006 aged 19.

Farewell from Nikki McAuliffe

Dear all, just a little note to say goodbye. I know I've only been with the Society for a short while, but the time has come for me to say farewell to you all as I look to further my career in psychology and hope to pursue a PhD. Since I started back in July last year, I've met, and advocated on behalf, of some really wonderful families and some fantastic children. I would just like to say it's been a pleasure working with you and some of my highlights include meeting some of the Scottish families at Almond Valley, attending some of the regional clinics and mingling with families at the International Fabry conference in Paris, not to mention some of the home visits I've made! Cheerio and all the best for the future.

Introducing... Tim Summerton

I am the most recently joined member of the board of trustees having become a co-opted Trustee in September. I viewed the invitation to become a trustee as a huge honour and an opportunity to give back something to the Society. One of the little surprises that comes with the job is having to write an article all about me!

So let me begin, I'll try and keep it brief. I have been involved with the Society for five years since my daughter Sophie was diagnosed with Sanfilippo disease. During this time the MPS Society has provided immense support and guidance.

I am married to Sally and we have two children, William and Sophie. We live in Hertfordshire, conveniently close to the MPS office.

I work for a large international organisation in the Finance sector. My job as a software development manager is extremely challenging, at times exciting and always pressured.

After graduating I started my career as a nutritionist in Lincolnshire. I met my future wife and moved to London where I developed my computer and finance skills designing and writing software for the finance department of a large shipping organisation. I then spent a number of years as an IT consultant, working on large and small projects gaining much insight into various aspects of human nature and the diversity of business needs.

Tim and Sophie (MPS III)

With a job and a daughter with Sanfilippo I don't have much spare time. I have been tempted in the past to list my leisure activities as attending hospital appointments, home adaptations and providing specialist child care! However, whilst these and many more are true I think it would be more accurate to say that these are not my true interests! I am a keen gardener and spend much time in the garden. I also enjoy DIY, reading and cycling not to mention spending too much time on the computer!

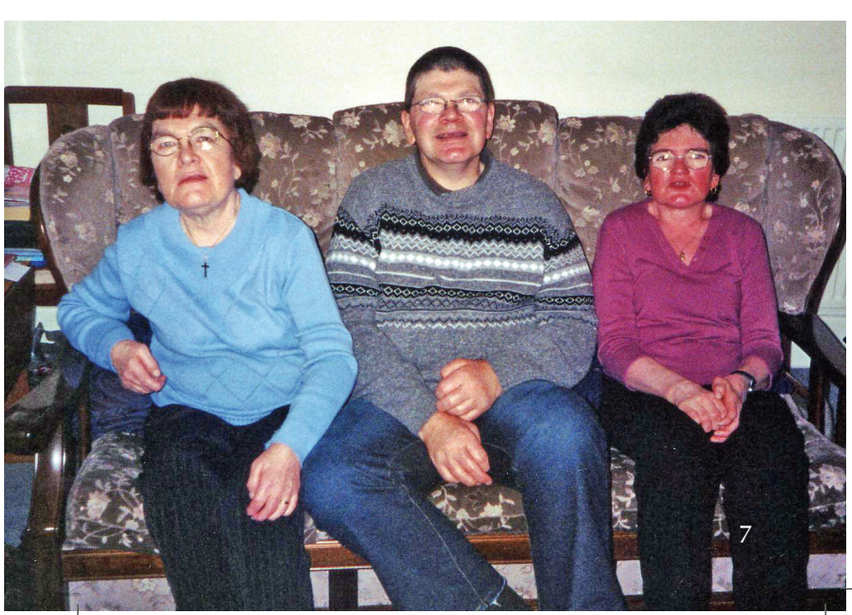
Christine Lavery appointed to Advisory Board

Christine Lavery will act as the Patient Representative for the next two years to the Advisory Council of the Department of Health's National Specialist Commissioning Advisory Group (NSCAG) for Lysosomal Storage Diseases.

Ann Canton (MPS IS) wrote to tell us about her 60th birthday surprise

For my 60th birthday it was planned I would spend some time with my sister, who lives close to Antony Selwood and Janice Wilkes, both fellow MPS sufferers, so we met up to spend the afternoon together.

As a surprise my sister bought me tickets for the Millennium Centre (I'd always said I wanted to go there). We had a meal in an Italian restaurant and then saw the show Saturday Night Fever, which was a little loud, but very enjoyable.



MPS National Centre

for Mucopolysaccharide and Related Diseases

In 2001 the Society for Mucopolysaccharide Diseases launched a capital fund to be used to create a centre as a national source of authoritative advice and information, a place where learning and teaching about MPS and Related Lysosomal Storage Diseases can be shared, and a focal point for the UK-wide MPS Society.

Society for Mucopolysaccharide Diseases
MPS House, Repton Place, White Lion Road,
Amersham, Buckinghamshire, HP7 9LP, UK
Tel: 0845 389 9901, Fax: 0845 389 9902
www.mpssociety.co.uk,
Email: mps@mpssociety.co.uk

After much searching, the MPS Society has found just the right building. The first floor provides ample space for the Society to provide all its advocacy and support services whilst the ground floor will provide a modest conference area and training resource plus library area.

We want the MPS centre to be vibrant, motivating and exciting, welcoming and involving, a 'heart and home' for the MPS community whether that is affected children and adults and their families and carers or any of the many professionals in healthcare, education, social services and the pharmaceutical industry that work with us.

MPS House is ideally situated for motorway access and public transport to all parts of the United Kingdom as well as to London, Heathrow and Luton airports.

The MPS centre will be fully accessible to visitors and we offer a warm welcome to everyone either contacting MPS remotely or visiting the MPS National Centre.

The Society would like to thank the following donors for their contribution to the development of a dedicated resource for MPS and Related Diseases: Jeans for Genes, BioMarin Europe Ltd., Shire Human Genetic Therapies UK Ltd., Actelion Pharmaceuticals UK Ltd., The Foyle Foundation, John Lewis Partnership High Wycombe.

Family Noticeboard

We have received a suggestion from one of our families for a 'Noticeboard' type column to appear in the MPS Magazine. This would ideally provide any hints and tips for other family members, ranging from suggestions on types of equipment used to support a sufferer to strategies for coping with varying stages of a particular disease. Often, something that seems logical or trivial to one family may not occur to someone else, but could go a long way to making another family's life so much easier.

If anyone has any ideas they would like to share with other members please could you either call the MPS office on **0845 389 9901** or send us an email to **mps@mpssociety.co.uk**

We are sure that you all have a mine of information stored away which could be of use to other families. So, thinking caps on please!

Dates for your diaries!

4 May	Northern Ireland MPS Clinic, Antrim Area Hospital Social Gathering, Hilton Templepatrick Hotel, Belfast
5 May	Northern Ireland MPS Conference, Hilton Templepatrick Hotel, Belfast
21 May	South West Family Gathering, Westbury, Wiltshire
30 May	Bristol MPS Clinic
15 June	Social Gathering, Hilton Edinburgh Airport Hotel
16 June	Scottish MPS Conference, Hilton Edinburgh Airport Hotel
22/23 June	MPS III Clinic, Great Ormond Street Hospital
29 June - 2 July	9th International Symposium, Venice
9 July	MPS Family Day, London Zoo
26 July	BMT Clinic, Royal Manchester Children's Hospital
28 July	BMT Clinic, Royal Manchester Children's Hospital
29 August	Bristol MPS Clinic, Frenchay Hospital
11 November	Cardiff MPS Clinic
16 November	Northern Ireland Clinic, Antrim Area Hospital, Belfast
30 November	Bristol MPS Clinic, Frenchay Hospital

Cardiff MPS Clinic

Cardiff City centre. Rush hour. Nightmare! After a hideous journey from the hotel to the hospital, which should have theoretically only taken 10 minutes, we finally arrived at the children's centre for the Cardiff MPS clinic.

This was another successful clinic and it was lovely to meet all the families who attended. The MPS Society would like to thank Dr Wraith and Dr Shortland for all their hard work and also to Christine Caveney and Sue Crownshaw for their part in the clinic's organisation and running.

Newcastle MPS Clinic and Get Together

7 - 8 February 2006 by Neisha Hall

We would like to start by saying a big thank you to Dr Wraith, Dr Rylance and Dr Leech for their ongoing support and time, and to the staff at the Royal Victoria Infirmary Hospital for making us so welcome and keeping us well supplied with tea, coffee and biscuits.

The clinic ran to time over the day and a half, and I hope all who attended benefited from their time with the doctors. It was lovely to meet you all and put names to faces and to see some familiar ones too.

Tuesday evening was our time for some socialising, and where better than in the bar of the Washington Holiday Inn Hotel. This was a time for families to get together and catch up, with an opportunity to speak to Sophie and myself about any issues in a more relaxed atmosphere. We both hope you all enjoyed the evening as much as we did - even though the football was on and held most of the men's attention, including Dr Wraith, who was kind enough to attend the evening.

Photos page 8 clockwise from top right: Stephen Jones (MPS III), Helen Skidmore (MPS IH), Megan Rennoldson (Mannosidosis), Abigail Harvey (ML II), Joseph Coleman (AGU), Christopher Jones (MPS III). Photos this page clockwise from top right: Callum McDonagh (ML III), David Jones (MPS IV), Luke Chapman (MPS III), Colin Arrowsmith (MPS II), Group at Newcastle Get-Together

MPS III Clinic

Great Ormond Street Hospital

On 26 January 2006 Great Ormond Street Hospital (GOSH) held its first MPS III clinic. As it was the first clinic only three patients were invited to attend. The purpose of the clinic is to provide a joint clinic appointment for patients to see the specialist consultants in MPS diseases, Dr Ashok Vellodi and Dr Maureen Cleary, the clinical nurse specialist Niamh Finnegan along with the specialist speech and language therapist Martina Ryan.

The clinic went extremely well and there are plans to hold another clinic in June 2006. All appointments for the clinic are arranged through GOSH with attendance from the MPS advocacy support team, who will be there to provide support and information as needed. Feedback from the first clinic was that it went extremely well but that the number of patients seen could be increased. This is something that has already been agreed and planned for.

We would like to extend my thanks to Dr Vellodi, Dr Cleary, Niamh Finnegan, and Martina Ryan for a very successful clinic and for making the Society feel so welcome.

MPS Family Day, London Zoo

Invites are being sent out to family members within the M25 boundaries and home counties including Buckinghamshire, Berkshire, Hertfordshire, Oxfordshire, Surrey, Kent, Essex, and Middlesex to attend a fun day at London Zoo on Sunday 9 July 2006. If you live outside of these areas and would like to join us, please contact Sophie Denham. We would love to see as many of you as possible and can guarantee a fun-filled day.

Childhood Wood Remembrance Day

The MPS Society would like to invite as many bereaved families to attend the MPS Remembrance Day. This is to be held on the Sunday 16 July 2006 at the Childhood Wood, Sherwood Pines. Prior to meeting at the Wood, we are inviting families to join us for lunch at Rose Cottage, approx. five miles away from the Wood. Rose Cottage is in a lovely part of Nottingham. It has its own children's outdoor play area and is opposite Rufford Abbey. After lunch we will make our way to the Wood to remember those individuals who have lost their lives to an MPS or related disease. This will include a balloon release. The day will conclude with light refreshments and the opportunity to explore the wood. We hope that as many of you as possible can join us.

Grant for Volunteers

Youngsters suffering from potentially life-threatening diseases could benefit from a new training course in Bucks – thanks to a National Lottery grant.

The Society for Mucopolysaccharide Diseases, based in Amersham, has been awarded £3,500 under the Awards for All programme which sets aside lottery money for community projects.

The cash will help the Society teach up to 35 volunteers to care for children and young people suffering from the diseases who attend activities and events at the centre.

Depending on the success of the trial the course could be rolled out to other MPS centres across the country.

Children born with MPS diseases are not able to produce the enzymes which help to break down used material in the body. This means that the body cannot dispose of the waste material and it remains stored in the cells of the body.

The diseases are progressive, as the amount of used material builds up over the years, and can particularly affect the brain and bones. Although research, especially into gene therapy, continues, there is currently no known cure for the condition.

Article appears courtesy of Bucks Examiner, January 2006

If you would like support from the Society's advocacy team please do contact us.
Phone 0845 389 9901
or email
advocacy@mpssociety.co.uk

You are important to us, please keep in touch.

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

Rotherham Advocacy Partnerships

Rotherham Advocacy Partnerships (RAP) is a voluntary organisation that provides advocacy for adults with a learning disability who live in the Borough of Rotherham, South Yorkshire. RAP has been in existence for 12 years and helps a range of people from those who have very complex needs to others who are very able but may struggle from time to time with what life throws at them. We are a small team, having three full-time and one part-time 'hands on' advocates to cover a well populated geographic area.

I had been working for less than two weeks with the family of 'S', a young lady aged 19 who sadly was in the latter stages of MPS III, Sanfilippo Disease. 'S' had lived with her gran for the previous 12 years of her life, attending a local special needs school and enjoyed visitors to the home from her large family. She was much loved and treasured and obviously considered very much part of the family.

At the time I became involved 'S' was no longer living with her gran and had been found a place by Rotherham Social Services in a small nursing home in Sheffield. There were many advocacy issues to be addressed for this young lady. Help and support was needed for 'S' and also for her gran and immediate family members.

Prior to meeting representatives from the MPS Society I knew very little about MPS Diseases, their effect and of the various stages and care that was needed. I had contacted a nurse who had previously had some involvement with 'S' but although she gave me an idea of Sanfilippo Disease, she admitted that she was not an authority on the disease as it was a specialism that she had not encountered previously.

With the Society becoming involved with 'S' virtually at the same time as myself, my role as advocate became much more effective. It was obvious that they both had a great deal of knowledge about MPS diseases and perhaps, more importantly, knew what was needed for 'S' to make her as comfortable as possible for the remainder of her young life.

Before meeting with the Society I had grave misgivings as to whether 'S's present home was meeting her needs and had voiced these to services. Prior to the involvement of the MPS Society, neither I, not it appears local professionals, had been aware that she had reached the stage of needing palliative care.

I do not intend to detail events, which were many, due to confidentiality to 'S' and her family. I do feel though that a very positive outcome was achieved for 'S' by joint working between RAP and MPS. The Society's involvement and expertise enabled me, as advocate, to have a great deal more influence when working with local professionals to achieve what was needed for 'S' in a much shorter time scale than was the norm. Indeed, without their knowledge of the disease and of how the time factor is of prime importance, the situation might still be in its early stages of being resolved, to the extreme detriment of 'S'.

From a personal point of view it was good to have the Society on board. They were very approachable and gave me lots of support as well as advice, whilst we kept each other informed of events and 'planned the strategy'.

I would welcome working with the Society again if the situation arose and it has made me realise the benefits of working with other specialist organisations if future referrals for advocacy support warrant it.

Jan Reed, Advocate
Rotherham Advocacy Partnerships

Do you have a story or an experience to share with us?
Send an email to newsletter@mpssociety.co.uk
or phone
0845 389 9901

Ben's Story by Peter Conlin

Our son Joshua Benjamin Conlin (Ben) was diagnosed with MPS I Hurler-Scheie Disease on 31 October 2003. This is our story.

Ben has, from almost the moment he was conceived caused problems. My wife and I had experienced several miscarriages. I'm not sure if this is related to Ben's condition or not, so when my wife became pregnant with Ben she was scanned at 7 weeks (very early). The scan showed twins and for just ten minutes Ben was a twin. We had mixed emotions of excitement and mild anxiety at the prospect of twins. Sadly, however we had suffered another miscarriage and the other baby was smaller than Ben and from its size it appeared to have died the previous week. Only one healthy heart beat was found.

We were advised that the other baby would simply be absorbed into the amniotic fluid over time. By the next scan the other baby had gone and only Ben remained.

The rest of the pregnancy flew and on 16 May 2001 I was working in York. We rented a place in Fridaythorpe in Yorkshire where I stayed Sunday to Thursday. On 16 May I was driving from York to Tadcaster when I noticed a familiar Claret Ford Escort in a lay-by. It was Tracey and my daughter Sarah… totally and utterly lost, going the wrong way around the outskirts of York. Luckily I was in my car and by pure fluke passing that lay-by. I stopped, laughed, and pointed them in the right direction.

I got home later to see Sarah and Tracey waiting and my dinner prepared - fantastic. Tracey had finished work for her maternity leave and had decided to come and visit me. That night when we settled down to watch TV, quite unexpectedly, Tracey went into labour.

Ben had decided to be awkward again. Tracey phoned two local hospitals for advice when suddenly we realised we had no one to look after Sarah. After phoning my parents we made the 90 mile journey to Stockton, dumped Sarah and headed for North Tees, Ben was supposed to be born in Durham, this is just typical Ben.

He was eventually born on 17 May 2001 in North Tees General Hospital. When he was first born I thought his only difficulty in life would be the fact that he's a ginger, but we could live with that.

Unfortunately, Ben had been delivered too quickly and in virtually one push. This had possibly caused him to have amniotic fluid on his lungs and as a result he went blue on a number of occasions, he was taken to intensive care and monitored. Tracey and her new son ended up staying five days in hospital, not the expected two.

When he was born he was diagnosed with a hernia and a hydroseal. Once the initial fright was over and Ben and Tracey left hospital, Ben appeared fine and our main concern was his hydroseal and we regularly asked for advice.

Ben met all his developmental stages, sitting up, crawling, walking, and speaking. As he developed we noticed subtle physical characteristics. His tummy was large and we used to joke that he walked like a rugby player. We were amazed about the size of his first cycle helmet, when he needed to wear an adult helmet.

When we returned to Stockton, we registered with a GP. When my wife asked about Ben's hernia and stomach this GP told her that Ben was overweight and we should change him to skimmed milk to get his size down. At this point we changed GP.

At Ben's first appointment at the new surgery we were exceptionally lucky as the usual GP was off and we were seen by the amazing and extremely clever Dr Scott. She asked Tracey if we were concerned about the size of Ben's head, she then suggested referral to Dr Verber, Paediatrian at North Tees as she felt Ben may have a storage problem – this meant nothing to Tracey or I.

Ben was duly referred to North Tees and a short while later we all went for X-rays, Ben was brilliant, a little frightened but quite compliant. He was apprehensive about the machine but could be encouraged to lie still long enough for the x-ray.

A short time later, Tracey took Ben to North Tees for the results and I happily and in pure ignorance of the seriousness of the situation went to work. (I feel such a pratt when I think about this now).

I just thought it was something simple and fixable. About two hours later I got a phone call, Tracey was in tears and within a few seconds she handed the phone to my mother-in-law who told me to just come home. My boss at the time was fantastic and I just told her I was going home.

The x-ray had shown lipping on some of Ben's vertebrae and seemed to confirm a metabolic problem. Dr. Verber asked if he could take a urine sample. Dr Verber said he was ignorant of the conditions, but he did know there were several types and some where treatable and some were not. The worst case scenario would be that Ben may have a life expectancy of about 10. Tracey asked if these conditions were degenerative and he confirmed they could be.

Two weeks later Tracey, Ben and I returned to North Tees for the results of the urine test results. This confirmed that Ben had Mucopolysaccharidosis (I still have trouble spelling it so we just say MPS), but again Dr Verber was unsure of the type and the prognosis. He first mentioned Dr. Wraith at Manchester. Dr. Verber asked to take a blood sample to send to Manchester and we would then be contacted by Dr. Wraith and his team.

Dr. Verber advised us to keep off the internet, which we did. We do, however, have a large supply of medical books which we read very carefully trying not to scare ourselves. I felt Ben was MPS VI, but could not be sure.

Anyway the fateful day arrived, 31 October 31 2003 and Tracey, Ben and I set off for Manchester. The journey was obviously tense and conversation limited this was definitely without doubt the worst period of my life. We had a two hour drive and the whole journey was just awful. At that moment I would have sacrificed anything to swap places with Ben; I made so many deals with God, every deal possible, so many prayers.

We arrived at the Willink Centre and introduced ourselves, Ben made himself at home and eventually we were introduced to Dr. Ed Wraith. We went into a clinic room, closed the door and I thought; Dr. Wraith gave us a number ……., 1. That number means so much, so very very much; in Ben's case it means the difference between relative normality and death.

Dr. Ed Wraith explained about genetics, luckily both Tracey and I have some knowledge of biology and understood most of what Dr. Wraith talked about. He explained about MPS and the consequences, about the missing enzyme alpha-l-iduronidase and the consequences.

Dr. Wraith also explained that since June 2003 a drug "Aldurazyme" was now available to treat MPS I, I could have kissed Ed, but really he's not my type.

Due to possible issues with funding Dr. Wraith suggested that the best course of action was to put Ben on the under 5's trial for the medication and in the meantime write to North Tees to request funding.

In February 2004 Ben started on the under 5's trial and began with a series of pre-study medical examinations in Manchester. Ben is brilliant with doctors, he lets them examine him, they joke with him, he asks lots of questions, he always impresses me the way he's so compliant, I think he's special.

Throughout this year my wife was brilliant, she got up at 5.30 a.m. very Friday for a year to drive Ben to Manchester to receive his treatment. I went as often as I could but we could not afford to both lose one day's pay each week.

She'd set off at 6.45 a.m. and drive for over two hours with Ben, she'd sit for four hours while he received his enzyme and then drive back for over two hours, amazing, but it's what you do for your children. At the end of the trial Ben's treatment has transferred to North Tees. Ben loves the nurses there. I miss Manchester and the amazing people we met there, very special people. We meet parents of other MPS sufferers and realise how lucky we are - Ben is MPS I H-S.

Ben is in school now and he loves it. He has a statement of Special Educational Needs, which was virtually written for us by Sophie Denham of the MPS Society.

Just recently Ben moved from part-time nursery to full-time reception. His statement has been increased from 12.5 hours support to 25 hours plus lunchtimes, so we are delighted with this outcome. This statement brings with it Mrs Hall, (Ben's carer) who he loves.

Ben still has some major difficulties but overall he is doing remarkably well. Everyone knows Ben at school. He always appears happy and he has lots of friends and numerous girlfriends, although I'm not sure I approve of this just yet. He gets invited to all the parties which costs an absolute fortune.

He still has lots of difficulties relative to his peers physically but without the work of Dr. Wraith and his amazing staff we would have to live with MPS and all the profound difficulties it brings.

Ben will always be behind his peers but just maybe his only difficulty will be his ginger hair.

Hannah's Story

Hannah was born on New Year's Day 2004. On 27 April 2005, aged 16 months, she was diagnosed with MPS I, Hurler Disease. Sadly, if left untreated, a child with MPS IH has a life expectancy of 5-10 years.

The only treatment available is a bone marrow or cord blood transplant; if the engraftment of new donor cells is successful then the new cells should start producing the missing enzyme. In addition to a transplant, Enzyme Replacement Therapy with a synthetic enzyme has recently become available. In itself it is not a cure as it has yet to be fully proven that the synthetic enzyme crosses the blood brain barrier but it does reverse many of the symptoms. To date, four transplants at Great Ormond Street Hospital, London have taken place in conjunction with once weekly infusions of enzyme several weeks before and post transplant. To date the results have been very encouraging.

Hannah had her first infusion of ERT on 2 June. Her cord stem cell transplant took place on 9 August just over three months after diagnosis. This article summarises Hannah's journey so far.

Pre – diagnosis

Hannah suffered from re-current ear and chest infections. She was admitted to our local hospital twice for chest infections and twice for peri-orbital cellulitis. In March 2005 tests were carried out for a suspected Lysosomal Storage Disorder.

Hannah had been slow to reach physical milestones such as sitting and crawling and was starting to display physical signs of MPS I such as coarse eye brows, low nasal bridge, prominent forehead, umbilical hernia and a curve in her lower back.

Diagnosis and Preparation for Transplant

The devastating diagnosis was given to us in Great Ormond Street Hospital at the end of April 2005. We were offered the option of a bone marrow transplant but it would need to be carried out before Hannah's second birthday to have the best chance of success. Transplant carries many potentially fatal complications such as graft rejection, and Graft Versus Host Disease. The Director of the bone marrow unit gave us a 75% chance of success. A transplant is not a cure but the best treatment available.

We decided our only option to give Hannah the best opportunity for the future was a transplant, we were fully aware of the risks but hoped a suitable donor could be found, only 8 months till her second birthday. Hannah started ERT a few weeks after diagnosis, within three weeks we observed improvement in her joint mobility; she was crawling and much more supple. Whilst the search for a donor was underway Hannah had a full medical assessment including visits to Ophthalmology, Cardiology, Dentist, CT scan of the brain, and E.N.T. department.

Two adult 8/10 bone marrow matches were found. One cord 5/6 match. The BMT Consultant felt the cord option was preferable as cord transplants carry less risk of rejection and GVHD., Hannah had her Hickman Line inserted at the same time as grommets and adenoid removal.

Transplant

Hannah was in GOS for 18 weeks for her transplant, she had a rocky time. A straightforward transplant with no complications takes 6-8 weeks.

The ward has 11 rooms with filtered air systems and individual bathrooms. Each room has a parent's bed settee, TV/video, phone and storage facilities. The medical staff are very specialised in their field of work, and play specialist, physio, and occupational therapists, social worker, dietician, teacher (school age children) make up part of the team. Meals for patients are prepared in a 'clean' kitchen, sealed

and delivered freshly cooked, they must be eaten within 45 minutes. Only three named carers are allowed to be with patients until neutrophil recovery allows for more family visitors. Everyone with access to a BMT patient must wash their hands thoroughly, wear clean clothes every day, wear gloves and aprons at vulnerable times and mustn't wear jewellery or watches. The patient must stay in the cubicle until neutrophil count starts to recover. Even now nearly six months post transplant, Hannah has to avoid crowded places. Minimising the risk of any infection during and after transplant is vital.

Conditioning

Ten days before transplant Hannah started her conditioning chemotherapy, this consisted of ATG, Busulphan, and Cycylophosphamide. The first part of the conditioning killed off her original bone marrow allowing space for the donor cells to grow and the second phase was to completely kill off any white blood cells which could fight with the new cells. At the time Hannah coped very well and continued to eat and play as normal.

Transplant

Many people, including ourselves, initially believed that a bone marrow transplant involved major surgery, however it is as straightforward as having a blood transfusion, although it is very special blood!

Post Transplant

Hannah had a stormy post transplant period. The first three weeks post transplant were very smooth. However in early September Hannah started losing blood from her intestine, at first it was thought to be caused by a stomach ulcer but later diagnosed as being from the lower small intestine but no definite cause could be identified. Hannah was put on total gut rest and had all her nutrition intravenously which is called Total Parental Nutrition or TPN. After two weeks of more or less continual blood loss and consequent large quantities of blood transfusions and drugs the bleeding stopped. During the same week as the bleeding stopping we were told that Hannah was producing 72% donor cells. This was fantastic news as if she had not showed any signs of producing new cells by the end of that week her old bone marrow would have to have been put back in to 'rescue' her.

As a consequence of all the drugs and blood clotting agents used to stop her intestinal bleeding Hannah developed Veno-Occlusive Disease. This causes an enlarged liver and spleen but was controlled by a two week course of drugs.

At the end of September, 46 days post transplant, Hannah became increasingly sleepy and lethargic, a CT scan of the brain revealed that she had developed acute Hydrocephalus and that a shunt was urgently required to drain the fluid from around her brain into her stomach as the pressure was too high. The operation went smoothly and Hannah made an excellent recovery and was far better seven days after the operation than she had been for weeks.

However for some unknown reason she developed a brain haemorrhage along the path of the shunt tube causing it to block and the pressure around the brain to increase. Within 24 hours her condition deteriorated considerably. She suffered four seizures and became non-responsive to pain stimuli, her coma score was 2; 15 is normal, 1 is a coma. Her condition was critical and life threatening. In emergency theatre an external drain was inserted, a bed was reserved on intensive care but Hannah was able to breathe unaided. She was moved from Bone Marrow to the Neuro Surgery ward for four weeks. A new internal shunt was fitted but blocked two days later. A second external drain was fitted and finally 16 days later another shunt was inserted. Amazingly Hannah has made a fantastic neurological recovery.

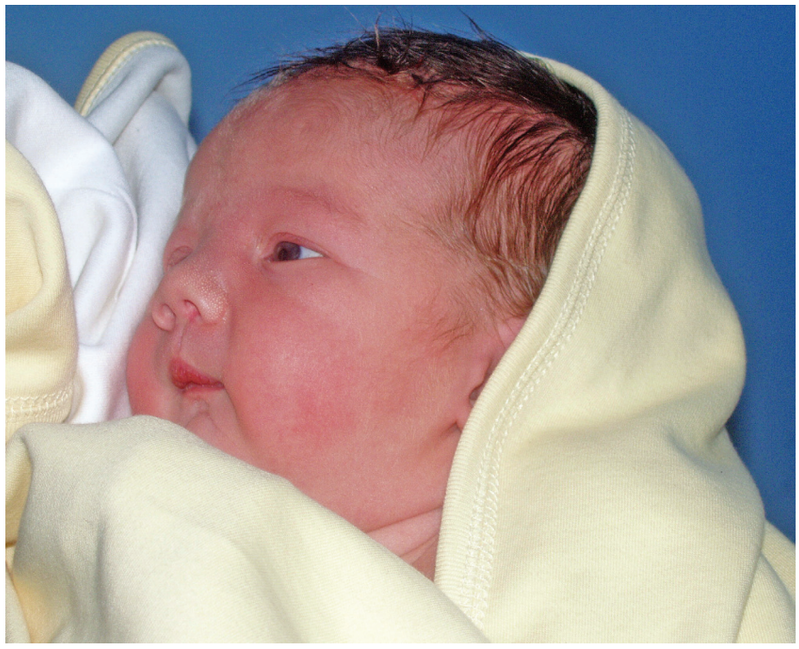
During the entire post transplant period Hannah had five positive cultures for bacterial infections in her Hickman Line, inevitable but easily treated. Close to discharge a new line was inserted to replace the original. Hannah was on TPN for a prolonged period. However four weeks before discharge she started Naso – Gastric feeds at 2ml per hour! On discharge Hannah could tolerate 4 x 200ml per day.

Discharge

Transplant took 18 tough weeks but we got there in the end! Her engraftment is increasing towards 100% and her body produces the once missing enzyme. Hannah adjusted to coming home really well and within a week of being home was eating, five months on she was having three meals a day plus snacks! Her legs are still very weak and Hannah finds standing almost impossible. In the short term Hannah has physiotherapy and needs feet splints.

We work closely with our community team and Great Ormond Street and will continue to do so for some time. Until her counts are better recovered she is restricted as to where she can go, who she can see and even the types of food she eats. We hope her quality and longevity of life have been vastly improved. Our thanks go to all the medical staff, family and friends that have and continue to support Hannah. This is only a brief summary of the months of pain, joy, hope and love we encountered.

For the full story visit www.caringbridge.org/visit/hannahcooper



Congratulations to Clare Cogan, Senior Advocacy Officer, and her husband, Will, on the birth of their baby, Joseph William Cogan, in January 2006.

Getting a Passport

The passport office has introduced strict new rules on how a passport photograph should look. For example, both children and adults have to be 'full face' looking straight at the camera with neutral expression, mouth closed and eyes open. Many MPS children would find it difficult to understand such instructions and some with physical disabilities would find it difficult to pose in the required manner.

Members need to be aware of the new requirements and to start applying for a passport well before it is needed, in case it takes several attempts to get a photograph passed as suitable.

Scottish MPS Conference

16 June 2006

The MPS Society is to hold a conference at the Hilton Edinburgh Airport Hotel on Friday 16 June 2006. We do hope families will be able to attend this event as it will provide an excellent opportunity to meet with professionals such as Dr Ed Wraith, Dr Maureen Cleary and Dr Peter Robinson, amongst others.

As part of the Society's programme of events for the first half of 2006, we are also hosting a family get-together on Thursday 15 June 2006 at the hotel between 5.30 – 8.30 pm to enable families to come together for a relaxing social evening over dinner. We look forward to seeing as many of you as possible and full details will be sent out to all families in the area in the near future.

4th Fabry Outcome Survey (FOS) Investigator Meeting

By Sophie Denham

On 21st January, I was fortunate to be invited to attend the 4th FOS investigator meeting, which was held in Brussels. The meeting was a one-day meeting, where professionals from around Europe came together to discuss the outcomes from the various working groups that have been set up to look at specific aspects of Fabry disease and its treatment.

The Fabry Outcome Survey (FOS) was established in 2001. It is a European database, which collects data on the natural history of Fabry disease and evaluates the efficacy and safety of ERT. There are approximately 752 patients across Europe who are enrolled in this study. This figure continues to rise and includes males, females and children.

Eight different working groups presented their findings and outcomes on various aspects of Fabry disease, such as Renal, Cardiac, Central Nervous System, Gastro-intestinal, Ear and Eye. It also included the studies on females and paediatrics as well as other studies that have taken place.

The results were all very positive and generated a lot of discussion. These meetings are invaluable to allow professionals in the field of Fabry disease to present their findings and discuss the outcomes.

Although there are huge developments and results are positive, the general consensus from all the working groups was that there is a need for more patients to enrol on the study to allow the data analysis and outcomes to be more effective.

Thank you for allowing me to be a part of this extremely informative weekend.

MPS Spain

The Spanish Society for Mucopolysaccharide and Related Diseases

Mercedes Lopes writes about the first national conference of the Spanish MPS Society which took place in October 2005

It is my pleasure to write this article for your magazine about the First National Conference of the Spanish MPS Society, 14-15 October 2005.

The conference was held in the city of Calpe (Alicante) because living there is a family affected by Sanfilippo Disease. Thanks to this family we were able to get a large room in which to hold the Conference and they obtained some sponsors. Of course, our Society organised the meeting.

It was very successful with the assistance of many families affected by MPS and Related Diseases and the doctors, Amparo Chabas, Ma Josep Coll, Antonio Balldellou and the researchers Lluisa Vilageliu and company.

The Conference started on Friday 14 October in the Culture Room with the Mayor of Calpe welcoming all the attending members, and then followed a dinner with the doctors and families affected.

The next day the Conference started and the topics covered included: the diagnosis of the diseases, genetic counselling given to affected families, an update on treatment and psychological advances.

Lunch was a typical Spanish paella for all the families with doctors and friends. In the afternoon Dr Lluisa Vilageliu and assistants from the Biology Faculty of the Barcelona University told us the latest news from their research into Sanfilippo Disease. This research has been made possible through a grant from our Society.

Then two parents with children affected by MPS I and MPS II told us about their experiences with Enzyme Replacement Therapy. The Conference was closed by the President of the Society, Jordi Cruz who explained all the work of the Society, the objectives, the grants for research and made a tribute to two affected families who work very hard for the Society. The charity dinner was very successful with raffle prizes donated to the Society.

During the day a team of volunteers looked after the affected children and babies in the crèche, while another team of volunteers went to the theme park all day. The children had a very enjoyable day in 'Terra Mitica'.

Benjamin's Story

by Rita Hausken Barkhodaee

A beautiful September day in 1996, Wednesday 29th to be exact, was shattered to pieces by very few words. "Benjamin is suffering from a progressive illness, either Hunter or Hurler, and will most likely die at a very young age."

What? When? "When will he die?" I asked in disbelief. According to the paediatrician the statistics said sometime between 7 and 12 years of age. How was this possible? In front of us we had a perfectly wonderful two-year-old boy! Did she really want us to believe we would only have him around for five more years? What should we do? How would he die? "It sounds like you are telling me that Benjamin has AIDS and will die in five years of pneumonia or some other weird thing," I said. She looked at us and then Benjamin and then back at us. "There is nothing to do but to make the most of what is now. And then move on afterwards."

These words – "There is nothing to do but to make the most of what is now. And then move on afterwards" – really upset us. Obviously it made us tremendously sad, but it also made me extremely angry. Who was she to tell us what to do? These words would turn out to be the key to Benjamin's life and what this story is all about.

We went to my parent's house after seeing the doctor and, typically, the diagnosis was presented as we had heard it – without any hope. And when my mother tried to put the word "but" in a sentence, I jumped at her. I was upset that she did not understand that "THERE IS NO HOPE!" Then she looked at us and said: "As long as there is life there is hope!" How true this is! And how this would turn out to improve Benjamin's life and ours.

Three weeks after we got the diagnosis we decided it was time to look on the bright side. We took a holiday together with my parents and went to the Canary Islands, where Benjamin then turned two years. After all Benjamin was doing well at the time and we had to enjoy it. Doing well may be a bit of an exaggeration, he was constantly struggling with all sorts of infections, joint stiffness, a painful umbilical hernia, and no more than 20 minutes consecutive sleep at night. But still, he was a happy boy.

"Nothing to do. Make the most of what is now." The words were resonating in my brain. Could we really just sit there and wait? Was there nothing to be done? If there was anything that could be done, the paediatricians surely must know. Or was that to be expected? Could they know it all? We must all take responsibility for our own actions or lack of action. Sure, we should expect the "experts" to tell us what to do, but in my experience, and I am sure, in many of yours, we know that is not always the case. So I decided to hunt for information. In Norway at the time, the information was limited. Also I felt it was not the right time to get in contact with other parents in Norway in similar situations or the small organisation that existed. Why not? Because I felt I had to be selfish! Someone reading this might be offended by me saying this, but we Norwegians are not known for being the most optimistic people in the world. Just read Henrik Ibsen's "A Doll's House" and you pretty much get the picture. So, I could only turn to people that somehow would give me a positive view on things.

In January 1997, I came across an article written by Dr. Emil Kakkis and I decided to send him a letter. He wrote back telling me that they were close to getting started on an enzyme replacement therapy on humans. However, he said I should not get high hopes for Benjamin. After all, Benjamin was diagnosed with Hurler, not Hurler Scheie. Still, this was music to our ears! Somebody was actually working on a drug for Benjamin! How wonderful! I also got in contact with a group of American families on a website and they made me feel good about what we were doing. It also turned out to be very useful for getting to grips with Benjamin's problems before they became a problem.

Since the paediatricians at the hospital noticed that we took a very active role in Benjamin's health and were able to contribute in a constructive manner as to what we should do in various situations, they listened to us. They also listened when I eagerly talked about the enzyme replacement therapy that "would come and be a wonderful treatment for MPS I". But I am not so sure they believed in it. This resulted in some doctors repeating the message that we had heard that dark day in 1996: "There is nothing to do. Make the most of what is now, and then move on afterwards. Do not waste your time on this." Okay, so you are telling us to spend our time and money on golf instead?

In 1999 we were involved in a survey where Dr. Kakkis and his group were looking for children to participate in a study. However, even though Benjamin was doing very well for a 5 year old, he was a Hurler, so he was not included. But these steps in the right direction of getting enzyme replacement therapy out to children was an indicator to keep "preaching" about this treatment. And I did talk about it, to everybody I believed could make a difference.

Anybody that I believed should know about this and could tell others. Looking back, I realised that none of the doctors I spoke to, even those with close connections to families with children suffering from MPS I, mentioned that this study that was going on. I am sure it was because they did not want anybody to raise their expectations for a treatment that might arrive too late for their children.

My question is: Was that the right decision to be made by these doctors? Would it be wrong to inform about a study being conducted with the aim to actually find a drug that can improve our children's lives? Would it be wrong to tell us that there are people out "there" that take such interest in our children's illness that they are willing to spend time and money on research? Would that be so much worse than telling us: "There is nothing to do. Make the most of what is now, and then move on afterwards"? Could this possibly make life worse?

As time went by, Benjamin's health was deteriorating. In 2001, when he was six-and-a-half, he was hospitalised due to respiratory problems. Due to low oxygen level he was given 100% pure oxygen through a mask in the hope it would help him. Unfortunately this was the worst thing he could get as he could not exhale and it resulted in a high CO2 level, and he went into coma. They tried to intubate, first one doctor then another, but it was impossible. Observing every step of the way in Benjamin's room was terrible. To suddenly realise that this illness would kill him, was in all honesty a shock. However, when I concluded that there was nothing more to be done I remember thinking: "You had a good life Benjamin and I am glad you could go to sleep now. We have been so blessed to have you with us."

However, Benjamin managed to pull through with the help of another anaesthetist that was called upon and had seen Benjamin before and knew of his throat problem. How lucky we were! A second chance! But it made me realise that all the work I did to make sure they all knew about enzyme replacement therapy might not benefit Benjamin after all, but it could still help other children. And this gave me more strength to carry on. The hope we had and the knowledge that somebody was working on a drug for Benjamin and others with MPS I really was of great help in the years from when he was diagnosed up until now.

We realised that Benjamin might not live to see the day of Aldurazyme, the name given to the drug, but the hope made us feel alive!

In 2002 I remember attending our first MPS get-together in Norway and I was so optimistic as I knew it was just a matter of time before the drug would be released. How disappointed I was when I understood that hardly any of the MPS families knew what was about to happen. Why did they not know? Some had seen specialists I had talked to about the treatment many times. One mother was furious as to why she had never been told. She did not believe there would be anything to do for her child but just knowing that something was being done would have helped her emotionally. Is it to be expected that all parents should do their own work to keep themselves updated on the latest research? Is it prohibited for doctors to talk about possible future treatments?

In the spring of 2003 we finally got the word that Aldurazyme had been approved by the FDA and was to be released on the European market shortly. Finally! This was exactly what we had hoped for. Somewhere along the line we had got the understanding that Benjamin had to start before he was 10. After that he would be considered "too old". We did not have much time as he was now 9. However, I was convinced that we would be rewarded for the work I had done in constantly informing "the right people" about this treatment. So when we started the

Continued overleaf

Benjamin's story

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application process in September 2003 we believed it would be approved quickly.

Does anyone think about how much a prisoner costs society? I never thought about this until March 2004 when our application for Aldurazyme was turned down. Why was it turned down? It was too expensive and nobody wanted to pay for it! So we include prisoners in the national budget and are willing to spend even more money on each of them to keep them safely behind bars, but treating a child with a medication that will extend his or her life, and make more liveable for both child and parents, that doesn't count. That we just cannot afford. How unfair!

In 1996 we had been told Benjamin would only live a few more years. Yet this message did not seem to trigger any sense, a full 8 years later, that his case was a matter of urgency whilst processing the application form. They managed to waste five months before they turned it down. Imagine how long five months of a 7-12 year life expectancy actually represents. And they honestly thought we would not fight back?

The hospital advised us to go via the proper channels, in other words send a letter to the health authorities and wait for their feedback. Alas, the only channel that works, apparently, is found in the media. On 16 March the story was released on one of the largest TV channels in Norway. This forced the Ministry of Health to review the case, which stunningly took only two hours before they instructed the local hospital to "pick up the bill" and get this boy his treatment.

On 14 April 2004 Benjamin got his first injection and he is still getting it every Thursday. He is doing very well and is bringing us lots of joy and happiness. His physical improvements are overwhelming!

His weekly presence at the hospital has also changed many people working there by giving them a feeling of making a difference. This is important for Benjamin, and for us, as it promotes emphasis on being positive and listening. Doctors increasingly listen to their colleagues, the nurses, the parents and patients. It has changed us as parents. How? We truly understand the value of being part of a team at the hospital. We have learned the value of good communication and trust. By exercising those two skills there is no room for misunderstandings as everybody involved has one interest, and that is to make Benjamin's life better. What more can any parent wish for?

Today we are looking back on the day of 29 September 1996 and we are grateful for the words "There is nothing to do but to make the most of what is now. And then move on afterwards." These words made us realise that we had to fight for a hope, and this fight kept Benjamin alive long enough to see the day that our hope could physically be touched, on 14 April 2004, when he started his treatment with Aldurazyme.

Do you have a great story to share with us?
Send an email to
newsletter@mpssociety.co.uk
or phone
0845 389 9901

Drug Information Association Meeting, 6-8 March 2006, Paris

The MPS Society is a member of Eurordis, the European Association for Rare Diseases. It is as a result of our relationship with Eurordis and the achievement of the MPS Society in the UK, ensuring our working towards members having access to new therapies that I was invited to speak on 'Unnecessary delays in patients' access to new medicines are medically, ethically and politically unacceptable' at this important meeting attended principally by the pharmaceutical industry and regulators from throughout Europe.

International Symposium on Mucopolysaccharide and Related Diseases

29 June - 2 July 2006
Venice Lido, Italy

The Symposium is possibly one of the most challenging to host with the complexities of bringing scientists, clinicians, the pharma industry and affected families together all to share their experiences of MPS.

From the outset the organisers, the scientific committee under the chairmanship of Professor Maurizio Scarpa and the Italian MPS Society have set out to make this meeting in Venice informative and inclusive for all those attending.

The MPS Society is privileged and grateful to have been able to propose speakers for the family conference. The following members will be speaking:

Hannah Donegani	'My brother and sister have Sanfilippo disease'
Myles Broughton	'Achieving my full educational potential'
Joanne Evans	'I'm at university'
Fer Pidden	'My daughter has a gastrostomy'
Peter Hawkins	'Living with Sanfilippo disease'

We are delighted that a few MPS families have booked for this International Meeting but for those for whom this meeting is out of reach the MPS Society is already planning its 25th birthday National Weekend Conference 29 June-1 July 2007 at the Northampton Hilton. Booking forms will be sent out with the Autumn magazine.

Spanish MPS Conference

In January 2006 Christine Lavery was a guest speaker at the conference organised by ADAC - Asociacion de Deficiencias de Crecimiento y Desarrollo. The subject of her presentation was the MPS Registry.

What was noticeable from the number of MPS families attending was that very few were affected by MPS III, Sanfilippo Disease. If we assume that Sanfilippo has the same incidence pattern as the UK and Germany then there has to be around 100 children and adults with MPS III in Spain. Christine would like to thank her hosts, ADAC, and in particular Susanna Diaz for the invitation.

Editor's Note: ADAC provides support to people with genetic diseases causing short stature.
www.adac-es.net

In 2004 Jordi and Mercedes Cruz set up a patient organisation for Sanfilippo Disease in Spain. This is now established to support all families in Spain affected by a Mucopolysaccharide or Related Diseases.
www.mpsesp.org

Clinical Waste

A delicate subject we know for many of you, but we thought it fair to share with you our experiences of clinical waste and the collection of it.

Clinical waste is classed as the used/soiled nappies for young children, young adults and adults. This does not include baby nappies as these can be placed in nappy sacks/bags. Sooner or later the dustbin collectors will and can refuse to collect your bin, which contains any clinical waste.

We have recently been successful in arranging for a clinical waste collection for a family who knew the collection existed but were having trouble getting it arranged for themselves.

If you are also struggling with getting a clinical waste collection arranged, please do not hesitate in getting in contact and we will happily point you in the right direction or contact the council on your behalf, to help get this arranged. This is something you are all entitled to.

Benefits Fact Sheet

The Contact a Family sheet 'An introduction to Benefits and other financial help' has been updated to reflect recent changes. The fact sheet outlines the range of benefits and financial support that families may be entitled to, as well as details of other organisations that may provide financial assistance.'

Copies of the fact sheet can be downloaded from www.caf.org.uk or telephone Contact A Family on **0808 808 3555**.

New portable pump adds mobility to infusion process

Fabry patients in the US have just begun using a new pump system for infusions. Although Fabry patients in the US and UK have been able to access home treatment for enzyme replacement therapy many still feel that this is difficult because they are still attached to an IV pole and pump.

Jason Frett, a Fabry sufferer from the US, states that 'doing home infusions allows me the flexibility of scheduling around work and activities. Now with the portable pump, mobility is must easier. This is a great process for travelling, or even riding in a car at the time of infusion, if necessary.' Jason continues 'I feel less restricted and more in tune with my family during this time.'

The Challenging Behaviour Foundation Parents' Email Network

The Challenging Behaviour Parents' Email Network is for parents caring for individuals with severe learning difficulties (both children and adults) who typically display behaviour which may put themselves or others at risk, or which may prevent the use of ordinary community facilities or a normal home life.

There are many local and national support groups for specific disorders like the MPS Society. However, different individuals are affected differently by different disorders and you may feel you are alone in trying to cope with your son or daughter's aggression, self injury, stereotyped behaviour or disruptive and destructive behaviours. If that is the case, then the Challenging Behaviour Parents' Email Network may be for you.

For more details of the Challenging Behaviour Foundation Parents' Email Network contact the Challenging Behaviour Foundation:
Tel: **01634 838739**
Email: info@thecbf.org.uk
www.challengingbehaviour.org.uk

Holidays at The Scout Holiday Homes Trust

The Scout Holiday Homes Trust is a registered charity founded in 1969 buy former scouts in order to provide suitable self-catering family accommodation for families who have a member with special needs, regardless of any Scouting connections.

The Trust has chalets and caravans at sites located around the country which are maintained by volunteers. The facilities are designed to be standard wheelchair access but are not suitable for individuals who are totally dependent on a wheelchair for mobility.

For more information visit
www.scoutbase.org.uk
or telephone **020 8433 7290**.

Gene therapy clinical trial for Mucopolysaccharidosis type VII

We have a pending gene therapy clinical trial for mucopolysaccharidosis type VII (MPS VII). The target of this clinical trial will be the hematopoietic (blood) system. In this clinical trial we will isolate the blood stem cells from a child with MPS VII and add a functional copy of the defective gene into those cells. We will then transplant those genetically modified blood stem cells back into the patient after a mild (myeloreductive) conditioning regimen. The genetically modified blood stem cells will then give rise to more mature blood cells that now produce the deficient enzyme, circulate through the body, and can share that enzyme with other organs. In this way we hope to provide a permanent source of the deficient enzyme to the patient. The basic procedure is similar to bone marrow transplantation but will use the patients own blood stem cells rather than stem cells from a donor. This will reduce or effectively eliminate a severe condition referred to as graft vs. host disease where the donor blood cells attack the patient.

It should be noted that this is an experimental therapeutic approach. We have shown that this approach works in human MPS VII cells in a humanised (xenotransplantation) mouse model of MPS VII [Hofling AA, et al., (2004) Molecular Therapy, 10:106; Hofling AA, et al., (2003) Blood, 101:2054]. We believe that this same approach will work in children with this disease. For additional information please contact:

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If any MPS VII families overseas are interested in learning more about this clinical trial, please contact the Washington University School of Medicine as above.

INOTECH starts **Morquio Type A clinical program**

Basel, Switzerland, 21 December 2005

Dear Morquio Community

It is a great honour for Inotech Biotechnologies Ltd. to announce that Saint Louis University, Prof. Shunji Tomatsu, and Inotech initiated to collaborate and conduct the Morquio Type A clinical program with Morquio families, Morquio Foundations, and MPS Societies.

Inotech is aware about the commitment and responsibility towards all Morquio children and families and will do its best to bring this program into clinic by the beginning of 2007. Inotech guarantees and confirms to the Morquio Community to take the Morquio project as the No. 1 priority project.

Two months ago, a close friend of mine asked me for support within the biotech industry to find a commercial partner for the enzyme replacement therapy. His daughter, Sophie (6), has Morquio and he has heard from Prof. Tomatsu having a new cell line producing said enzyme. That's where we entered into the Morquio Community and since then, I am very touched about Sophie and as a result, highly motivated to set-up as soon as possible this respective enzyme therapy.

Who is Inotech?

Inotech is a biotech company based in Basle, Switzerland. We focus on the GMP production of biopharmaceuticals derived from mammalian cells. We show a R&D department, clean rooms and respective bioreactor facilities as well as all regulatory know-how and persons. Inotech has its core competence in the development and GMP production of recombinant biopharmaceuticals as e.g. monoclonal antibodies, hormones, and enzymes. We have a highly motivated team with long-term industrial background.

What are the next steps?

Today, we have started the Project with the production of enzymes for a confirmatory pre-clinical study which will be conducted together with Prof. Tomatsu. In parallel, we will file the respective regulatory papers towards the FDA and organise all topics in order to be able to start the first clinical trial in Q1 / 2007 according actual planning. This initial clinical trial will be executed and supervised by Prof. Tomatsu and his team at St. Louis University.

As a prerequisite for these clinical studies we need your help. Data collection of as many as possible of Morquio children is vital. Together with the International Morquio Organisation (the Morquio registry program) and MPS Societies, we will inform you precisely about the status of the project on a regular basis.

Yours sincerely

Dr. Christoph Heinzen
CEO
Inotech Biotechnologies Ltd.
Basel, Switzerland

MPS IV

Research Update

MPS II

Shire Pharmaceuticals announced on 24 November 2005 that it had submitted a Biologics Licence Application (BLA) with the US Food and Drug Administration (FDA) for idursulfase under the trade name Elaprase formerly referred to as 12S. If approved Elaprase would be the first human enzyme replacement therapy for the treatment of MPS II. Idursulfase has already received fast track designation from the FDA, and Shire has requested priority review of this submission, which would result in a six month review.

The BLA contain data results of the pivotal clinical trial which studied 96 patients during a 52 week period, and is the largest study ever conducted for an MPS disorder. The primary efficacy outcome assessments were distance walked during six minutes (six minute walk test) as a measure of endurance, and percent predicted forced vital capacity as a measure of pulmonary function. The primary endpoint combined these two components into a composite score. Patients who received 0.5mg/kg of Elaprase on a weekly basis showed a statistically significant difference in the primary efficacy endpoint, compared to patients receiving placebo. Additional data demonstrated improvements in key secondary endpoints

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

On 1 December 2005 Shire announced that it has submitted a Marketing Authorisation Application (MAA) to the European Medicines Agency (EMEA) for idursulfase for the treatment of MPS II. Review of a MAA by the EMEA typically takes 12 months.

MPS III

Shire Pharmaceuticals Group, as part of its research to evaluate new approaches to the problem of treatment of the central nervous system, is hoping to move its MPS III A program forward. If the trial to directly administer the enzyme into the central nervous system of individuals with MPS II is successful, they hope to expand their research initiatives to include MPS III A. The Shire Web site is www.shire.com.

MPS IV

In December 2005 it was announced that the Swiss company Inotech will be promoting a clinical trial of ERT for MPS IV A. The natural history information is critical for any drug development and follow-up. If you are a sufferer of Morquio disease or the parent of a son or daughter with Morquio disease we would be pleased if you would ask for a questionnaire to be sent to you to enable the Society to update your/your child(ren's) information on the MPS registry and provide anonymised data to Dr Tomatsu and Inotech that may assist in the natural history study.

MPS VI

On 15 September 2005, BioMarin Pharmaceutical announced that the Committee for Medicinal Products for Human Use, the scientific committee of the European Medicines Evaluation Agency, issued a positive opinion on the company's marketing authorisation application for Naglazyme. The committee's proposed label states that Naglazyme is approved in the European Union as a long-term Enzyme Replacement Therapy in individuals with MPS VI to treat the clinical manifestations of the disease.

MPS VII

Drs. Emil Kakkis and William Sly have received a grant to develop enzyme replacement for MPS VII. Good progress is being made in production development. At this point there is no timeline for a human clinical trial.

MPS VI

BioMarin Receives Marketing Approval for Naglazyme in European Union

Novato, Calif, January 30, 2006 – BioMarin Pharmaceutical Inc. (Nasdaq and SWX: BMRN) announced today that the European Commission has granted marketing authorisation for Naglazyme™ (galsulfase), the first specific treatment approved in the European Union for patients with the genetic disease mucopolysaccharidosis VI (MPS VI; Maroteaux-Lamy syndrome). As the first drug ever approved for MPS VI, Naglazyme has been granted orphan drug status in the European Union, which confers 10 years of market exclusivity.

Naglazyme has been approved in the 25 member states of the European Union, Iceland and Norway for long-term enzyme replacement therapy in patients with a confirmed diagnosis of MPS VI. All clinical post-authorisation commitments requested by the Committee for Medicinal Products for Human Use (CHMP) will be fulfilled through a voluntary surveillance program that will monitor patients on commercial therapy. No additional clinical trials are required. BioMarin will launch Naglazyme in the European Union on a country-by-country basis.

“I have observed the dramatic improvement enzyme replacement therapy with Naglazyme can have on patients afflicted with MPS VI and am pleased that the therapy will soon be available to individuals outside of clinical studies,” stated Ed Wraith, M.D., Consultant Pediatrician at the Royal Manchester Children’s Hospital located in Manchester, United Kingdom, and a principal investigator for Naglazyme clinical trials. “Naglazyme holds the potential to positively impact the lives of MPS VI patients and their caregivers.”

“We are excited that BioMarin has succeeded in bringing the first enzyme replacement therapy for MPS VI to market, first in the United States and now in the European Union,” stated Jean-Jacques Bienaimé, Chief Executive Officer of BioMarin. “With European commercial operations in place, we are ready to launch Naglazyme. Additionally, we are now positioned to partner with companies looking to bring other products for rare diseases to the European marketplace.” Mr. Bienaimé continued, “We are pleased with the growing sales and profitability of Aldurazyme, and with Naglazyme now approved in Europe and the United States, we expect combined worldwide sales of Aldurazyme by our joint venture and Naglazyme by us for 2006 to be in the range of $118 million to $132 million.”

About MPS VI

MPS VI (also known as Maroteaux-Lamy syndrome) is a debilitating, life-threatening genetic disease caused by a deficiency of the enzyme *N*-acetylgalactosamine 4-sulfatase. This enzyme deficiency leads to the accumulation of certain complex carbohydrates, glycosaminoglycans (GAGs), in the lysosomes, giving rise to progressive cellular, tissue and organ system dysfunction. The majority of individuals with MPS VI die from disease-related complications between childhood and early adulthood. Additional information can be found at **www.mpsvi.com**.

About Naglazyme

Naglazyme is the first and only enzyme replacement therapy indicated for the treatment of MPS VI. As the first drug approved for MPS VI, regulatory agencies in both the United States and European Union have granted Naglazyme orphan drug status, which confers seven years and ten years of market exclusivity, respectively. Additional information can be found at **www.naglazyme.com**.

Naglazyme is indicated for patients with MPS VI. Naglazyme has been shown to improve walking and stair-climbing capacity. The most common adverse events observed in clinical

trials in Naglazyme-treated patients were headache, fever, arthralgia, vomiting, upper respiratory infections, abdominal pain, diarrhea, ear pain, cough, and otitis media. Severe reactions included angioneurotic edema, hypotension, dyspnea, bronchospasm, respiratory distress, apnea, and urticaria. The most common symptoms of infusion reactions included fever, chills/rigors, headache, rash, and mild to moderate urticaria. Nausea, vomiting, elevated blood pressure, retrosternal pain, abdominal pain, malaise, and joint pain were also reported.

No patients discontinued Naglazyme infusions for adverse events and all patients that completed the double-blind portion of the trial continue to receive weekly infusions of Naglazyme. Nearly all patients developed antibodies as a result of treatment, but the level of the immune response did not correlate with the severity of adverse events or impact the improvements experienced in endurance. Because antihistamine use may increase the risk of apneic episodes, evaluation of airway patency should be considered prior to the initiation of treatment. Consideration to delay Naglazyme infusion should be given when treating patients who present with an acute febrile or respiratory illness.

About BioMarin

BioMarin develops and commercialises innovative biopharmaceuticals for serious diseases and medical conditions. The company's product portfolio is comprised of three approved products and multiple clinical and preclinical product candidates. Approved products include Naglazyme™ (galsulfase) for mucopolysaccharidosis VI (MPS VI), a product wholly developed and commercialised by BioMarin, Aldurazyme® (laronidase) for mucopolysaccharidosis I (MPS I), and Orapred® (prednisolone sodium phosphate oral solution) for inflammatory conditions. Investigational product candidates include Phenoptin™ (sapropterin dihydrochloride), a Phase 3 product candidate for the treatment of phenylketonuria (PKU). For additional information, please visit www.BMRN.com. Information on BioMarin's website is not incorporated by reference into this press release.

The MPS Society has produced an MPS VI news-sheet to coincide with these latest developments in MPS VI.

Copies are available free of charge from the Society.

www.mpssociety.co.uk
0845 389 9901


MPS VI News

A description of MPS VI

Mucopolysaccharidosis type VI (MPS VI) is a lysosomal storage disease in which affected individuals lack the lysosomal enzyme N-acetylgalactosamine -4-sulphatase (also known as arylsulphatase B, ASB). The disorder is recessive, affects males and females equally, and results in an inability to degrade the glycosaminoglycan (GAG), dermatan sulphate (DS). DS is an important constituent of the extra-cellular matrix, joint fluid and connective tissue throughout the body and in affected patients partially degraded DS progressively accumulates within the lysosome, ultimately causing cell, tissue and organ dysfunction by a largely unknown pathophysiological mechanism.

MPS VI is a rare disorder with a published prevalence varying between 1:500,000 and 1:1,000,000 live births. Historically, MPS VI has been divided into a 'mild' and a 'severe' form but in reality the disorder is a clinical spectrum with wide extremes.

SYMPTOMS

Clinically the disorder is progressive and the diagnosis may usually be made at 6-24 months of age when the children show progressive deceleration of growth, enlarged liver and spleen, skeletal deformities, coarse facial features, upper airway obstruction, and joint deformities. Progressive clouding of the cornea, communicating hydrocephalus, or heart disease (usually involving the valves but very rarely the heart muscle) may occur. Developmental progress is potentially normal but many children miss large amounts of schooling as a result of the serious nature of their disorder.

As the disease progresses children may become increasingly disabled as a result of the severe skeletal dysplasia and the majority of affected children without treatment die in their second decade from cardiopulmonary failure associated with respiratory infection or a surgical procedure in the setting of chronic cardiac disease.

One distressing complication that may occur in patients is a rapidly progressive visual loss which ends in complete blindness. This is usually associated with raised intracranial pressure and requires shunt surgery. Dysplasia of the odontoid with instability at the craniocervical junction also requires careful follow up and prophylactic decompression and fusion may be required in some patients.

There are a small number of patients who have a much less aggressive form of MPS VI although many have significant health problems. These patients may present with short stature and the diagnosis is established when minor radiological abnormalities are detected. Occasionally, the diagnosis is made in the ophthalmology clinic because of corneal clouding or by the rheumatologist who recognises the cause of the fixed flexion deformities of the fingers or other joint movement limitations.

In these patients, the disorder is still progressive but the rate of progression is slower when compared to patients with the early onset form of MPS VI. As adults these patients often require cardiac valve surgery or corneal transplant. Fertility is not affected in either sex.

Jonathan's Story

Jonathan is the third of four children and was diagnosed with MPS at the age of 8 months. At the time of the diagnosis, the most striking symptoms were a moderate blurring of the cornea, deafness and slight skeletal anomalies. During his first year he developed the full spectrum of MPS VI-related symptoms. Otherwise he became a happy and open-minded child who enjoys living with his healthy siblings.

Today, Jonathan, at the age of seven, is very short in stature (97 cm) and suffers from joint-contractions and a hip-dysplasia. For this reason, he uses a wheel-chair. Since Jonathan also suffers from apnoea, he wears a CPAP-mask at night which improves his sleep significantly.

Jonathan entered a school for physically-disabled children at the age of six. He enjoyed the place, was well integrated and progressed in skills rapidly. However, within a few months, his vision declined from 30% to less than 1%. This led us to move him in September 2005 to a school for blind children because he could not follow the regular school lessons in the first school. Since September 2005, Jonathan is collected from home daily by a school-shuttle and returned home on his individual school-schedule.

Jonathan has a healthy child as his best friend with whom he plays free of any restrictions. Max, his friend encourages him to move and play physically. His siblings also tend to treat him as a normal child. Like in any other family, the children occasionally fight and struggle with each other, but normally they get along very well and play very creatively with each other.

Since April 2005, Jonathan has received Naglazyme. He is treated weekly at the Mainz-University clinic. The infusion takes four hours. Except for the burden of driving the way to Mainz once a week, we are very happy that ERT for MPS VI is available. Since he receives regular treatment, his health has not deteriorated. Even Jonathan's vision remains stable and ERT has improved his fitness and mobility. Through the day, Jonathan is less tired and more mobile. His skin and hair have softened. He is now able to pull a shirt over his head which he never could do before.

Our hope is that for the near future ERT at home will be feasible since keeping everything organised for the three siblings during the weekly stay in Mainz is a great burden. Our three healthy children need to stay at their friend's home. The therapy in Mainz including the driving takes a whole day which could be avoided if home-service would become available.

We are full of expectations and hope to see how Jonathan will improve physically or at least stabilise his current health-condition.

Jonathan

Society for Mucopolysaccharide and Related Diseases

National Registered Charity No. 287034

Become a Friend of MPS

Would you like to show your support by becoming a Friend of MPS? We would welcome relatives, friends, overseas MPS families, professionals or indeed anyone interested in the work of the Society or the field of MPS & Related Diseases. This would encourage us, help us plan for the future and bring about more public awareness for this group of rare, genetic, life-limiting diseases.

What are the benefits of becoming a Friend of MPS?

Membership number and card
Quarterly colour MPS magazine
Quarterly colour fundraising newsletter
Annual report and accounts
Regular publication updates
Information on and preferential rates at MPS events
Priority ordering of MPS & Corporate Christmas cards

Phone 0845 389 9901
or download a form from
www.mpssociety.co.uk