MPS

Summer 2006

Care Today, Hope Tomorrow

Society for Mucopolysaccharide Diseases

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

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:
Faith and Victoria Parrott

CARE TODAY HOPE, TOMORROW

MPS Magazine

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Newsletter Deadlines

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

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CHIEF EXECUTIVE'S REPORT

I want to take this opportunity to share with you the staff team activities and an insight of the work we have been involved in and the tasks that lie ahead.

Many of you will remember Clare Cogan our Senior Advocacy Officer who is on maternity leave having had a baby boy, Joseph in January. Clare has decided not to return to work full time and left the Society's employment at the beginning of May. Sophie Denham who has been deputising for Clare has now been appointed Senior Advocacy Officer. This month we have had two new Advocacy officers join the team, Ashley Siberini and Steven Cotterell. Cheryl Pitt our MPS researcher who has just finished her three year project on the Psycho-social outcomes of MPS I patients post BMT went on maternity leave at the end of May. Cheryl will be back at MPS in the New Year to start her five year research programme that includes psychological projects for Fabry, Morquio and Sanfilippo disease. Lastly after nearly two years with the MPS Society Linda Norfolk said Goodbye at the beginning of June. We wish Clare and Linda well in all they do in the future, offer our congratulations to Sophie on her promotion, look forward to news of the birth of Cheryl's baby boy and warmly welcome Ashley and Steven to the Advocacy Team. At the end of July we will also be joined by Charmaine Scott who has accepted a one year, hopefully to be extended when the Department of Health confirm the second and third year's funding, post of Volunteer and Event Co-ordinator.

Up in Nottingham we are making excellent progress with the developments to the Childhood Wood. The new Memory Board has been ordered and should be constructed and ready for the Childhood Wood Remembrance Day on 16 July at Sherwood Pines. We are so pleased that the gathering will involve over 50 parents, brothers and sisters, friends and family who have lost someone very dear to them from an MPS or a related disease.

Perhaps one of the most sensitive matters we need to share with you is the future of funded Enzyme Replacement Therapy under the National Specialist Commissioning Advisory Group. None of us can have failed to hear of the budgetry difficulties facing the National Health Service. As a consequence we should advise that in two parliamentary questions we initiated recently the Secretary of State indicated that she hasn't made a decision whether the current arrangements for funding the diagnosis, clinical management and Enzyme Replacement Therapy for Lysosomal Storage Diseases will continue after 31 March 2007. A second question asking when the Secretary of State will make a decision resulted in a reply to say she doesn't know! We are talking to the Department of Health, politicians, NSCAG centres and the industry and will be in touch with those affected individually over the coming weeks with a plan of action. Let us hope our worst fears of going back to PCT funding or strategic health authority funding are unfounded.

Finally I want to welcome to the Management Committee our newly elected Trustees, Bob Stevens from Guildford in Surrey, Paul Moody from Bradford and Angela Brown from Livingstone in Scotland. Peter Conlin from Stockton has also become a co-opted Trustee. The Society is fortunate to have a very committed and vibrant board and I have no doubt that Bob, Paul, Angela and Peter's strengths and enthusiasm will enhance the board further.

Christine Lavery
Chief Executive

News from the MANAGEMENT COMMITTEE

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Management Committee Meeting held on 28-29 April 2006.

Personnel

Trustees were informed that the recruitment of two new advocacy officers was underway. Trustees were also informed that Clare Cogan had decided not to return to her post as Senior Advocacy Officer with the Society following the end of her maternity leave and Trustees thanked Clare for her hard work. Trustees agreed the promotion of Sophie Denham to Senior Advocacy Officer with effect from May 2006. It was agreed that the contract for Monica Hartwell, Fundraising Consultant, should be renewed for a further six months until December 2006. Trustees were informed that, following a full staff consultation, the MPS Office is now to remain open over the Christmas/New Year period to reflect the on-going needs of the Society's members.

MPS Annual Report and Accounts

The Trustees agreed the Annual Report and accounts for year ending 31 October 2005 and they were duly signed by the Chairman.

Governance

The Trustees reviewed the Society's current MPS Risk Register and agreed that there are no changes to be made at this time. The Society's Strategic Plan 2005-2010 was also reviewed and agreed by Trustees.

Policies

The Trustees reviewed and agreed the following policies: Data Protection, Child Protection, Moral and Ethical Policy, Moving and Handling Policy, Care Plans Policy, Policy Statement on Research, Policy on the Death of an MPS Sufferer.

MPS Office

Trustees were informed about several events which had taken place at the new MPS Office including the opening launch of MPS House on 12 April 2006. Trustees welcomed similar opportunities for the future.

Introducing Ashley

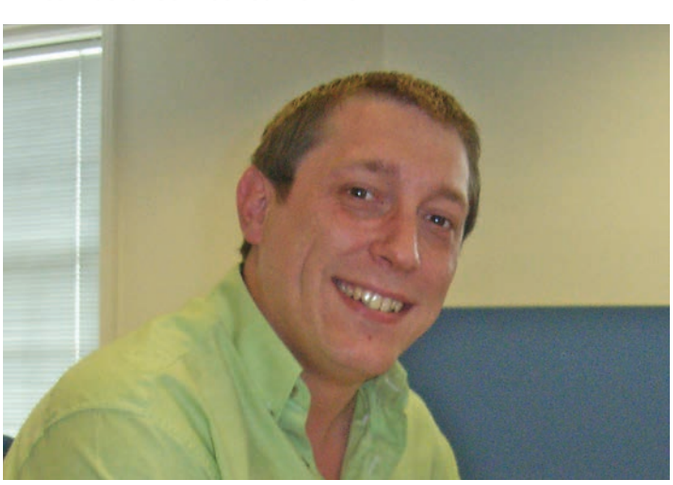
Well, it's that time of year again. New soldiers fully equipped and ready to march for the MPS Society. One of which is me joining the Society's advocacy team as an Advocacy Support Officer.

After a fairly disappointing and unfruitful career in the media I decided that I wanted to work in an environment where you can actually go about helping people achieve meaningful and rewarding lives. I began work with Buckinghamshire County Council in a day centre for adults with physical and learning disabilities. The rewards were instant. I had never been in the company of people who were truly grateful for what seemed to me to be the smallest amount of effort on my part. It was a humbling learning experience. I liked the idea of accessing existing community resources and helping the clients raise the profile of their needs, aspirations and crucially their human rights!

I began wondering what other areas I could put this new found bundle of ideas into effect. I began working with adults with mental health problems. Much of the challenges they faced were much the same as those with physical and learning difficulties. The main challenge was ignorance of the conditions! These were exciting times, new attitudes were being implemented and the work was more varied than it had ever been.

Now the "who is Ashley" bit. I'm quite a simple creature. I survive on a diet consisting mostly of football, loud live rock gigs, the gym and a few fine ales! When at home I mostly watch sports or spend quality time with my pets, Suzy the rabbit and Benny and Bertie who are rats.

I enjoy travel too, which is kind of handy when working for the MPS Society! Hopefully I will have the chance to meet as many of you as possible over the next few months, and if I can be of service let me know!



Announcements

Farewell to all by Linda Norfolk

I would like to say how much I have enjoyed working with everyone involved with the MPS Society over the past 18 months and hope you will continue to benefit from the support the Advocacy Support Team offers to you.

Shuban Selvaranjan was born on 27 February 2006 weighing 7lb 12oz. Many congratulations to Selva and Vani Selvaranjan on this wonderful news.

We were delighted to hear from Geraldine and Jake Rudham that **Isabella Rudham** was born on 7 March 2006. Isabella is a sister to Emily (aged 8) and Archie (aged 5, MPS II).

Many congratulations from all at the MPS Society to the Rudham family.

New Members

Aydin Kulle has recently been in contact with the Society. Her sister Suzan has a diagnosis of Fucosidosis. Suzan is 38 years old and lives with her family in London.

Mrs Christine Hedges has recently been in contact with the Society. Christine has a diagnosis of Fabry Disease. The family live in the South West.

Mr and Mrs Rodden have recently been in contact with the Society. Their daughter, Demi-Leigh, has a diagnosis of Hurler Disease. Demi-Leigh is nine months old. The family live in the North of England.

Ms Natasha Toghill has recently been in contact with the Society. Natasha has a diagnosis of Fabry Disease and lives in the South East.

Mr Peter Dine has recently been in contact with the Society. Peter has a diagnosis of Fabry Disease and lives with his wife in the South East.

Deaths

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

Reece Kemp who suffered from Hurler Disease and who died on 8 April 2006 aged 5 years.

Adam Miles who suffered from Hurler Disease and who died on 14 April 2006 aged 7 years.

Victoria Hendry who suffered from Hurler Disease and who died on 1 May 2006 aged 10 years.

Terri Hambly who suffered from Hurler Scheie Disease and who died on 6 June 2006.

Introducing Steve

Hello, my name is Steve Cotterell. On 5 June 2006 I joined the team here at the MPS Society as an Advocacy Support Officer. The areas that I am covering are Wales, Northern Ireland and the West of England. Prior to this I have worked in a care setting for the National Society for Epilepsy and then more recently for Buckinghamshire County Council as a Person Centred Planning Development Worker. Here I supported people with learning disabilities, their families and support staff in accessing services, developing support plans and advocating on their behalf.

In my spare time I **try** to play golf, I also enjoy cooking, gardening and pub quizzes. I have had the pleasure of meeting and speaking with a few families in my short time here and I am very much looking forward to meeting with and speaking to many more. If you would like to speak to me or need some support you can contact me on the usual office number or e-mail me at s.cotterell@mpssociety.co.uk

Family Noticeboard

In the last edition of the MPS Magazine we asked whether any of our readers would like to submit something for our family noticeboard.

This type of column had been suggested by one of our families. It could include any hints and tips from 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.**

Louise Lewis (Mum of Georgia, MPS III) writes:
I have a suggestion/information, which may be useful for other families. As constipation can often be common for children with Sanfilippo (and other MPS diseases), I thought I'd share our experience.

When Georgia was younger, she used to suffer with severe diarrhoea Fortunately, she outgrew that, but is now prone to constipation! At times, this was very painful for her, so our GP prescribed 'Movicol'. Initially, this worked well, however, it didn't guarantee a bowel movement everyday and I really didn't like the idea of giving Georgia yet another medication to take!

I started giving her fruit smoothies, half a glass every morning and evening and also adding raisins to her cereal (porridge oats) every day. This has worked really well. I haven't given the medication for over five months now, and her bowel movements are completely normal.

Dates for your diaries! 2006

16 July	Childhood Wood Remembrance Day
26 July	BMT Clinic, Royal Manchester Children's Hospital
29 August	Bristol MPS Clinic, Frenchay Hospital
20 October	Childhood Wood Planting
16 November	Northern Ireland Clinic, Antrim Area Hospital, Belfast
17 November	BMT Clinic, Royal Manchester Children's Hospital
29 November	Cardiff MPS Clinic, University Hospital of Cardiff
30 November	Bristol MPS Clinic, Frenchay Hospital

CHILTERN
District Council

MPS receives grant from Chiltern District Council

The Society has been awarded a grant of **£1538** from the Chiltern District Council Community Grant Aid Scheme 2006 for training for volunteers enabling them to work with children and young people with MPS. This will give them the opportunity to learn about the very rare metabolic conditions and the care needs of affected patients.

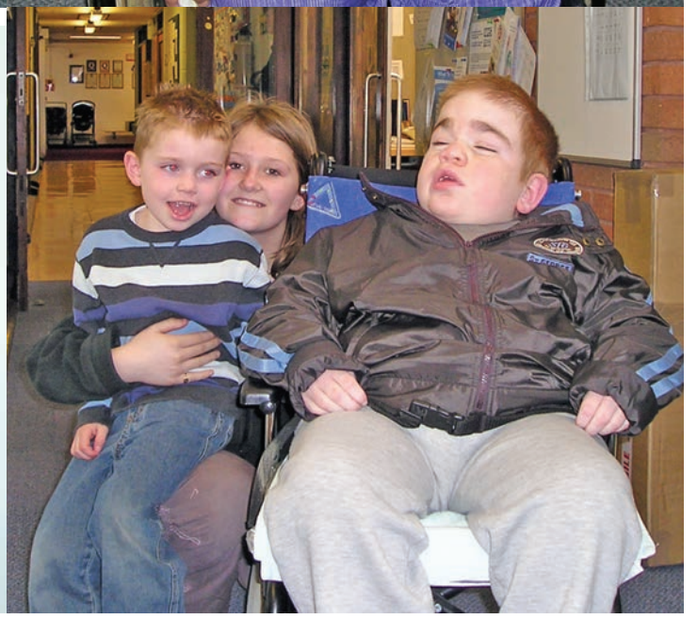
Birmingham MPS Clinic

by Sophie Denham

On a cold and very wet spring day, 19 April 2006, Birmingham held its third MPS clinic at the Victoria Special School in Northfield. Six patients attended this clinic and were seen by Dr Chakrapani, Dr Hendriksz and other professionals involved in the care and support of patients. Di Aspin stepped into Jo Hardy's shoes, who unfortunately was unwell for this clinic and for Louise who was on maternity leave, following the birth of her son William Oliver on 10 November 2005. (Congratulations to Louise from all at the MPS Society).

The clinic went well despite the poor weather conditions and as always there was plenty of parking with direct access to the entrance to dodge the rain clouds.

It was great to meet up with the families, some of you I have not seen for sometime. Our thanks go to Dr Chakrapani, Dr Hendriksz and Di Aspin for another successful clinic.

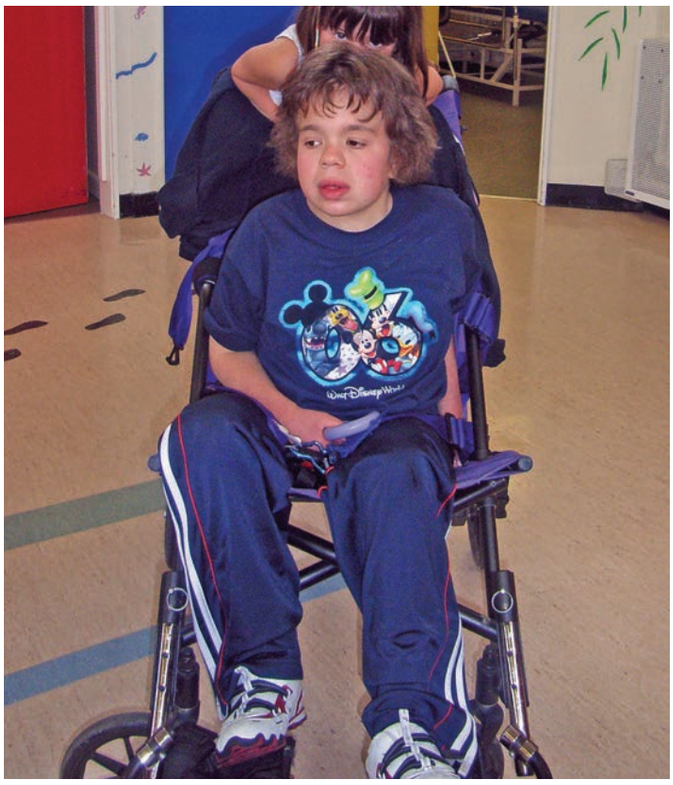


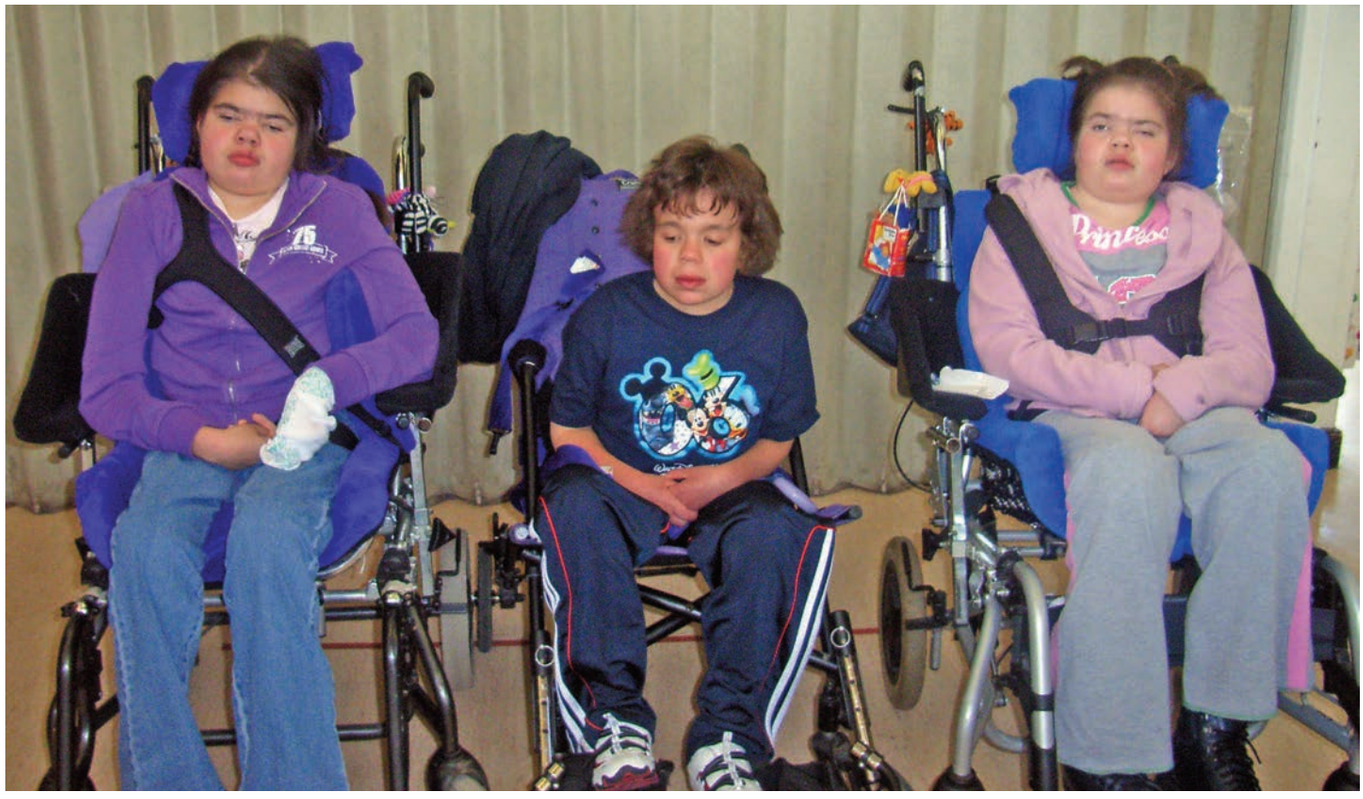
Bristol MPS Clinic

by Sophie Denham

On 30 May 2006, Bristol held their first clinic for 2006. There are three clinics at Bristol and we are holding further clinics in August and November. The clinic was very quiet which is unusual for Bristol as it is usually bustling with families catching up with new and old friends, enjoying tea and biscuits, children running around enjoying all the games, toys and dressing up clothes available and various professionals going about their daily work.

Even though the clinic was quiet, tea and biscuits were offered in abundance, even though some of the chocolate biscuits were a little melted by the end of the day. The clinic went well and I would like to thank all the families I met as well as saying a big thank you to the staff at Frenchay for all their help in setting up another successful clinic. Thanks also to Dr Jardine and Dr Ed Wraith who, without their commitment and support, this clinic would not be possible.





Photos this page: Top right: Hannah Chisling (MPS III), Bottom: Francesca and Josephine Kembry (MPS III) and Hannah Chisling (MPS III)

Photos page 8 clockwise from top right: Adikah Batool (MPS IV), Jack Onion (MPS II), Ali Anwar Khan (MPS IV), Landon Owen (MPS III)

Events in Northern Ireland

4-5 May 2006 by Linda Norfolk

Northern Ireland MPS Clinic

Sophie and I landed at Belfast Airport in torrential rain, having left a mini-heatwave in London! The clinic was held in Antrim Area Hospital near Belfast on Thursday 4 May 2006. As always, there was a full clinic list with Dr Ed Wraith, Dr Fiona Stewart, and Aoife Bradley from the Genetics Unit in attendance. The clinic ran very smoothly and it was a pleasure to meet the families face-to-face. I am working my way through the evaluation forms returned for any comments or suggestions regarding the clinic and the facilities so kindly offered by the staff in the hospital.

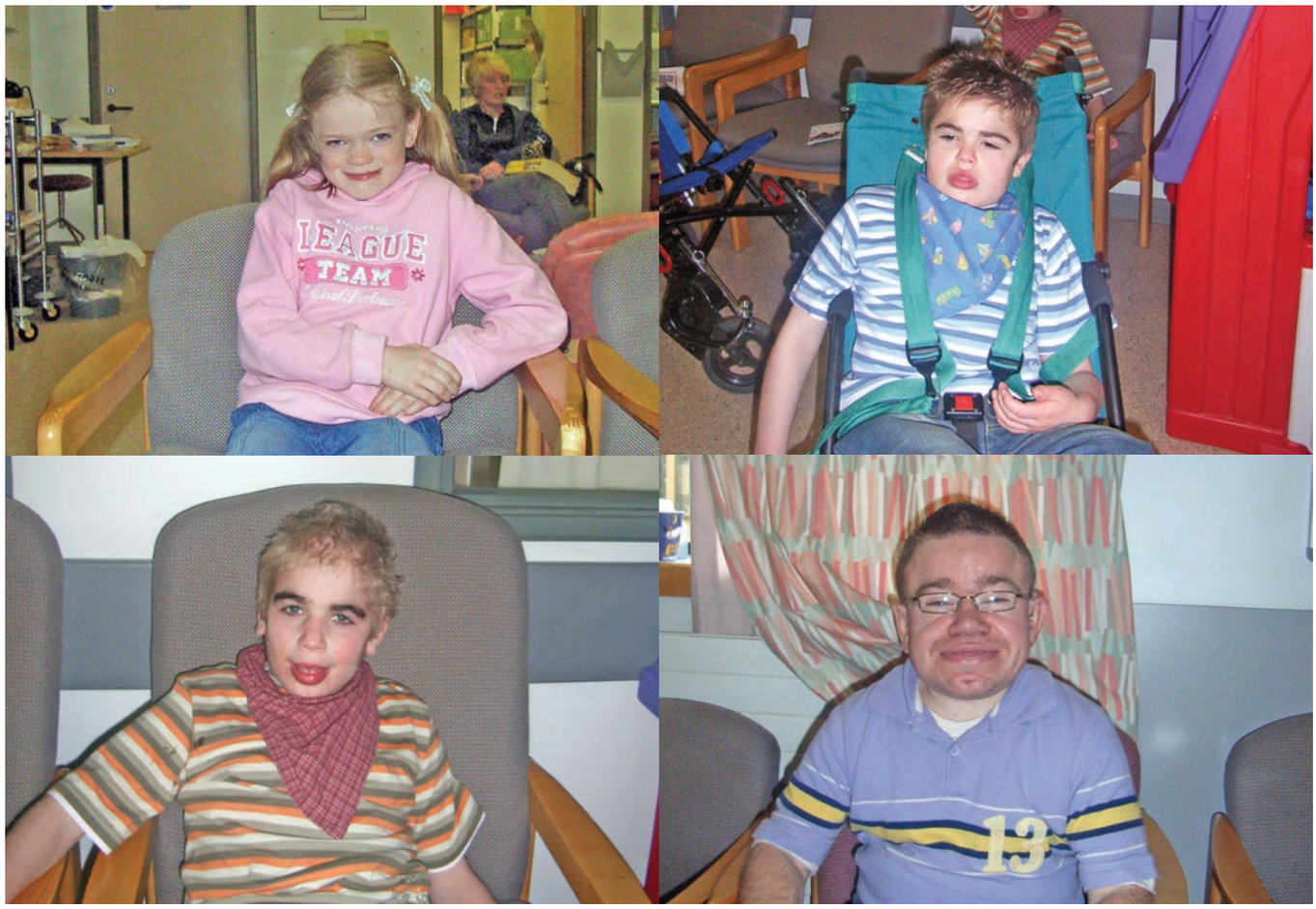
Our thanks go to Dr Stewart, Dr Wraith and Dr Stewart's secretary, Sandra Smith, for ensuring the smooth running of the clinic and also for the staff in the maternity and antenatal outpatients department who accommodated us and made sure we had the corner with the playhouse and toys!

Northern Ireland Social Gathering

Following the clinic at the Antrim Area Hospital, a Social Gathering was held at the Hilton Templepatrick for families, with 18 adults and 10 children attending. A fun evening was enjoyed by all over an excellent meal in the main restaurant of the hotel. A colouring competition and a mini quiz for the children were ably organised by Sophie Denham and some of the photos of the event are shown below. Many thanks to all those who attended for making this event such a success.

Photos page 10, Clockwise from top right: Dean Doherty (MPS III), James Stewart (MPS II), Aaron Doherty (MPS III), Chloe McCauley (MPS I HS)

Photos page 11, Clockwise from top right: Sarah, Roma Drayne (MPS IV), Kilian Drayne, Philip Walker, Kathleen Shannon, Hannah Shannon (MPS III and family)



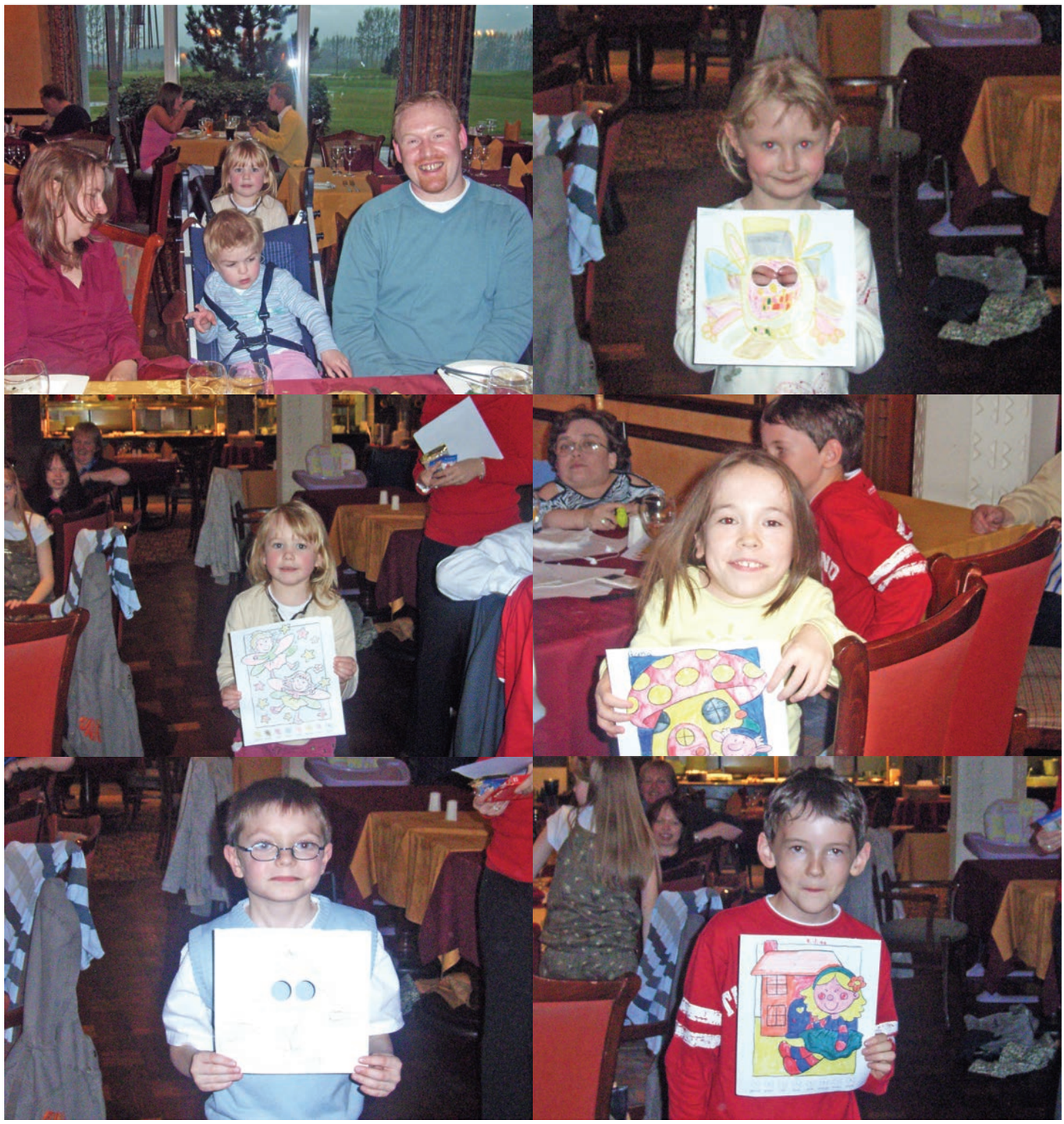
Northern Ireland MPS Conference

The MPS Conference in Northern Ireland was held at the Hilton Templepatrick in Belfast and was extremely successful with a very good turnout. Dr Alex Magee kindly agreed to Chair the Conference on our behalf.

In the morning, following registration and coffee, Dr Fiona Stewart gave her first presentation on Clinical Management of MPS and Related Diseases. Dr Bob Taylor gave a presentation on Anaesthetics - Risks in MPS, followed by a presentation on the Role of the Northern Ireland Children's Hospice by Karen Bleakley. After a short coffee break, our very own home-grown Sophie Denham, Senior Advocacy Officer, gave a presentation on Environmental and Practical Approaches to Behaviour Management in Sanfilippo Disease. Bernie Drayne, an MPS Society member, gave a presentation on Accessing the Education system in Northern Ireland - a personal perspective, followed by Mr Niall Eames presenting on Managing Scoliosis in MPS Disorders.

Following a delicious lunch the afternoon began with a presentation by Dr Ed Wraith on Current and future therapies for MPS and Related Diseases. Dr Fiona Stewart made her second appearance presenting on Accessing ERT in Northern Ireland and the Conference was very ably rounded off with a presentation by one of our members, Jennifer Johnston, with her talk on a Personal Experience of ERT.

All of the presentations were highly professional and very informative, with feedback from the delegates very positive. Requests for copies of the presentations made have been received and, where possible, will be passed on.



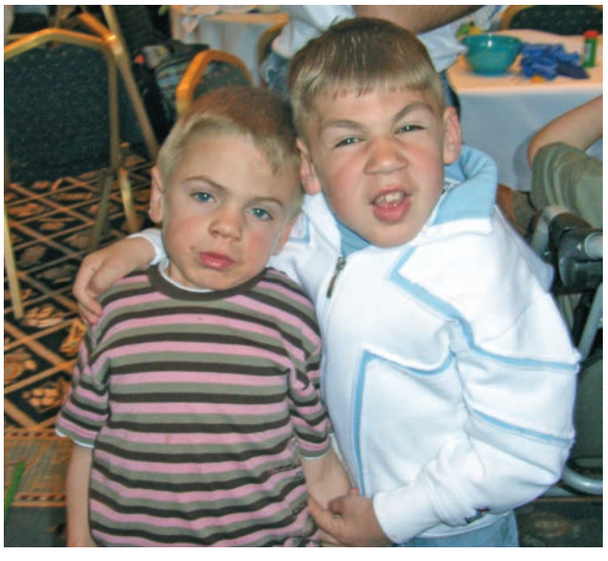
MPS AGM and Fun Weekend at Alton Towers

The MPS Fun Weekend at Alton Towers took place at the end of April 2006, taking advantage of the bank holiday weekend. Many families attended and enjoyed a fantastic weekend at the Splash Landings Hotel and Cariba Creek Waterpark followed by a day at Alton Towers theme park itself for those who didn't want the fun to stop!

The hotel and waterpark were great places to splash around and have fun, and many of you arrived early on the Saturday to do just that! The MPS AGM 2006 took place on Saturday evening, followed by the Gala Dinner. Meanwhile, the children were entertained by a merry band of childcare volunteers who oversaw all the games and activities. After the dinner finished, the temporary walls were drawn back and children and adults mingled for the Family Disco.

Once again, this weekend was a great success and we were pleased that so many families could join us and meet up with friends, new and old alike. We do hope you enjoyed yourselves!

On these two pages are a selection of photographs from the weekend.



Hi!
Just a note to say thank you all for the hard work that went into arranging the Alton Towers weekend.
We had a fantastic time.

Love from
Sue, Emma and Mark Jenkins

MPS VI Expert Meeting and Launch of MPS House

On 12 April 2006 the Society hosted an Expert Meeting on MPS VI, Maroteaux Lamy Disease followed by a Reception to jointly celebrate the launch of Naglazyme, the Enzyme Replacement Therapy for MPS VI, and the opening of MPS House. We were delighted that so many professionals and MPS VI families could join us on this very special occasion. We extend our great appreciation to BioMarin for sponsoring the day, to the BioMarin representatives who took part in the Expert Meeting and to Dr Emil Kakkis for unveiling the MPS plaque.

MPS VI Expert Meeting by Sophie Denham

On 12 April 2006, I was fortunate to be able to attend the MPS VI Expert Meeting held at our very own MPS house. For the best part of the day I was able to listen and learn about the new developments for MPS VI and the breaking news that ERT had been agreed. However, it has to be said that the majority of my day was worrying about the presentation I had to give in the afternoon. For some, public speaking comes naturally but for me unfortunately I go a dark shade of red that starts off as random blotches then slowly joins together (a bit like a dot to dot picture that you then colour in).

The morning was more scientific with talks from Dr Ed Wraith, who gave an overview of MPS VI and Dr Emil Kakkis from BioMarin who gave a summary of the clinical trial undertaken for MPS VI. After a hearty lunch (for some anyway) we carried on with the programme for the day which consisted of talks from the clinical side looking at infusion management, information on NSCAG guidelines, the support provided by the MPS Society (aaaargh, my bit) and treatment expectations, a very heartfelt presentation from a family's perspective.

The conference was a great success as was the reception and official opening of MPS House that followed.

We would like to extend our thanks to all those who helped make the conference the success it was, particularly to all the speakers and BioMarin who funded the Expert Meeting and who produced the enzyme for MPS VI. Thank you!

Photos clockwise from top right: Bill Aliski and David Boothe from BioMarin with Christine Lavery; Emil Kakkis and Christine unveiling the plaque; Mr and Mrs Kynaston, Mr and Mrs Coney and Mr and Mrs Moody (MPS VI member families); Christine Lavery presenting Julie Kelly and Sheila Bone with bouquets during her speech at the MPS Reception.

Neil and Kim Coney tell us

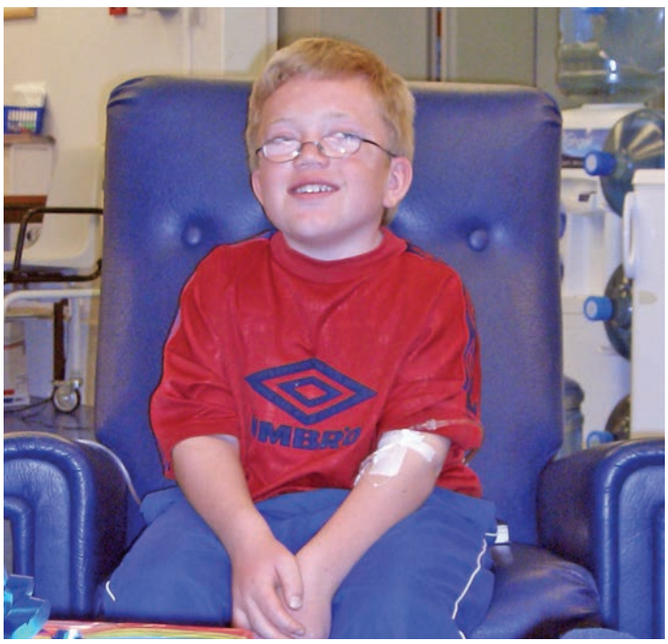
about ERT for their son, Thomas (MPS VI)

Thomas is a ten year old with MPS VI. He has received three treatments to date and all seems well with no adverse reactions. We are gradually getting into a routine of travelling to Manchester Children's Hospital, the early morning starts and long days, but pleased to be travelling in the summer months.

The first morning of ERT seemed like any other trip to Manchester for Thomas's routine check ups. It was difficult to believe that this day had finally come at last. It seemed such a long time since Thomas was trying to get onto the ERT trials and was too healthy, hence the reason why he didn't get on them. Added to this, the expected dates that the drug would finally be given approval had kept getting pushed back.

It was a 5:30am wake up call for a 6:30am drive to the hospital. We slapped on Thomas's "magic cream" ready for the needle through which he would receive his cannula later on. We arrived in plenty of time, and Jane and Sarah were there to meet us. The Moody family were also there, with their son Oliver. Thomas received his cannula through which he would be infused Naglazyme. We were feeling quite nervous but until we were able to see the drug being infused into Thomas's body we couldn't, and actually dared not, think that this day the time had finally arrived, and at that point the drug was with the pharmacist being concocted.

Once the drug had arrived at the Willink Unit in Manchester, the infusion machines were set up and that all-important moment arrived as the nurses linked the drug line to the cannula and the infusion started. This was an emotional moment for us all and a lump came to my throat. We relaxed, sat back and let the infusion work its four hour course.



This means such a lot to the whole family. This treatment opens a door for us to think more about Thomas's future than the inevitable decline in his condition that we felt before. We were always optimistic but this has increased our optimism. We have heard of testament to such great improvements in MPS I sufferers who have undergone treatment.

What does Thomas hope for? "I hope that the treatment will help my knee and ankle joints not to ache and my belly to go in."

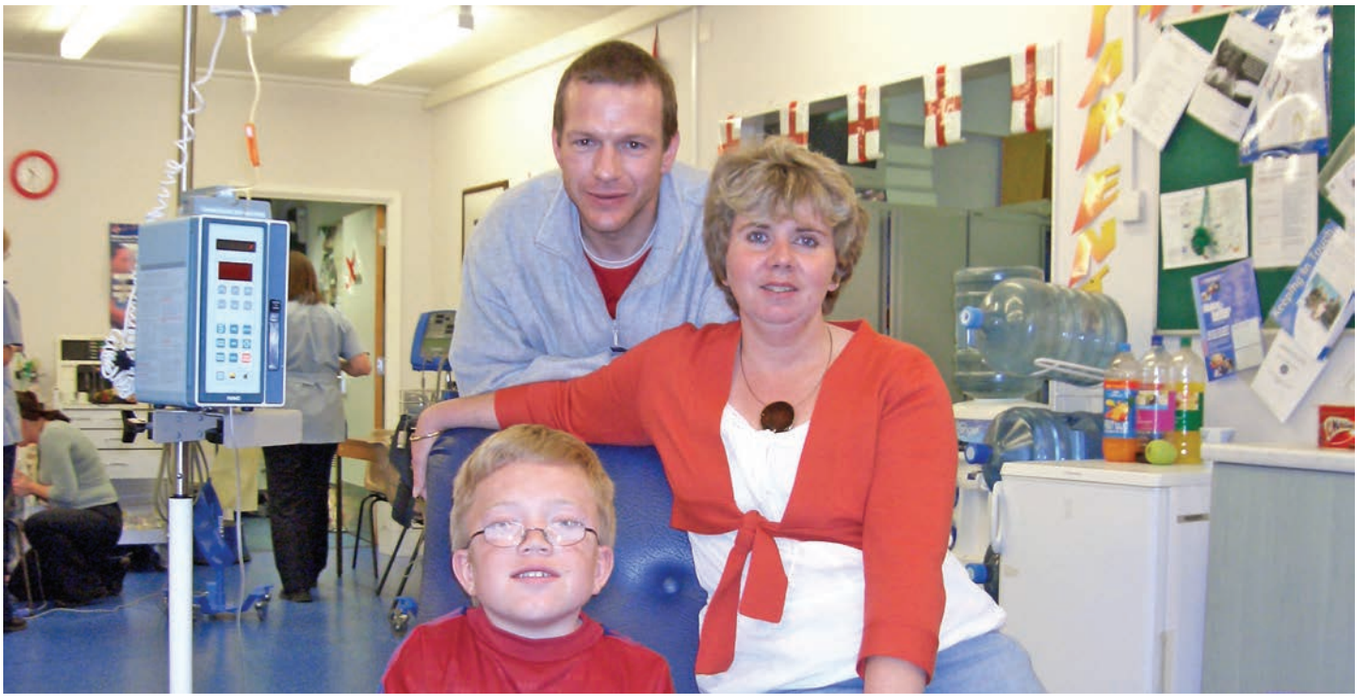


Photo top right: Thomas Coney (MPS VI); Neil and Kim Coney and Thomas

Expectations for the Future

This is a transcript of Paul Moody's presentation for the MPS VI Expert Meeting.

Good afternoon, I am Paul Moody. Dawn and I are the parents of Oliver, who is now eleven years old and was diagnosed with MPS VI at the age of five. The purpose of this presentation is to concentrate on the future, not the past, but just to set the scene, I feel I should give you a very brief update on how we reached this stage.

The first person to recognise Oliver's symptoms was actually a radiographer who alerted the specialist because she thought she recalled seeing the symptoms somewhere before. But this was only after Oliver had undergone many weeks of tests and consultations without the condition being identified. However, this time the consultant summoned us at short notice saying he had news for us.

We arrived at the hospital the next day, not knowing that it was to be the worst day of our lives, and were immediately taken to a small room where the specialist wrote a very long word on the white board, abbreviated it to MPS, and within a few minutes our whole world collapsed beneath us.

The specialist, who by his own admittance knew very little about MPS, explained that he believed it was a very serious and fatal storage disorder, and told us we may have 5 - 10 years. That was our lowest moment!

But, we have come a long way since then... Christine at the MPS Society was the very first person we spoke to who knew about MPS and she was a great help and comfort. Christine mentioned Dr Wraith's name, and to cut a long story short, within 48 hours we were sat in Dr Wraith's office. From that time on, things began to get better!

Ed Wraith diagnosed Oliver as having MPS VI the moment he saw him, and the blood tests later confirmed it. But Ed gradually educated us on the symptoms, on its progression, and most importantly of all - about the work being carried out by Biomarin on possible ERT treatment. All of a sudden, we had something back which we had lost, and that was hope!

And now to the present... It is at this stage that I would like to change the heading of this presentation, from 'what do we expect' to 'what do we hope'.

We have learned not to expect anything, because we have spent the last seven years on an emotional Roller Coaster. Every twinge that Oliver has, every headache, every minor health issue causes us to fear the worst, and yet those same symptoms in his brother and sister will simply be dismissed as growing pains!.. and so, hope has become a very big word in our lives.

At this moment Oliver is waiting to go onto the ERT and so we still hope…
That his MPS symptoms will remain as mild as possible
That the progression of MPS will be as slow as possible
That Oliver will receive the treatment in time to benefit him

If he does receive the ERT soon, then our hopes increase, we hope...
That to a certain extent, Oliver will receive his life back - that no longer can his lifespan be determined by those that have had the disease before him
That the ERT will prolong Oliver's life and improve his quality of life
That the progression of MPS will be slowed down, in fact in some aspects, we hope it may stop the progression completely
That Oliver can remain mobile
That Oliver can keep his vision, even if this means waiting for further developments in ERT

We still know very little of the effects of prolonged usage of the drug but we hope Oliver may still have a relatively normal life, for as long as possible.

Maybe the ERT is not a miracle cure, we don't know, but to us it is almost that! It will change our whole outlook on life, and enable us to start living again, and to do less worrying, which should improve our own health too!

Those who know Oliver will know what an incredibly happy young man he is, always smiling, laughing, and being positive. In the top stream at school, just qualified to attend an exclusive education academy, living life to the full. Small in stature but big in personality. He does, however, have an annoying habit of always having the last word!

It is we, the parents who worry, and it is we the parents who will be crying with joy, the moment we see the enzyme dripping into his body!

We are cautious however that prolonging Oliver's life may bring new problems, but we hope that they can be challenged and may be overcome in the fullness of time.

So, we would like to take this opportunity to thank Biomarin for giving us hope, because to parents of an MPS child, hope is everything! And yes, I know Biomarin is a business and money has to be made, but what a wonderful business to have - saving children's lives!

Thinking back to the specialist who gave us 5-10 years, we are now midway between the 5 and 10 years, but, rather than preparing for the worst, we are actually looking forward with optimism and hope! Believe me, that's a wonderful feeling!

So, thank you to Christine and the MPS Society, thank you to Dr Wraith and thank you to Biomarin. As usual Oliver was determined to have the last word, but because he couldn't be here, he insisted on sending a photographic message to you all….Thank you!

Dad's thoughts...by Paul Moody

The transcript above is from a five-minute presentation I was asked to give at the recent launch of Naglazyme at the new MPS Society headquarters, in the company of many medical professionals and representatives from Biomarin in USA.

Bearing in mind that I often make presentations for my work and am quite happy speaking in front of people, I could not understand why I felt so nervous about this one.

Maybe it was because I was not familiar with speaking about something so personal; maybe it was because I was emotionally involved. But the moment I stepped forward and glanced at the pictures of Oliver being displayed on the screen behind me, then my confidence disappeared and my voice began to tremble!

Maybe a little frustration was also creeping in, because although we were celebrating the launch of the drug that could dramatically improve and extend my son's life, he still wasn't on it, nor had I any idea when he would be on it.

Later in the day we met with other MPS VI parents who also had no idea when it would happen. In fact, we had heard that much red tape still existed that could further delay the whole process.

I confess to driving home that night feeling a little depressed, (not an unusual feeling for an MPS parent) but I was unaware of the dramatic events that were to follow:

The very next day, Dr Wraith informed me that the treatment was imminent and that the red tape had been 'sorted'. They were in fact, now trying to establish a date to start the infusions! We were delighted! (Another example of that emotional roller coaster).

After a few discussions and delays, the date was actually set for 17 May, and when we received some Emla cream in the post from Jayne at the Willink, we knew at last we could celebrate, and we did!

When we sobered up, some time later, (please bear in mind, we had been planning that party for seven years!) the wonderful news really started to sink in. So we had more parties and more celebrations!

When the 17th came, it was almost too much to take in. We had tried to imagine that day for so long. Every New Year's Eve since 1999 we had wished each other a Happy New Year and said "lets hope it's this year", but it never was!

But suddenly we were driving to the Willink. It all seemed a blur….the welcome by Jayne and Dr Wraith, the insertion of the delivery tube; the arrival of the drug, the countdown, the pressing of the button… and at last, the enzyme was dripping into his body! It was all too much to take in; we cannot ever describe that moment.

All we could do for four hours was watch each drip going into his body and thank God that it was happening at last!

Oliver's thoughts...

My first day of treatment was on 17th May. When we were travelling to Manchester, I felt happy, and surprisingly not nervous!

When we arrived, we met Thomas who is 10 years old and was also going to start the treatment; everyone was excited and taking pictures of us.

Jayne put the needle in my arm, but I didn't feel a thing at all, then they connected it to a tube and held it on with a plaster.

We both sat down and everyone started to count down…Five, four, three, two, one…and they pressed the button on the drip. I saw a tear come from my Mum's eye, but it was quickly wiped away!

We then had to wait four hours for the drip to work so to pass the time we played on a play station and read magazines.

My teacher had given me a brown envelope when I left school the night before, which said 'homework to do in the hospital, open when you get there' but when I opened it, it was a football magazine sent with best wishes from my teachers! So I read this magazine!

On the second week of treatment we met a new person starting on the treatment, another Thomas age 3.

I am now having my fourth treatment, but we think we might be noticing two slight changes already! My tummy seems to have shrunk a little, and my hair seems softer! I am hoping for more changes, but they could be a while away.

My hopes are to grow taller, to be able to lift my arms higher, to straighten my legs and that I may be able to exercise more without getting really stiff! I also have problems with my wrists, which I hope we can make better.

I want to thank everyone at the MPS Society, and at the Willink for giving me this treatment.

Scottish Family Gathering and MPS Conference

Scottish Family Gathering

By Neisha Hall

Thursday 15 June 2006 saw myself, Sophie and Ashley flying up to Edinburgh for the start of the Scottish Conference. We had arranged for a Family Gathering at the Hilton Hotel at the airport where the conference was to take place the following day.

In total nine families attended the meal, which we all commented on how fantastic the food was. The ladies even managed to keep the men away from the bar where the England football game was being televised! All in all an enjoyable evening was had by all, and both the children and adults were kept well entertained with the Quizzes, Word Searches and Colouring. We would like to thank all the staff at the Hilton Hotel for accommodating us so well.

Children's Entertainment

By Ashley Siberini

The Scottish MPS conference in Edinburgh at the Hilton Hotel was more productive than most people in attendance may think. While adults were busy debating the more intricate details that such conferences thrive on, a far more light-hearted and artistic venture was in full swing just a few small steps around the corridor.

The MPS Society enlisted the help of the appropriately named "Crafty Sharon" No, she wasn't a scoundrel playing tricks on people, she was in fact equipped with a large box of art and craft goodies. The children in attendance had the chance to paint a plate that they could take away and use.

Sharon also had a large supply of modelling clay and dozens of designs which the children could use to make ornaments. Not limited to just those skills, Sharon had an array of plain bags which the children decorated using various shiny and textured materials.

The Hilton Hotel had a special room for us to ply our craft in. On arrival to the Hamley's room the children uncovered a box full of unused and unopened toys. From toy cars to crayons, and from a football game to an odd looking Thomas the tank Engine wendy house, every toy was used to its full potential.

Around the back of the Hilton, outside the swimming pool area was a climbing frame with soft wood-chip floor. This gave the kids a chance to let off a little steam in safety and enjoy some fresh air after a delicious lunch. Taking advantage of the television and DVD player there was also a screening of the most recent Harry Potter film. The audience were supplied with some afternoon sweets and drinks and relaxed before the adults returned from all the big serious talking next door to take them home. This side of the conference was certainly a lot of fun.

The MPS Society would like to thank Crafty Sharon and our volunteers Helena and Kate for helping out with the children on the day.

Scottish MPS Conference by Sophie Denham

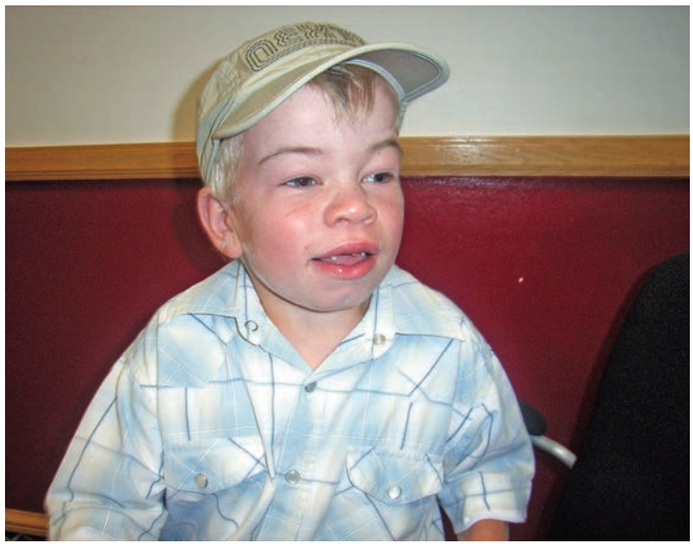
On 16 June 2006, the MPS Society hosted another Scottish MPS conference, the last one having taken place in 2003. This conference was held at the Hilton Hotel Edinburgh. It afforded us the comfort and space we needed and, luckily for us, they already knew how our conferences run and what to expect as their current manager used to manage the Northampton Hilton Hotel. The conference was well attended and had a good balance of professionals and families. Dr Peter Robinson chaired the meeting and we also had Doctors from Manchester Children's Hospital and Great Ormond Street Hospital.

In the morning Dr Cleary gave an overview of MPS diseases, focusing on the clinical management of these diseases. This was followed by Dr Wraith who spoke about Fabry disease and the management and treatment available for this condition. Dr Brad Williamson then spoke about the management of the spine and how procedures have changed during his time as a surgeon and his preferred method of surgery at present.

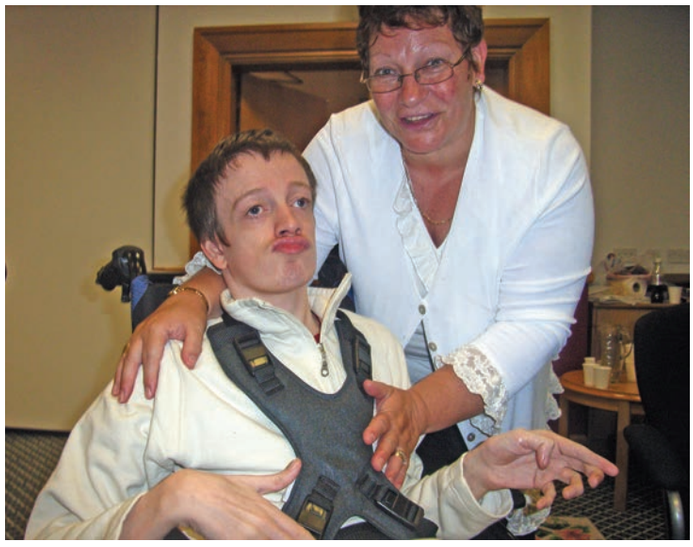
After coffee, Dr Fitzpatrick spoke on working with rare diseases in general medicine, emphasised the need for everyone to work in partnership and recognised that the parents were the experts in many of these diseases. Then it was my turn to talk about the environmental and practical approaches to behaviour management in MPS III. After quite a full morning we were all ready for lunch. This was a real treat, especially the home-made puddings which were extremely naughty but very nice.

Margaret Robertson opened the afternoon with a heartfelt presentation on the role of Robin House and it was great to see how the hospice was set up and what it is able to offer the families of Scotland. Following on from this, Dr Wraith shared information on current and future therapies, before Jennifer Johnston gave an inspirational talk on life after ERT. This was emotional for many, especially given the difficulties in Scotland for many who are being denied treatment. This was highlighted in Dr Robinson's talk and the panel discussion afterwards, where a number of families raised their concerns that treatment was being denied for some in Scotland and the unfairness of this. All the professionals recognised this and emphasised the need to work together, and not against one another, in challenging the Scottish government.

I also spoke about the Society's position and confirmed that we were continuing to challenge Scottish government but that we needed families to be the driving force behind it as we cannot do anything if we do not have the support of the Scottish families. The conference went very well, even though it ended on a very emotional note.Our thanks go to all the doctors and professionals who gave their time to speak at the conference and to also thank so many people for attending this meeting.



Jack Ferguson (MPS II)



John Paul O'Neil (MPS III)

South West Family Get-together

By Sophie Denham

On 21 May 2006, four families from the South West got together for Sunday Lunch. We met at the Cedar House Hotel in Westbury, Wiltshire, where after drinks in the bar we moved to the dining area for lunch and general chat.

We had planned to have a big party with children's entertainment at the local village hall but due to the small numbers this was not viable. However, the lunch was lovely and the families who attended appeared to have a good time and enjoyed catching up with old friends.

Fabry Conference by Sophie Denham


Sophie

On 25 April 2006, Cheryl and I attended the Fabry patient meeting held at the Royal Free. This meeting is run jointly with Addenbrookes in Cambridge and it was the Royal Free's turn to host it this year.

This is the second Fabry patient meeting I have attended and it was once again extremely informative and good to catch up with all the recent developments as well as meeting not only the professionals but also the individuals and families, some of whom I have met in the past but some who I met for the first time that day. Cheryl also found the conference to be extremely informative for future research she may be doing and it also gave her the opportunity to network with some of the leading specialists in the field of Fabry disease.

As some of you may know from attending the conference, I personally had to stand up and share with the audience the role of the MPS Society and how we as a support group provide support to our members. As always I was extremely nervous as public speaking is something new to me and I would like to thank all those who offered me their kind words and support.

I would like to thank the Royal Free Hospital and Addenbrookes, Cambridge for a very informative and well-structured meeting.

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.

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

Serendipity by Bernie Drayne

With so many cheap flights now available it was very hard not resist the temptation to take a short break in Rome at Easter. I really detest flying, but an offer came up which we could not refuse especially as our daughter Roma who has read and reread all the Dan Brown books was exerting pressure to visit the lovely city which bears her name. As it was Easter week, the city was buzzing with pilgrims and the atmosphere was alive. The five of us including Kevin, Roma's Dad (another Dan Brown fanatic) and Kilian and Peter really enjoyed touring the ancient sites although access was a problem with so many steps everywhere. The boys got to see Lazio v Roma football match and I thoroughly enjoyed eating out in the restaurants so we were all catered for.

However, the highlight of our trip happened by complete surprise, unplanned and purely by chance. One morning we were in St Peter's Square waiting to join the queue to view the Vatican when Kevin fell into conversation with a young man called Dony from Dublin. Dony had been working as an Irish sculptor in the Vatican area and he was with his friend who also a wheelchair user. Dony knew his way around the Vatican and was able to show us all the shortcuts and how to avoid queues. He accompanied us to the Sistine chapel and explained everything to us, he was so knowledgeable about everything and his enthusiasm was infectious.

Dony suggested to us that he could arrange for Roma to have a private audience with the Pope and to our amazement he spoke to a Swiss guard who produced a pass for myself and Roma to attend an audience the next day, known as Spy Wednesday. It was all quite surreal the way it happened and we were really unprepared for this.

On Wednesday Roma and I went through the security procedures and we were led up onto the open air podium in St Peter's Square to sit in a queue with the other twenty or so wheelchair users. The square was full of pilgrims all cheering as Pope Benedict was driven around in his open air Pope mobile waving to everyone.

The mobile was driven up onto the podium, and the Pope made his way to his throne from there he addressed the crowd in several languages who cheered wildly when their language was spoken. We were surrounded by papal dignitaries and security people who carefully watched the crowd. After about half an hour the security people came over and told us we were to be the first to be presented to the pope. I began to feel slightly nervous, just what do you say to the Pope? Roma was very laid back as usual and really enjoyed everything.

When we were presented to him, he shook my hand and he placed his hand on Roma's head and hands and blessed her Rosary beads. To this day neither Roma nor I know what he said to us, or what we said to him, it was all so unbelievable. As we were led away from him we were so bemused but absolutely thrilled. Roma promised never to wash her hand again which he had touched - and it looks like she's sticking to that.

It was certainly the highlight of our trip to Rome, so unforgettable and it made us feel really special. And to think it all happened by a chance meeting with a young man called Dony who we never met again.

Photo appears courtesy of the Photographic Service, L'Osservatore Romano

New 0845 telephone number

The MPS Society telephone number is 0845 389 9901. This number has been introduced as a benefit to MPS Society members as call costs will be charged to the caller at 'local' rates. Please remember to use this number when contacting us.

Your letters

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

Aiden's story by Angela Brown

When Aiden was born he was kept in hospital for an extra 24 hours to be treated for what was thought to be jaundice. After a short time under a heated lamp we were allowed to go home. At three months old he was admitted to hospital for a hernia operation and then at four months they found that he had an enlarged liver and spleen. Aiden continued to be a very sickly baby for no apparent reason. His development did not progress at all and he did not appear to be able to see anything. He also had a large curve in his spine.

Aiden was finally diagnosed with Hurler disease when he was ten months old. When I was told that Hurler disease was genetic, a double-decker bus could have hit me lighter. I always new that there was something wrong with my wee boy, but not this. I felt cheated that I had never heard of Hurler disease. I could not pronounce Mucopolysaccharide, never mind understand it. I kept looking at Aiden, who was rolling around on the floor playing with a car, thinking that the doctor must have got it wrong and was talking about another Aiden. It could not be my baby that was going to die. I thought Aiden just had a few wee problems which they could fix.

Although there is no cure for Hurler disease, if the condition is diagnosed before too much damage has been caused, and a suitable donor is found, a bone marrow transplant (BMT) can be an option. For some children, a BMT can alter the course of their disease, with the donated cells providing a source of the missing enzyme.

We were told that BMT was an option which would help to improve Aiden's quality of life, but were also told that there was a one in 20 chance of losing him in the transplant process. Although we knew the risks, we didn't really have a choice. If we found a match within the family, we had to go ahead.

Aiden was one of the lucky ones because his brother was found to be a perfect match. After being checked from top to toe, Aiden underwent his transplant when he was 14 months old and Aaron was three years old.

After the transplant, we brought a new baby home. He had a new layer of baby-soft skin, new nails, his blond hair had fallen out and it was coming back jet black and the clouding of the corneas of his eyes had cleared. He had learned how to sit up in hospital and his hearing had improved – he was now making noises. It was like a miracle. We felt for the first time since Aiden was born that we had a future for our son.

No-one knows what Aiden's prognosis is now, because of the progressive nature of the disease, every child who has had a transplant is different. Aiden still has major problems with his bones and has developmental delay by around two years, but he is catching up quickly. He also has a leaking valve in heart and major problems with his eyes. He had a spinal fusion when he was three which was the start of a lifetime of corrective surgery.

Aiden just shines now. He has a wicked sense of humour and a very infectious laugh. We enjoy what we have and never take for granted today what we might not have tomorrow.

Sam's surprise makeover! by Vicki Brockie

Sam had a makeover for her 15th birthday present. It was a total surprise for her as I didn't tell her until the morning we were going! Sam had her hair and make up done and really enjoyed the pampering.

Sam is doing really well on the enzyme replacement therapy at Birmingham children's hospital every Friday. I'm very lucky as my Dad does most of the trips (thanks Dad!) and Sam prefers him to take her as she gets spoilt.

She is now in her last year at school and doing a work placement in the local hotel for two weeks in July, then she is off to Dorset for three days with school. I can't keep up with her! Despite missing a lot of school her determination has the school predicting good GCSE results.

Enzyme replacement has made such a difference to Sam, who has a very positive future along with her laid back and positive attitude. Nothing gets my girl down!

MPS Support to John and Joanne Allen

Bethany was diagnosed with Sanfilippo disease on 15 December 1999. We were told this was a terminal disease and life expectancy was around 14 years of age, she would become more physically and mentally disabled throughout the course of her life. Of course we were devastated. The only help offered at that time was from the MPS Society. Jo was finding it hard to cope and was prescribed anti-depressants by our G.P. After three years John had a major break down, couldn't work or control his emotions, had panic attacks, and also prescribed anti-depressants.

It was at this point the MPS Society was involved again, they asked us who our social worker was, and we said we hadn't got one. The MPS Society made enquiries and we were assigned a social worker who was brilliant and immediately set about the task of supporting us as a family.

Unfortunately she retired and we were back to square one , left in the dark not knowing who our social worker was and no-one contacted us.

We had a parents' meeting at Bethany's school where Joanne broke down crying and we explained how we were struggling as a family. Bethany's teacher suggested direct payments as a way of helping us. We started the procedure and had a meeting with social services. We were asked to come to Tamworth council offices and bring Bethany with us which was very distressing for her; she was crying and very upset. The social worker suggested John took Bethany away for a while. The meeting, an (ASSESSMENT) of Bethany's and the family's needs, was held with Jo, Jasmine and Tom. They were told within a couple of weeks how many hours would be allocated and from that moment we were never contacted. A few months later we tried to chase it up, was given the name of our new social worker and she would call me back. The school tried to find out more, they were told someone will call us but no-one ever did. The school were told it was all in hand. Someone else who was also chasing for us was told all direct payments were on hold until June due to training, but still no one had contacted us.

The MPS Society wrote a letter asking the question why no -one had responded to us. Then out of the blue we got a call from our new social worker asking if she could come and meet us, which she did. She told us she would be our social worker for the next five to six weeks until direct payments were sorted. Other issues we had were not worth getting involved with because we would be passed on to the long term team. We were told by someone else that it would be the disability team but when this was mentioned to our new social worker she said Bethany would never make the criteria. This proved to be the wrong information again as Bethany is now with the disability team.

During the whole time Bethany was never fully assessed and the social worker spent very little time with her. The social worker turned up to meetings without relevant paperwork so things could not proceed, and when the application was made it wasn't what we had requested. When this was pointed out we were then told she may have to be re-assessed. I was not happy as this was now around nine months after first meeting.

Bethany was first offered 24 hours a month because the social worker had requested an overnight stay which we had never asked for. We always said we wanted flexible hours which is what attracted us to direct payments. Now she has been given 13 hours a month, which is a lot less.

The social worker then paid us a visit and told us that we were now holding up the procedure because we hadn't opened the bank account. Considering it had taken the best part of a year for them to get to this stage we felt insulted and angry that we were now to blame for its hold up. IMMEDIATELY after the meeting we opened the account and left the details as asked by the social worker. We never heard from her again.

Some months later we were then assigned another social worker. She said she was having trouble retrieving Bethany's file but had seen that no-one had contacted us and so felt she should. We explained the trouble we were having obtaining direct payments and the poor support we were getting. She was very apologetic and vowed to put it right.

She had a meeting with our previous social worker before seeing us later that day. Our new social worker asked why direct payments had not started. The old social worker said she had not had the bank account details, yet that same morning I had a letter saying direct payments money had gone into Bethany's account. So, again, a lie/excuse was given for the delay.

The whole thing had taken 12 months and the treatment and support given to us has been nothing less than appalling, very un-professional and a total let-down. All support was dropped before the disability team eventually took over.

Since being assigned our current social worker we have no complaints at all. She tries all lines of enquiry and doesn't give in, she keeps us well informed verbally and in writing and is very professional and supportive.

We were then offered a meeting with the Area Manager (East Staffordshire and Tamworth) Social Services, which took place at our house. The Area Manager apologised profusely and admitted full responsibility for everything that had happened and the inappropriate treatment we had experienced.

Thanks to the MPS Society and our newfound friend, Neisha the ROTTWIELER, this fight has now reached conclusion with full apology and extra help being offered this summer school holiday. All I can say is a BIG THANK YOU to all at the MPS Society, and a message to all you parents - hang in there, don't let these people like social services and many others grind you down. Do it for your children as they deserve it. Also, don't be afraid to ask for help, the added stress these organisations create passing you from one place to the next is not needed and you will find as I did when the MPS Society get involved it took them one phone call to get an answer. This is what's wrong and frustrating. So get yourselves a ROTTWIELER and fight back.

Thank you MPS Society for your support.

A heart-stopping tale

by Robert Kenton - a person with Anderson-Fabry disease

Recently completing my first swimming session for three months I feel a goal has now been reached, one that I've had doubts about for some time and which, having been achieved, has given me the confidence to finally put pen to paper.

Health problems can vary greatly for Anderson-Fabry patients and one area where there can be a variation is in the heart; valve malfunctions have been found in some cases, although it is not clear whether this is due to the disease or not, as statistics show that about 1 in every 20 people may have a slightly deformed mitral valve. This condition can often remain undetected if it does not cause any problems but can promote tiredness and shortness of breath; as many readers may be aware, proper function of the valves is important for efficient transfer of blood within the heart chambers and circulation around the body.

The phrase 'prolapsed mitral valve' meant little to me in real terms, although I knew it was something that I had been diagnosed as having when I was found to have Anderson-Fabry disease, and could have been inherited. Until I was told quite unexpectedly that the mild leakage of the heart valve had progressed and needed attention. Not something to be thought of as a future event any more but a problem to be dealt with now.

I hadn't been feeling any different physically when I was given the information, at one of my routine six-monthly check-ups courtesy of the Royal Free LSD Unit, and found it difficult to come to terms with the facts; I either needed to have some serious heart surgery pretty soon or, if I left things as they were, there was the possibility of a less successful operation in the future.

I was given some respite while further tests were carried out; an angiogram to check that my coronary arteries did not require attention (at the same time) and a trans-oesophageal echocardiogram, which examined the valve from inside the chest and gave the consultants a better view of the problem.

This gave me time to accept the situation and the options open to me; my health was currently good and I was advised by the Anderson-Fabry and Heart Hospital teams that it would be better to deal with it now before the heart function was further impaired.

I decided I wanted the operation to go ahead, so I began to collate material to help me understand the information I was being given. Of great help here were the leaflets and website provided by the British Heart Foundation which provide details of what is involved from very much a patients viewpoint. It was good to read that my feelings of doubt and unrest were natural and how these thoughts might develop as time and the situation progressed.

Heart surgery has advanced an awful lot in recent years; it is still major surgery for each individual and great skill is required from the surgeons, but so many operations are now carried out that much of it is considered to be routine.

I was told about the results of the tests; a repair to the valve looked difficult but was preferred to replacement if possible. This is because when a replacement is fitted, it may have a limited lifespan and the patient will be required to take an anti-coagulant drug which needs careful monitoring, for the rest of their life. I was sent to see a specialist in that area of cardiothoracics; the repair would be attempted with replacement as a possibility and, although I felt confident that everything would be done to try and achieve the former, I needed to understand the outcome of either, and be able to accept whatever happened.

During these months of investigation, I developed symptoms which could have been due to the poor functioning of the valve, the situation I was in or something else not connected to either; it has remained something of a mystery but I felt very unwell at times and had scans which revealed a foreign mass inside my lungs, at one stage threatening my suitability for the operation. I knew it was important to keep as fit as possible prior to the operation and found it frustrating and stressful to be plagued by this problem.

I had a Bronchoscopy (a tube via the mouth down to the lungs to take a biopsy) to try and diagnose the unknown ailment. The area could not be reached but the procedure may have helped in some way because I was feeling a lot better almost immediately after it; the following scan showed that the mass had dispersed leaving me, and everyone involved in my treatment, feeling very much relieved. I was told that the operation could go ahead.

My appointment date arrived in the post; I was pleased to find that I only had a short time to wait and was soon attending the pre-operation day. A final ECG and x-ray were taken here, and at a group session with others about to have heart surgery I was given more details of the operation and invited to ask any questions. It was an informative but somewhat exhausting day that left me feeling emotional and drained but relieved that I had been passed fit for my operation in a week.

In many ways, having Anderson-Fabry disease helped me cope with the situation; I'd had to think about my health most of my life and deal with things as they occurred. I'd been a volunteer for the enzyme replacement therapy trials involving biopsies and other clinical procedures so the operating theatre would not be a new experience. And I must mention all the encouragement and helpful advice I received from everyone at the Royal Free and Heart hospitals which gave me extra confidence and optimism.

The final week passed and I had my last swim, wondering when I would be back at the baths next (I told myself I would be back); I also called in at a local convalescent home with a view to spending some time there post-operation.

I spent the night before my operation at the hospital feeling more relaxed than I had for some time. I was fortunate to be in the cardiothoracic Atkinson- Morley wing at St. George's hospital in London; very clean and modern and full of friendly patients in various stages of heart surgery who wished me well.

I had brought with me an Anderson-Fabry publication provided by the Royal Free LSD Unit and this proved to be a valuable asset which created interest and awareness of my condition to some of the hospital staff; in addition it gave me confidence to know that they had taken the information from it into account before going ahead with the operation.

The outcome of my operation was still not certain but whatever, it was out of my hands now, I was as fit as I could be and I felt confident I had made the right decision.

The general operation procedure is that, under general anaesthetic, the blood vessels entering and leaving the heart are connected by tubes to a Heart-Lung machine. The heart is cooled and paralysed to stop it beating so that the work can commence, and blood is diverted to the Heart-Lung machine which oxygenates it, removes carbon dioxide and returns it to the body. Afterwards the heart is restarted with an electric shock and the circulation is restored.

It made me smile to see a recent television re-creation of a heart surgery operation where the patient was conversing with their family within the hour; in reality, about 24 hours will be spent in the Intensive Care Unit, most or all of which the patient will not remember. This was the case with me but I did feel quite comfortable once I was back on the ward and the news that the repair had been successful gave me a big lift and a lot less to be concerned about.

The first couple of days were not very comfortable; there was oxygen and a heart pacing machine, chest drains and a catheter attached to me, all making movement and sleep difficult, but looking back these were only minor irritants which every patient was dealing with and the important issue was that I was progressing.

Friends and family visiting were surprised at my progress and I was soon moving around the ward; I found this incredibly exhausting at first and sometimes doubted my ability. Never had I looked at a flight of stairs with such trepidation, but things became a little easier in time and after about ten days they were satisfied that I was ready to leave.

The convalescent home was a worthy stepping stone to the outside world and for the next two weeks I slipped into the quiet ritual of recovery along with the other 'inmates', all of whom had their own stories to tell and were good company. There was constant supervision if required, good food, plenty of rest and a gradual increase in exercise and improvement; I found it beneficial for all these things but also it was good for taking the pressure off friends and family.

I felt very uncertain about stepping back into my life outside even though I was keen to do so after the two weeks; it was mid-winter and cold out there after the warmth and caring security of the home, I felt quite weak and vulnerable having to take responsibility for myself again.

The first weeks were not easy; I had some bad days with aches and pains I had not experienced before. I felt weak, tired and depressed at times but read that all these feelings were normal and would pass with time - and so they have, but it is a gradual process and a lot of patience is required.

Cardiac rehabilitation classes from my local hospital have been very useful and fun too. Although my diet and exercise has been carefully thought out with Anderson-Fabry disease in mind, I have learned more about these topics from a different viewpoint and been able to add to my knowledge in general. The staff are all very friendly and keen to see progress being made and I have been impressed with this.

To date, I have not had any recurrence of my pre-operation problems; my heart appears to have lost its old slow beat and developed a quicker rhythm to go with the repaired valve. Results from recent visits to the Royal Free and back at St. George's have shown that the repair is successful and working well. I hope that my progress will continue and that the hard work put in by all those involved will prove worthwhile; I will certainly be doing my best to help.

At the recent Anderson-Fabry patient meeting I had the chance to talk about my experience and, although this has undoubtedly been an important part of my life I am now beginning to think of it as a past event.

With thanks to the teams at:
Ben Weir ward, St. George's Hospital
LSD Unit, Royal Free Hospital
Rustington Convalescent Home
and
British Heart Foundation (www.bhf.co.uk)

Global Organisation for Lysosomal Diseases

New web-based LSD information from GOLD

Dr Ann Hale, Executive Director, GOLD

GOLD, the Global Organisation for Lysosomal Diseases is an international collaboration of scientists, clinicians, patient organisations and commercial organisations dedicated to improving the lives of patients and families with lysosomal storage diseases (LSD.)

GOLD has now developed two new areas of its website **(www.goldinfo.org)** where member organisations can view video presentations or get online access to Scriver's Online Metabolic and Molecular Basis of Inherited Disease. As the MPS Society is a member of GOLD, our individual members are able to access these new areas of the GOLD website. We hope you will find them very useful.

Scriver's Online Metabolic and Molecular Basis of Inherited Disease

This textbook is the "bible" of inherited metabolic disease and we are extremely fortunate to be able to access the whole section of 20 chapters on LSD, free of charge, via GOLD. The entire work is now so extensive that it can no longer be published as a multi-volume textbook. Apparently, it would have weighed 32lbs! The online version of this prestigious publication was launched by McGraw Hill Medical Publishing in November 2005. Contributing authors are renowned experts in their fields, and the chapters are regularly updated, so that information is always current.

Video Presentations

Initial presentations on this site include two scientific meetings, the National Tay Sachs and Allied Diseases Association Annual Conference, held in Alexandria, VA, USA in April 2006 and a GOLD Meeting in collaboration with the Institute of Child Health, University College London, held in London, England in May 2006.

Presentations at NTSAD's Medical and Research Update included:
"Genomics and Beyond" by Paula Gregory PhD
"Clinical Trials in Late Onset, Juvenile and Infantile Tay-Sachs", by Edwin Kolodny, MD, David Korosec, RN, Gustavo Maegawa, MD and Cynthia Tifft, MD, PhD
Overview of Current Research efforts: A summary of a NINDS worskshop "Glycosphingolipids in Health & Disease" by Bruce Bunnell PhD

At the GOLD UK meeting, Dr John Hopwood gave a Plenary Lecture "The Research Pipeline for Lysosomal Diseases" summarising the current status of research into LSD and the directions of future research and therapy. GOLD will be adding further video presentations to this site.

How to Access OMMBID for GOLD and GOLD's Video Presentations

Both these areas can be viewed from the GOLD website at www.goldinfo.org. In the menu, select "Education and Information" and then either "Video Presentations" or "Scriver's OMMBID".

You will need to register at the website, if you have not done so before - instructions are on the relevant entry pages. Registering only needs to be done once. You will be asked to give your email address as a username, choose a password and select the organisation of which you are a member, so please select MPS Society. GOLD will not give your information to third parties.

OMMBID

This is a special version of Scriver's The Online Metabolic and Molecular Bases of Inherited Disease developed for GOLD's member organisations by McGraw Hill. This allows online reading access to the section on Lysosomal Storage Disorders, free of subscription charges.

To access McGraw Hill Education's OMMBID you will need to be registered as a Member user of the GOLD website. Select "education and information" on the index of the gold website homepage www.goldinfo.org. Select "Scriver's OMMBID" from the sub menu. If you are not already a registered user, you will be prompted to register here. Log in via the entry page on the GOLD website **www.goldinfo.org**.

This will take you to a new log in page. Here, you will need to register again (the first time you use Scriver's OMMBID for GOLD), as a user for McGraw Hill. GOLD apologises for this double registration and log in, but this is because GOLD and McGraw Hill do not exchange data, and for technical and security reasons we were unable to effect entry to the McGraw Hill OMMBID database without this additional step. We hope you will not be inconvenienced by this, in order to access this very informative online publication.

Subsequently, log in with your username and password. This takes you through to the index page of section 16 "Lysosomal Storage Diseases" of OMMBID.

Click on the chapter you wish to read. Please note that the facility to read other sections, or download chapters as download as a pdf file are disabled.

How to view the video presentations

Log in using your username (email address) and password. Click the login button and you will be taken to the Video Presenter Page.

Scroll to the panels underneath the screens at the top of the page.

In the Right hand panel, select the Presentation you would like to View:

NTSAD - presentations at the National Tay Sachs and Allied Diseases Family Conference 2006 Medical and research Update session.
GOLD-UK - A lecture by Professor John Hopwood at a meeting of GOLD's UK members, 2006 "The Research pipeline for Lysosomal Storage Diseases"

Still in the right hand panel, click on the tile of the presentation you would like to view. Details will appear in the left hand panel.

Under the picture of the speaker, click on the option to play, in either Real Player or Windows Media, depending on which programme you have installed.

The Presentation will auto-load. Load time depends on the speed of your internet connection and make take a minute or two.

The slides appear in one frame, the speaker in another.

If Using Real Player:
Holding the cursor over the icon buttons displays their function.
If you wish to view just the speaker, full screen, use the "Full Screen" icon underneath the slides screen. To return to slide view, use "esc".
You can scroll forwards or back using the "next slide" icon.
The icon "View Chapter marks" underneath the slides screen switches between slides view and outline of slides view. You can scroll through the slides and double click on the one you wish to resume viewing. Double click on the slide and the presentation will re-synchronise. Return to the slide view using the "View Chapter marks" icon again.

If using Windows Media:
Holding the cursor over the icon buttons displays their function.
If you wish to view the speaker full screen, double click on the screen. (Note, the "full screen" icon does not work in Windows Media Player) To return to slide view, use "esc".
You can scroll forwards or back using the "next slide" icon.
The icon "View Chapter marks" underneath the slides screen switches between slides view and outline of slides view. You can scroll through the slides and double click on the one you wish to resume viewing. Double click on the slide and the presentation will re-synchronise. Return to the slide view using the "View Chapter marks" icon again.

If using a MAC:
You will need to use Firefox as the web browser. Safari will not support the slide presentation view, although the video can be viewed using Safari.

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The Neuronal Ceroid Lipofuscinoses, Sandra L. Hofmann / Leena Peltonen

Archives of Disease in Childhood

The Archives of Disease in Childhood is inviting children and young people to contribute their experiences of ill-health, chronic disorders and health services to their family page, Backchat. The journal goes to all paediatricians in the UK, as well as many around the world.

Submissions can be up to 800 words and be written or illustrated. You can submit your experiences online at www.archdischild.com, following the links to submit a manuscript, or by post to Dr Harvey Marcovitch, Backchat Editor, ADC, Honeysuckle House, Balscote, OX15 6JW. Anyone under 16 should obtain their parent or guardian's permission before submitting an experience.

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.cafamily.org.uk** or telephone Contact A Family on **0808 808 3555**.

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

Does your child have epilepsy?

Below are some of the things we have found out since the death of our son Oliver aged 10 who had Sanfilippo Disease and also had epilepsy and died of Sudep.

There are over 450,000 people in the UK with epilepsy, 1 in 20 people will have a seizure in their lifetime. There are 1,000 deaths a year (more than cot deaths and AIDS combined).

500 of these deaths are **SUDEP** (Sudden Unexpected Death in Epilepsy). Most of these occur in young adults who are otherwise healthy but they rarely occur in children.

Most deaths occur at night and Sudep is therefore likened to cot death. Sudep is rare in many types of epilepsy, but risks are individual. 70% of people with epilepsy can be seizure-free if treated with the appropriate drugs.

The facts above have been taken from a fact sheet produced by the charity Epilepsy Bereaved. More information can be obtained from the website **www.epilepsy.com**.

May we suggest that if you are at all concerned about the above information you should contact whoever deals with yours or your child's epilepsy to find out more?

Bernie and Eddie Hall

Bereavement and Learning Disability: Help Needed!

Although life expectancy in learning disability has been widely researched little has been done to investigate the needs of those left behind when an individual with a learning disability dies. The University of Wales, Bangor project aims to investigate what these support needs are and look at how these needs might be provided for. The project will investigate the grief experiences of families in which a child or adult with an intellectual disability has died.

The European Social Fund and Mencap Cymru are jointly funding the project. Led by Deirdre Reilly, the study will involve an interview with bereaved family members asking about their experience. Participants will also be asked to fill in some questionnaires relating to their loss. It is hoped the study will include family members from all over the UK and Ireland, which will give us an insight into how experiences differ in different areas.

If you wish to take part in the research please contact Deirdre as follows:

Deirdre Reilly
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University of Wales
Bangor Gwynedd, LL57 2AS
Wales
Tel: 01248 351151 ext. 8706
Email: d.reilly@bangor.ac.uk

Turkish MPS Family goes online

In the Winter 2005 MPS Magazine we featured an article by Nalan Cetin, the mother of Emre who has MPS II, Hunter Disease.

Nalan and her family live in Turkey and for almost 12 years they have cared for Emre despite little help and information from the outside. Over the past two years a website has been developed including a page in English, which tells more about their experiences, and hopefully will be a source of courage and inspiration for other families affected by this disease.

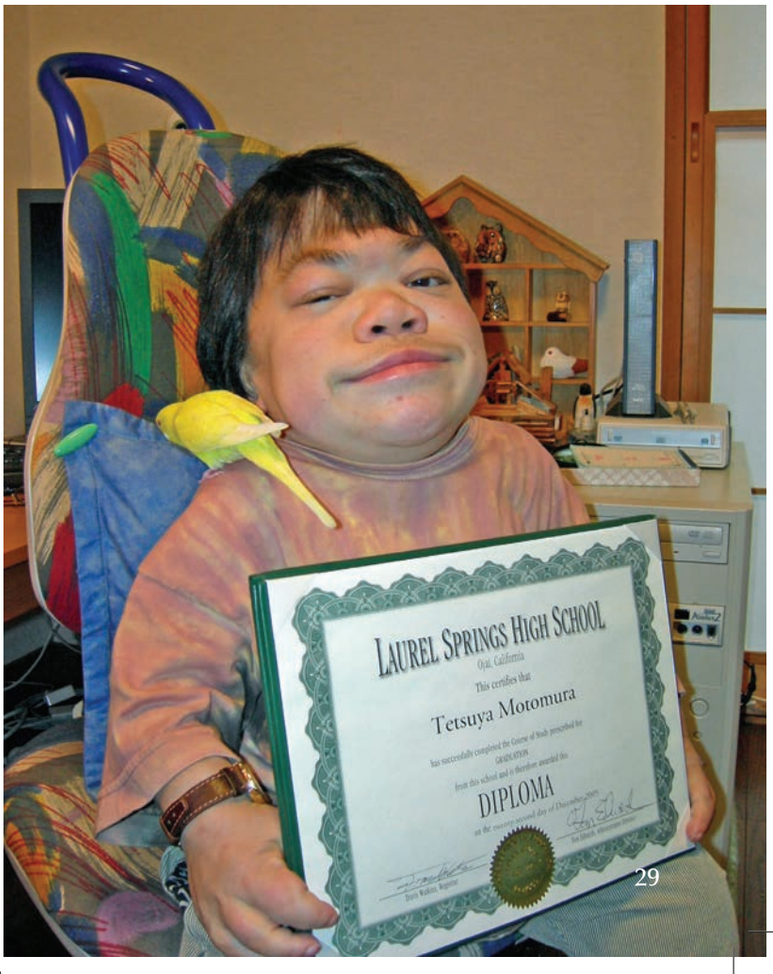
Visit **www.mpsturk.sitemynet.com** to learn more.

Tetsuya receives his High School Diploma

In the Summer 2005 MPS Magazine, Tetsuya Motomura wrote about his life, and his experience of living with Mucolipidosis III (ML III). Tetsuya gave a moving, poignant and inspiring account of the obstacles he has overcome and which have shaped his life. He wrote that life is sometimes frustrating and that he has to work a little harder to fit into Society.

So, we were absolutely delighted when Tetsuya recently told us how proud he felt to have just graduated High School. He is now moving on, and will start the next phase of his education. About two months ago Tetsuya and his family moved to a new apartment in Tokyo which is close to the station. The previous occupants had adapted the apartment to be easily accessible. Tetsuya also now has a pet budgerigar called Yellow Apollo!

Many congratulations to Tetsuya for his achievements. If you would like to read more about Tetsuya's experiences visit **www.freewebs.com/tmotom**.



Overcoming a Genetic Defect

Kiel University coordinates European research project on alpha-mannosidosis

The European Union has approved a grant of about € 2.4 million to a team of European scientists, led by the Kiel biochemist Professor Paul Saftig, to carry out research on the rare hereditary disease alpha-mannosidosis. The project, HUE-MAN (Human Enzyme Replacement Mannosidosis), will be coordinated by Professor Saftig for the next three years at the Christian-Albrechts-Universität (CAU) in Kiel. The aim is to develop a drug that will compensate the patient's body for the genetic defect.

In Europe about 400 people, mostly children, suffer from alpha-mannosidosis. It causes coarse facial features, retards the development of speech and mental ability, weakens the immune system, and damages bones and muscles. Professor Saftig explains the importance of his research: "At present, without a very risky bone marrow transplant, many of these patients die between the ages of 10 and 20". The cause of this lysosomal storage disorder is a genetic defect. Lysosomal enzymes - cell constituents that are specifically programmed to break down metabolic waste products - normally decompose the waste products sufficiently to enable the cell to continue the process. If a certain gene, which controls one of these enzymes, is defective, that enzyme cannot be produced. The waste products, which in the case of alpha-mannosidosis are mannose sugars, build up in the cell and completely block its function. As a result, not only muscle cells, but also brain cells, cease to work.

The European team of scientists are building on their previous basic research in an EU project that has been completed. In that work they discovered that the "accumulated garbage" in the cell became dispersed when diseased mice were given injections of the missing enzyme at fortnightly intervals. "It is especially new and interesting that we even succeeded in treating the damage to the animals' central nervous system", explains the Kiel biochemist. However, there is still a long way to go before an effective drug can be developed. "Nevertheless, we hope that in three years we will have progressed far enough to begin the first clinical phase", says Saftig.

In addition to the Kiel researchers, about 15 other scientists are working on the project - in particular paediatricians from Göttingen and Mainz, from Norway, Great Britain, France and the Czech Republic, as well as from a pharmaceutical company in Denmark. Each one will be responsible for a specialist area: while Saftig will coordinate the project and develop drugs for animal tests, the paediatricians will study the detailed course of the disease in patients. For example, the Göttingen scientists will investigate changes to the neighbouring genes.

The € 2.4 million funding for the project has come from the European Union's Sixth Framework Programme Life Sciences. A proportion of this programme's funding is reserved for research into rare diseases. Of the total project grant of € 2.4 million, the amount allocated to the Christian-Albrechts University of Kiel is € 600 000. For the Institute of Biochemistry this will fund three new posts: a technical assistant, a post-doctoral student, and a secretary.

More information about the project can be found at the website:
www.uni-kiel.de/Biochemie/hue-man

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Psychosocial Outcomes of Bone Marrow Transplant for MPS I Hurler Disease

by Cheryl Pitt

Since the late 1970s bone marrow transplant (BMT) has been used to treat MPS I Hurler disease. Although not a cure, BMT has helped many affected individuals by ceasing the progression of many clinical features of the disease and by arresting any further disease progression in the central nervous system. Unfortunately however, the treatment is limited, as it cannot reverse neurological damage already incurred prior to transplant, and it cannot stop the bone and joint disease that is characteristic of the condition from continuing to progress.

Some children and young people who have undergone BMT experience difficulties adjusting to society and the demands of everyday life. Some can have difficulty carrying out simple tasks such as shopping or opening a bank account. There may also be emotional issues, particularly relating to social acceptance, peer relationships, sexual awareness, and frustrations arising from physical disability. However, some individuals cope well with their disabilities and have found ways to adapt to live independent lives. The main aim of this research was therefore to identify the factors in these patients' lives that contribute to their overall quality of life, including disease- and disability-related factors, as well as parenting, familial, and social factors.

The study was based in the UK and focused on a cohort of patients affiliated with one of 5 main tertiary centres that specialise in genetic disorders of metabolism and bone marrow transplant. These centres were Great Ormond Street Hospital for Children, Royal Manchester Children's Hospital, Royal Hospital for Sick Children Bristol, Diana Princess of Wales Hospital Birmingham, and University Hospital Wales. It was carried out over three years by a psychologist working for the MPS Society. After three years this research project is now complete. A qualitative pilot project was conducted by the Society in 2003 and work on the main study, which employed a more

systematic approach and quantitative research methods, began in 2004. A total of 44 families participated, which was 90% of the total population of MPS IH patients living in the UK post-BMT at the close of data collection in May 2005. The age range of the patient participants was 15 months to 25 years.

Findings of the study illustrate how aspects of the MPS disease itself, such as physical and cognitive functioning can impact upon patient adjustment. Adaptive skills and internal coping resources such as self-esteem were found to contribute to more favourable patient outcomes however, as did certain parenting and familial factors. The study revealed patient adjustment to be multifactorial, with aspects of the MPS disease, physical and learning disability, family functioning, and parental beliefs and attitudes all contributing. More detailed publication of these findings will be available in due course.

Update on future research projects

Hello everyone, just a quick note to let you know that I will be on maternity leave from the beginning of June until the end of the year. From January 2007 I will be making preparations for two new research projects however, and soon after that I will be inviting children and adults affected by Fabry disease and Morquio syndrome to volunteer to participate. Both studies will be exploring the personal experiences of living with these two conditions, including the rewards and challenges that they can bring to your lives. Before the studies get properly underway however some time will be spent in development. For this process it would be useful to talk informally to some of you about your experiences. This will be done either at clinics or on a one-to-one basis at your home or similar location. More precise information regarding this will be sent out to you some time in 2007. I very much look forward to meeting you. Have a great summer!

Cheryl Pitt

How are you going to make an impression?

Everyone likes to get dressed up.
So on **Friday 6th October** jump into your jeans and make a donation to help raise £4 million to fund pioneering research and vital support services for children affected by genetic disorders.
Make an impression on a child's life today.

For your FREE fundraising pack visit:

www.jeansforgenes.com

or call freephone:

0800 980 4800

Jeans for Genes Day

Friday 6th October 2006

Dress to Impress

Ten national charities working together to help children affected by genetic disorders

The net proceeds from the 2006 Jeans for Genes Appeal will be distributed among the charities