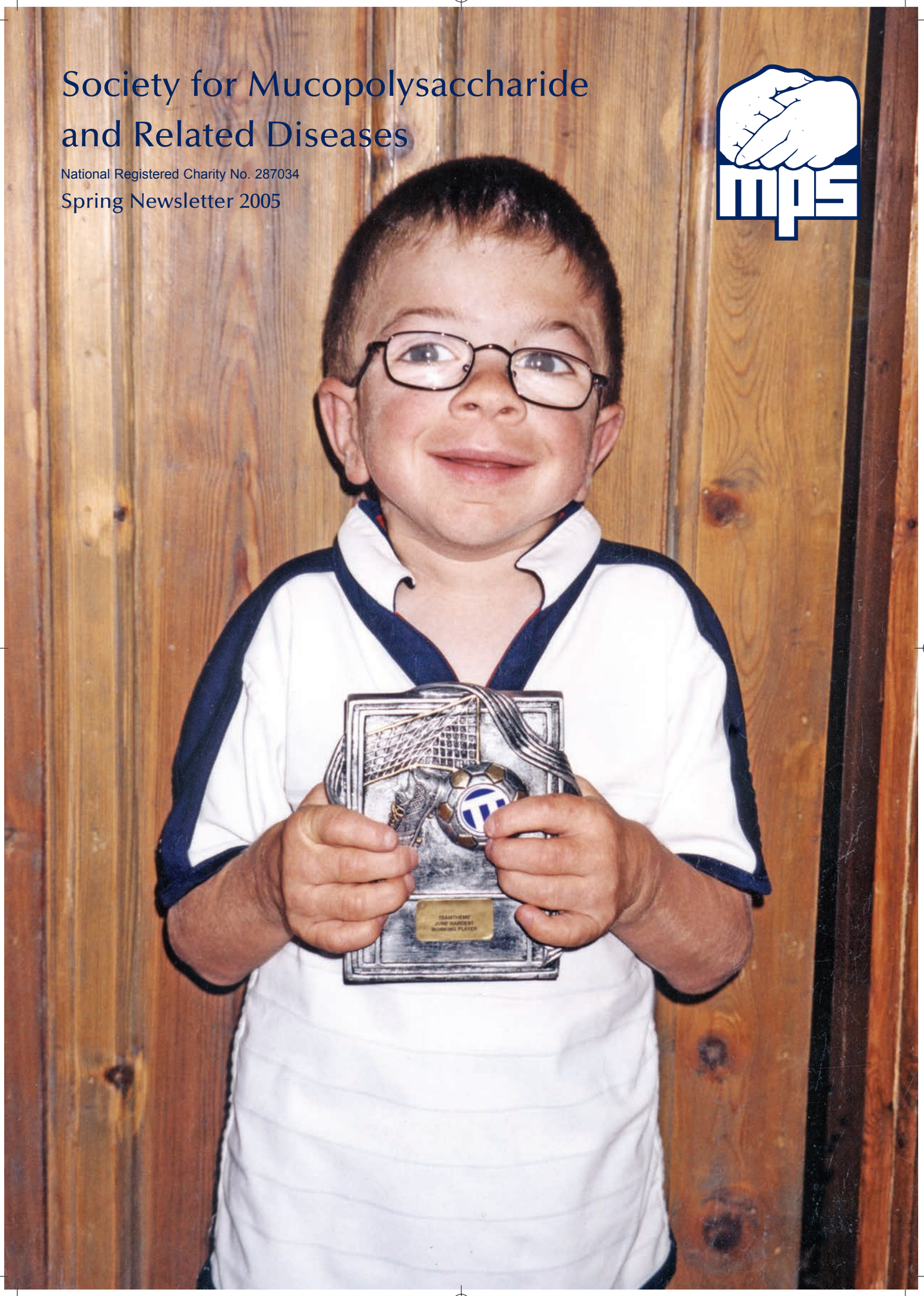
Society for Mucopolysaccharide and Related Diseases

National Registered Charity No. 287034

Spring Newsletter 2005



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 1000 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 Young People & Adults 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 including a video

Quarterly Newsletter
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

Read more about our cover star, Bradley Evans, MPS I, on page 6

CARE TODAY HOPE TOMORROW

Newsletter

MPS Society

46 Woodside Road, Amersham
Bucks HP6 6AJ
T: **01494 434156**
Out of Hours: **07712 653258**
F: **01494 434252**
E: **mps@mpssociety.co.uk**
www.mpssociety.co.uk
Registered Charity No. 287034

Newsletter Deadlines

Summer	1 Jun 2005
Autumn	1 Sep 2005
Winter	1 Dec 2005
Spring	1 Mar 2006

Become a Friend of MPS

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

CONTENTS

Welcome to the MPS Newsletter Spring 2005. Spring is a new beginning and we've been working hard on a brand new website, a new look for the newsletter and the launch of Friends of MPS which you can read more about in the following pages.

We are always very pleased to hear from you with your stories, articles and photographs so keep them coming in. By sharing your experiences you can help and inspire our other members just like Bradley, our cover star.

Antonia Crofts
Editor

THE ROALD DAHL FOUNDATION

This MPS Newsletter has been funded with the kind support of a grant from the Roald Dahl Foundation.
www.roalddahlfoundation.org

CHIEF EXECUTIVE'S REPORT

Spring 2005

There is a lot to look forward to this coming year at the MPS Society, but we start the year with the sobering knowledge that Andrew Wragg, Dad of Jacob (MPS II, Hunter) is standing trial for murder of his son during March. Other families have endured the loss of loved ones suffering from MPS and since the last newsletter yet more families are coming to terms with the devastating news that a family member has an MPS or Fabry diagnosis.

Looking forward to the summer and with still three months to the MPS Conference Weekend, it is absolutely wonderful to see so many families booked – over 70 to date. We can still accommodate a few more of you, but only smaller families whose children can manage stairs. We have a very exciting programme planned for the children and volunteers and can only hope that the sun will shine even more brightly from 1-3 July.

Many of you have complimented the Society on the newsworthiness of the Newsletter. It wouldn't be possible without all of your help in providing your stories and news. We hope you would agree that the newsletter could be of value to a wider circle of people, grandparents, friends, and local professionals. With this in mind we are launching 'Friends of MPS'. Everyone signing up as a friend and paying an annual subscription will receive regular information including the quarterly newsletter plus invitations to MPS events at preferential rates. Wouldn't it be wonderful if every MPS family in the United Kingdom signed up three friends? To help you on the way we are enclosing three 'Friends of MPS' subscription forms. There are plenty more where these came from! Becoming a 'Friend of MPS' is a wonderful way of spreading public awareness.

Lastly, I would like to leave you with the exciting news that the MPS Society has awarded £341,635 in research grants. £274,035 from Jeans for Genes, £60,000 the proceeds of the 'Ollie G Ball' organised by David Gosling of Country Wide Special Events and £7,600 from the proceeds of the Charity Shop based in Bristol and run by Marina Foster and her three volunteers. You can read more about these research grants later on in the newsletter.

As the year unwinds, the MPS team looks forward to meeting as many of you as possible at the Weekend Conference, clinics and MPS events.

Christine Lavery
Chief Executive

News from the Management Committee

The Society's Board of Trustees meet regularly. Here are the key issues discussed at their Budget meeting held on 27 November 2004 and the Trustees meeting held on 4-5 February 2005.

MPS Budgets

The Chief Executive spoke to both the Draft Income and Draft Expenditure Budgets for year commencing 1 November 2004 and a full discussion took place. The draft budgets were agreed without amendment.

Proposed Barry Wilson
Seconded Bob Devine

Generating Funds

The Finance Officer spoke about the raffle and Trustees asked whether the cost of tickets should be increased? The Trustees agreed unanimously that raffle tickets for the Winter 2005 draw should increase to £1 a ticket and be issued in books of five. Trustees were also advised by the Chief Executive that she had had confirmation from the organisers of the 'Ollie G' Ball that the Society is to receive a minimum of £118,000. Half of this sum is allocated to research.

Policies

The Trustees reviewed and agreed the following policies: Confidentiality, Health & Safety, Recruitment and Management of Staff, Recruitment of Ex-Offenders, Out of Hours Support, HIV & Aids, Financial Assistance Scheme, MPS Copyright, Organising Regional Events, Display Screen Equipment Eyecare, Whistleblowers, Internet and E.mail, Handling Poor Performance, Reserves, Animals in Research, Use of Electronic Equipment, CRB & Disclosure Policy, Managing Stress in the Workplace, Sickness & Absence and Gift Policy.

Risk Management

The Trustees received a report from Titixia Shah from the Department of Trade and Industry who as part of her 'week in industry' undertook a review and scoping exercise to inform Trustees. Trustees agreed that the report was very helpful and asked that their appreciation be extended to Titixia.

New Office Premises

Trustees shared their disappointment that the owner of Flint House had decided not to sell and affirmed their agreement that the Chief Executive continue with the help of the agents to identify other suitable premises that would enable the Society to operate more effectively and offer space to develop an information resource.

MPS Research Grants

The Trustees were joined by Tristan Millington Drake representing the Shauna Gosling Trust and received presentations in respect of five of the six short-listed grant applications. It was agreed the decisions will be taken by 1 March 2005. The Trustees also confirmed Year 2 funding for a grant to the Institute of Child Health, London for the 'Identification of Biomarkers for the Mucopolysaccharidoses'.

Publications

The Chief Executive gave Trustees details about the book written by Richard Dunn, 'A Chance of Life' relating to his grandson Isaac who has undergone Bone Marrow Transplantation. It was unanimously agreed the MPS Society as per the discussions with Mr Dunn take forward this publication.

Childhood Wood

The Chief Executive informed Trustees of her discussions with the Forestry Commission. The plans for taking forward the development of the Childhood Wood were agreed along with a budget of £5,000.

Proposed Sue Peach
Seconded Bob Devine

Jeans for Genes

Trustees were informed that the total is now very close to the £3 Million mark. Several Trustees were keen to understand more about Jeans for Genes and it was agreed to invite the J4G Chairman to address Trustees at a future meeting.

INTRODUCING...

The Society has recently recruited two new members of staff and they introduce themselves to you here...

Linda Norfolk

Hi! My name is Linda Norfolk. I joined the MPS Society at the end of November 2004 as Advocacy Assistant - they tell me you don't have to be mad to work here but it helps, so I fit in rather well!

Before joining the MPS Society I had two part-time jobs working for Age Concern Hillingdon as an Administrator and also as a Ward Clerk in the Elderly Day Unit based at Mount Vernon Hospital in Northwood. I decided that as my son was now 15 years old, I no longer really had an excuse not to work full-time, so here I am. I am enjoying myself so far, the day just flies by as I am so busy, and I hope that I will be able to put my skills to good use in supporting members in the future.

Maureen Cummins

Hello everyone! My name is Maureen and I've joined the MPS Society team for six months. Everyone here is really friendly despite being very busy! Before I came to work here all I knew of the MPS Society was the Jeans for Genes collection we had every year at the place where I used to work.

The Society has a database of information about patients, including their disease and symptoms. My job will be to make sure that all the details we have in paper files are transferred to this database. I will also be contacting families to find out more. Since this information will be used for research into MPS diseases, it is important that it is as full and accurate as possible. To begin with I'm working on data relating to international patients living outside the UK. I've been surprised at the number of different countries with which the Society has contact.

I'm very impressed so far by all the Society does, so perhaps when my six months is finished I will be able to be involved in some way as a volunteer.

Meet our cover star, Bradley Evans, aged 10, MPS I, by Pam Evans, Bradley's Mum

Bradley's favourite sport is football. I think it is safe to say he is football crazy. When Bradley brought the letter home from school asking could he attend an after school football club that was being run by a outside agency, I was a little apprehensive. But Bradley was so excited and all his friends were going to be there so I liased with school and we decided it would be fine for him to attend. I wrote down all Bradley's complaints and the football coaches agreed to take him.

The course ran for four weeks and Bradley's only concern was which kit should he wear! Bradley had a fantastic time getting 'professionally coached' as he called it and then playing matches and finally a tournament.

The last week came and all the parents were asked to attend for the presentation. Off we set, Mum, Dad and big brother Elliott. The trophies were given in age group order. When it finally came to Bradley's age group, the coach gave this speech about how hard this young lad had tried and nothing had phased him and he had given his all, even though he was smaller than most in his group. At this time Elliott turned round and said 'It's Brad' and I thought no. Then the coach said 'He's already a legend at this school, come on up Bradley Evans'. Bradley played it really cool, went up, shook hands with the coach and then turned round to all his fans and lifted his trophy high to an almighty cheer from his friends. It was a moment we perhaps thought we would never see and is one we certainly will never forget.

Announcements

New Members

Helen Tandy has recently been in contact with the Society. Helen has Fabry Disease and lives in the North West of England.

Deaths

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

Cathleen Hedgecock who died at home on 31 December 2004. Cathleen had Fabry Disease.

Sam Mullen who died at home on 28 January 2005. Sam had MPS IV, Morquio Disease.

Evie Mason who died in hospital on 21 January 2005. Evie had ML II.

Jonathan Heisig who died in hospital on 18 March 2005. Jonathan had MPS III, Sanfilippo Disease.

Congratulations to the Ingram Family

Matthew became a big brother on 16 February 2005. His sister is called Melissa and she was a chunky 8lb 2oz at birth. Matthew is already very loving and protective towards her and no signs of jealousy so far.

Congratulations to
Sally and Nick Massey,
married on 11 June 2004

Sally's brother, Ross, suffered from MPS II, Hunter Disease, and died at the age of 16. Throughout her younger years, Sally and her family were frequent participants at MPS conferences and regional events held in Wales. We wish Sally and Nick every happiness.

ANIKA HAJI-AFZAL
wins Good Carer Award

Anika Haji-Afzal is a very special young person, a fact that was recognised during a ceremony in October 2004 when she was one of only seven winners of the Douglas Allen Spiro Annual Children's Award. 12 year old Anika was nominated by the Befriending Service and was the clear winner for the 'Good Carer' award.

Anika's two sisters were cared for at Richard House Hospice. Anika also has a befriender from Richard House who spends regular time participating in activities with her.

Nominations, for outstanding children aged 16 years or under, were received from all over East London and West Essex and a panel of judges found the competition so tough that they finally awarded seven prizes across four categories. Anika's prize was £1,000 worth of travel and she was the only prize winner in her category. Sadly, soon after this award, Anika's two sisters, Alisha and Azaria, died from ML II on the same day.

Anika Haji-Afzal and her family

Why don't you become a Friend of MPS?

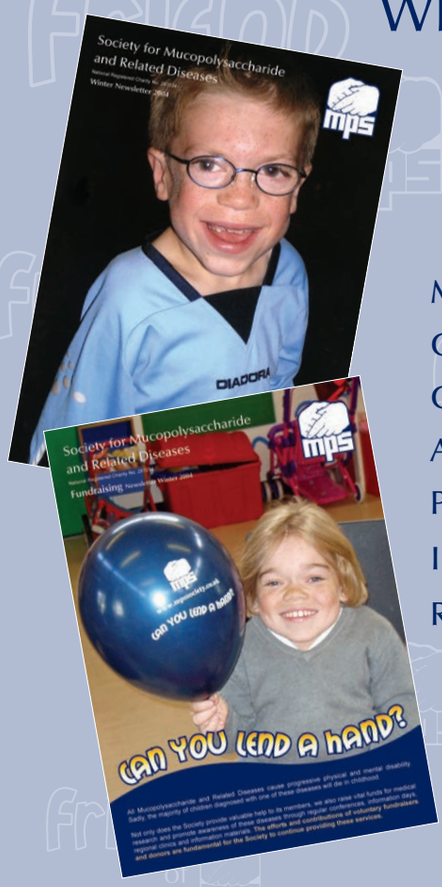
To promote the Society to a wider audience and encourage more support we are launching a new initiative called the Friends of MPS.

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

If you are not eligible to become a member of the Society why not become a Friend of MPS? Complete the enclosed application form, phone us, or download a form from our website www.mpssociety.co.uk.

All our existing newsletter subscribers will automatically become a Friend of MPS. Over the next year you will be receiving the latest editions of the newsletter, fundraising newsletter and annual report but there are other benefits. Read on to find out more...

What are the benefits of becoming a Friend of MPS?



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

If you want to do more than just become a Friend of MPS ask for a fundraising pack to find out more.
Phone us on 01494 434156

As a Friend of MPS you will help the Society support its members.

Here are just a few ways...

Childhood Wood

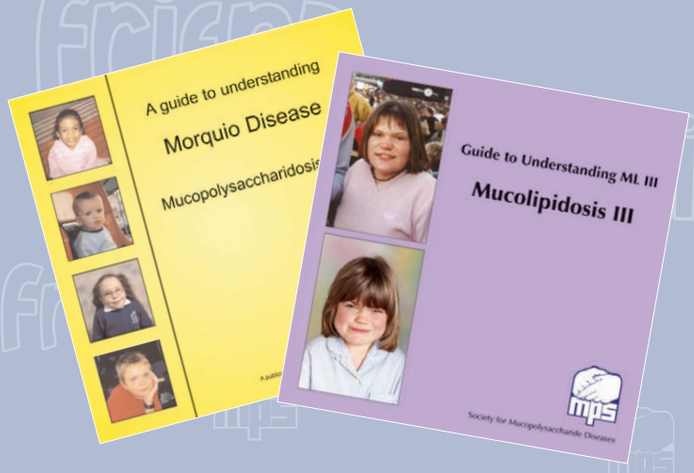
In 1993 the Society was given under licence an area of Sherwood Forest to create a wood of oak saplings cloned from the Great Oak as part of the Forestry Commission's Sherwood Initiative. Each of the oak trees celebrate the life of an individual who has lost their lives to these cruel, degenerative diseases. Families who have lost sons or daughters can remember their loved ones in the tranquility and beauty of the Wood. Each tree has a plaque inscribed with a name and short message. 'We are acutely aware of how little we spend on this planet. What we do now is left as a legacy to our children and their children. We have a responsibility to leave the Earth a better place than we found it. If the Childhood Wood helps a little then all those individuals who have been born with MPS will have helped in their own way'. **Becoming a Friend of MPS would help the maintenance of the Wood and leave a lasting legacy for the future.**

Advocacy Support

As parents we nurture and guide our children through each stage of childhood. From those first few wobbly steps, their first day at school, through to full independence and on to adulthood. Parents of those suffering from MPS and Related Diseases are denied such pleasures. These parents have no choice but to watch their children suffer progressive physical disability. Since being founded in 1982, the Society has become recognised worldwide for its expertise in supporting individuals affected by MPS and Related Diseases. The Society offers practical help in areas such as health, welfare, housing, education, respite care, specialist equipment and palliative care plans. **Becoming a Friend of MPS would contribute to this unique advocacy service.**

Over the past year the Society's advocacy support team supported over 600 individuals and their families.

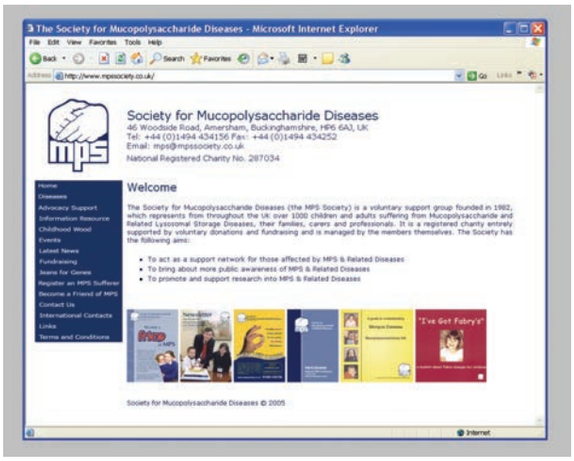
Information Resources



The Society has developed specialist information booklets in disease management for individual members, their families, carers and professionals. A range of booklets for affected children, their siblings and school friends has been developed with the children themselves to help explain in language accessible to children about their diseases and how that makes them different. As the Society is only a small charity with considerable commitments to both research programmes and advocacy support, **becoming a Friend of MPS would help us to reach as wide a readership as possible and would contribute towards printing and postage.**

Events

The Society organises regional MPS clinics throughout the UK, information days and conferences in Scotland and Northern Ireland. A National Conference is also held offering individuals and families the opportunity to learn from professionals and each other. Seminars, workshops and key speakers who are experts in the field of MPS Diseases attend this event. From time to time, the Society organises other special events. **Becoming a Friend of MPS would help to subsidise the attendance of an MPS individual at one of our events by up to 50%.**



www.mpssociety.co.uk

Have you checked out the new MPS website? Over the coming months we hope to make it as informative and accessible as possible. Containing lots of valuable information on MPS and the Society why not check out the new fundraising pages, donate online, download order forms, request a fundraising pack and get inspired with new fundraising ideas.

School Talks by Sophie Denham

Since September of last year, I have been involved with sharing information and giving presentations to a number of schools across the UK.

These have been in response to requests from families and from schools themselves. These talks have been key to my current work at the Society and have supported individuals and families in a number of ways getting for example the resources they need, informing Special Educational Needs Statements, greater knowledge and awareness of individual diseases and how they can affect individuals.

In undertaking these talks and the giving of information, I try to work closely with families and include them within the planning and giving of the presentation, promoting joint working and working in partnership. This method of working also allows the talk to be tailored to the individuals bringing in the needs of the child and how they should be best met by the school.

These talks have proved invaluable in educating schools, teachers and carers on specific MPS diseases and the response from parents has been that the talks had made a difference and the schools had taken a more positive approach in not only securing important resources but also in best meeting the child's needs.

I have really felt honoured to be a part of this and hope to continue to help make education a positive and enjoyable experience for individuals and their families.

If anyone would like a talk to be given at their child's school, please do not hesitate to contact the Advocacy Support Team on 01494 434156 or email advocacy@mpssociety.co.uk.

Birmingham Regional MPS Clinic

After close liaisons with Birmingham Children's Hospital, a new outreach clinic has been established in the Birmingham area. This clinic is to take place twice a year at Victoria School, Northfield (south Birmingham) with the first clinic held on 30 March 2005.

This clinic is now an integral part of the metabolic service at Birmingham Children's Hospital (BCH) and is being run by the Birmingham Metabolic Team. Patients attending this clinic will be seen by Dr Ed Wraith from the Royal Manchester Children's Hospital as well as either Dr Anupam Chakrapani or Dr Chris Hendrksz from BCH.

Priority for appointments will be given to those patients who are traditionally Dr Ed Wraith's and those for whom the facilities, which the BCH team has assessed as more suitable for older children and adults who are wheelchair dependent, are specifically needed. We have also been reliably informed that there is ample parking, which is definitely an added bonus. Adults in the West Midlands for whom there is no other expert clinical service have also been invited to request appointments at this clinic.

The clinic is being co-ordinated by Joy Hardy (formerly Joy Wright - congratulations on your recent marriage) who will be liaising with individuals and families to secure appointments.

Whilst the MPS Society is no longer arranging the clinic we are still very closely involved and will be attending the clinics as we have done in the past. I am looking forward to meeting those individuals and families who have been given appointments at these clinics. For the first time ever we have the facility of a room to allow individuals and families the opportunity to speak to us in private, something that many of you have requested in the past. We are extremely pleased that a clinic for this year has been secured and are very grateful to both the BCH team and Dr Ed Wraith for their commitment to providing a clinic facility in the West Midlands and working with MPS in setting up this clinic.

Any queries about the clinic or about appointments for this clinic need to put forward to Joy Hardy at BCH. If however, you have any questions about our role at the clinic or you wish to discuss anything or need us to bring something to the clinic please do not hesitate to contact us.

Newcastle MPS Regional Clinic

On a very early and cold Tuesday morning, Ellie and I set off to catch a plane to Newcastle for the annual MPS clinic. Although the weather was cold, at least we did not have snow, which has in the past caused individuals and families to miss their appointments. 11 individuals and their families were seen at the clinic which ran smoothly and allowed appointments to run relatively to time.

It was lovely to meet all those who attended the clinic and to be available to offer advocacy support as needed. A benefit of the Newcastle clinic is that it is able to afford us our own room, which allows individuals and families to speak in confidence away from the hustle and bustle of the general waiting room. This is a provision we hope can continue to be afforded to us.

As always we would like to extend our thanksto Dr Wraith, Dr Rylance, Dr Leech and the nursing team, for their continued support and dedication to the clinic.

Photo top right: Dr Ed Wraith, Callum McDonagh (ML III), Mrs McDonagh, Dr Rylance and Dr Leech; Photo bottom left: Daniel Muers (MPS II)

Clinic Dates 2005

Bristol
Tuesday 7 June
Tuesday 29 November

Cardiff
Wednesday 30 November

Northern Ireland
Thursday 12 May
Thursday 17 November

A week in the life of an ADVOCACY SUPPORT OFFICER

by Clare Cogan

Monday

In early, bit bleary eyed after flying back from Rome the previous evening where Ellie, Christine and I had been supporting families attending the International Fabry Patients Meeting from Thursday through to Sunday. This was a good opportunity to meet families and individuals and talk to them about a range of issues. Today was my 'catch up' day, it's amazing how much can happen when you are out of the office for two days!

Tuesday

Spent most of the day with another member of the advocacy team working through an issue concerning a family and the placement of one of their children. It's definitely a case where two heads are better than one to ensure that the family's views are fully and accurately reflected in all correspondence. In between phone calls and letter writing, gathered everything together for Thursday's Northern Ireland Clinic, making sure booklets, membership and registry forms are all to hand, oh and don't forget the Christmas cards. By the time I had finished there was no room for clothes; I've learnt very quickly you travel light when you work for the MPS Society.

Wednesday

5am start. I am a morning person but leaving my bed at 4:30am I would definitely class as the middle of the night. One cup of hot chocolate with marshmallows and whipped cream later and I was ready to go (I don't think Ellie could believe I was drinking that at 6:15am!) First challenge of the day, how to negotiate Belfast city in rush hour, in a hire car. Lesson 1, be sure you test the brakes before you start to discover they are a lot more sensitive than your own car's so you don't send your manager through the windscreen! Lesson 2, go in the direction your navigator tells you to! Teething problems over, our first visit was a meeting at a family home with representatives from housing to discuss future housing/adaptations. Provided information about the disease and issues to consider when looking at properties and potential adaptations. Agreed to write a report in support of the family's needs and to clarify points discussed in the meeting. On leaving, Ellie had to prevent me from acquiring the family's six-month old Alsatian puppy; I relented only because I didn't think she would fit in our hired Kia Picanto!

Afternoon visit involved a drive across the beautiful Irish countryside, meeting the Children's Community Nurse in a service station so we could follow her to the house. Met with the family and discussed issues relating to housing, a pertinent issue and funding for domestic appliances to support the family in the care of their disabled child. After several telephone conversations, also managed to arrange the repair of the washing machine. Also provided some bereavement support following the recent death of another of their children.

Back to the hotel to plan the induction for a new member of staff starting next week, it never stops…

Thursday

To the hospital for a full day of appointments at the six-monthly Northern Irish Clinic. Met one new family and arranged to visit them on my return to Northern Ireland in a fortnight to support them with completing their Disability Living Allowance forms. Also spoke to others and picked up support issues, mainly housing problems. It was good to meet some of the families for one of the areas that I cover, the clinics being a very good way of meeting face to face.

Then, back up in the air for a very bumpy ride back to Heathrow. I feel at the moment that I am spending more time in the air than on the ground! Arrived back at 10pm ready for a day off tomorrow to let my body catch up and be ready for whatever next week brings…

Focus on the Individual Advocacy Support Service

CARE PLANS

One of the services provided by the Society's Advocacy Support Team is to write careplans for individuals affected by an MPS or Related Disease.

These careplans include specific details on various issues to be considered when providing personal care support. They also give details of an individual's cognitive ability & address some pertinent medical considerations which are essential to be aware of when caring for a child or adult with an MPS or Related Disease.

A careplan may be written for any one or all of the following reasons:

To provide detailed information which the family or individual can use to inform local carers and professionals of the issues which need to be addressed in caring for the child or, in the case of an independent adult, themselves or...

To act as an information and assessment tool to support local professionals in carrying out a full and accurate assessment of need which in turn will inform the nature and amount of local support implemented or...

To provide a resuscitation policy which gives details of the family's or individual's wishes in respect of life-saving treatment in the event of a medical emergency or prolonged illness.

A careplan is written specifically for each individual in partnership with the individual & their family, medical practitioners and, as appropriate, local professionals and support workers.

Each care plan is dated and a review date is included to ensure that information in an old document is not acted upon inappropriately. When a careplan is up for review, it is the responsibility of the individual or family to contact the Advocacy Support Team to inform them an update is required.

Two doctors, who clarify that the information given is medically correct and that any protocols are appropriate for the individual concerned, sign the careplan.

When a resuscitation policy is included the Advocacy Support Team work very closely with the key specialist doctor to ensure the implications of the policy being signed up to are fully understood.

An MPS careplan is by no means required by every individual and family as social services, school and other local agencies have a duty to complete one when their services require it.

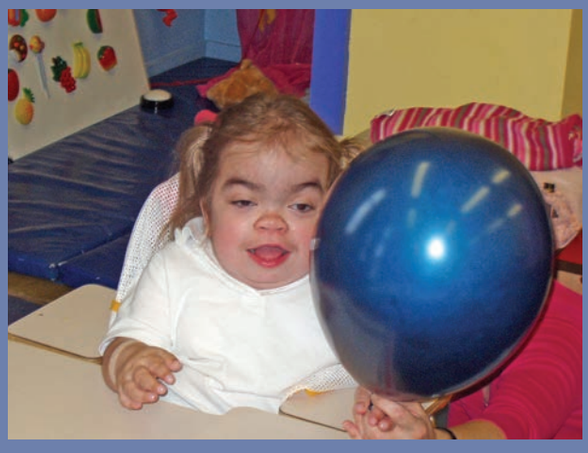
MPS and Related Diseases are rare conditions so an MPS careplan can help inform the local careplan or be used in its place. An MPS careplan has also, on occasions, been an extremely useful tool used by the Advocacy Support Team to advocate for alternative or increased local support services as it clearly demonstrates the needs of an individual with an MPS or Related Disease.

For more information about MPS Careplans or the work of the Advocacy Support Team please contact the Society.

In July 2004 a nurse contacted the Advocacy Support Team advising that Courtney's local doctors were requesting an updated MPS care plan. Courtney is 8 years old, lives in Scotland and suffers from MPS I.

After contact with Courtney's mother Karen a meeting was set up to which local professionals who had a constructive input to give into the careplan were invited. The meeting required a very sensitive and skilled approach with Karen's need for support to voice the significant deterioration Courtney had suffered since the previous careplan had been written (she had lost her sight and speech and could no longer weight bear) to meet and balance with the professionals input whose perspective at times differed significantly to Karen's.

Having updated the care plan Karen fed back how she was reassured that her daughter's needs were recorded for all to understand and that she was able to use this document rather than describe all aspects of Courtney's care to every new person involved.



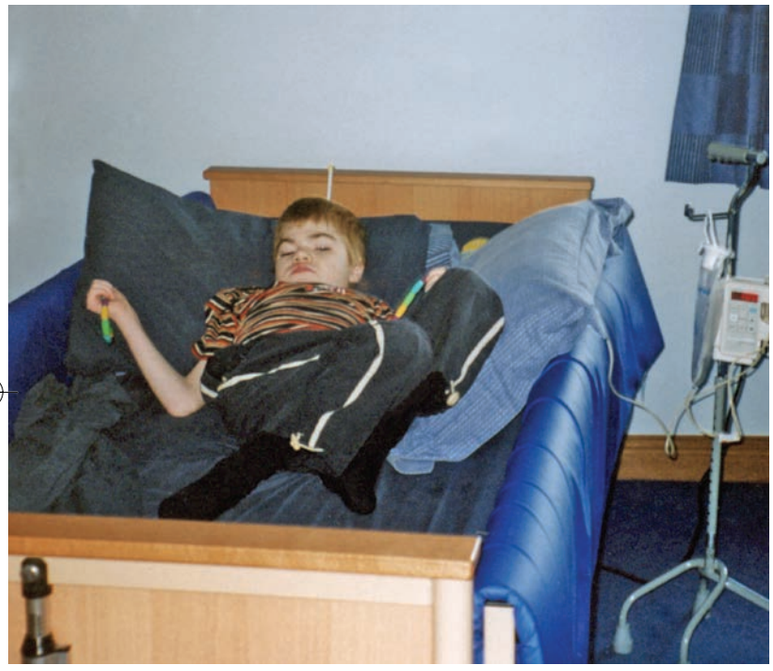
Case Study: Courtney, MPS I

Kyle and his gastrostomy tube

Following on from an article that appeared in the MPS Newsletter Autumn 2004, Alison Shields, Kyle's mother, updates us on Kyle's progress.

Kyle had his gastrostomy as planned and has recovered well. He was admitted to Barbour Ward in the Royal Belfast Hospital for Sick Children on 24 August 2004 for his operation the next day.

Kyle was assigned a bed in the middle of the ward across from the nurse's station. His named nurse went through all his details such as his weight, allergies, medication, previous operations, hospital visits and so on. Thankfully I was prepared and had them all listed.



Kyle soon moved to his own room, which had a large adjacent bathroom with shower, and also a folding bed, which I was able to put down beside Kyle at night. It was a lot quieter and had plenty of room for his buggy and all his bits and pieces.

Kyle had to have the dreaded blood test. The doctor informed me just after 8pm that Kyle's platelets were dangerously low and they would have to do another blood test to double check. As things stood they would not be able to operate because Kyle's blood would not clot as it should.

So, after the 'magic cream' was applied to dull the pain, it took three attempts this time to get blood and a lot of praying between times. At about 9.30pm I was told that the sample was fine and Kyle would be first in theatre the next morning. Kyle, oblivious of what was ahead of him, had quite a good night's sleep, and woke just after 8.30am.

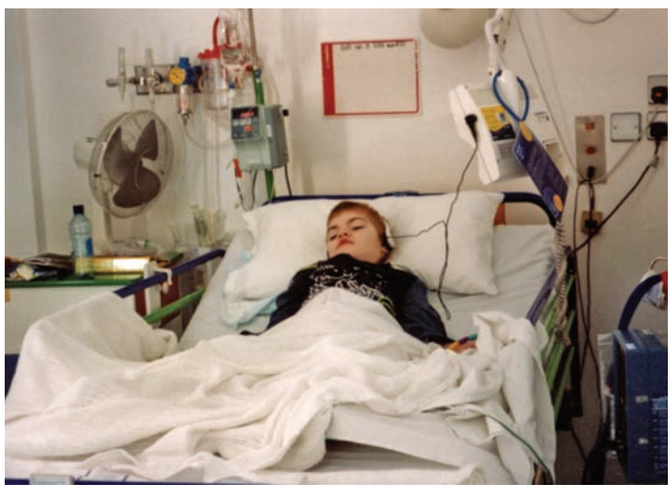
Kyle was wheeled down to theatre in his bed and lifted over onto the operating table. The anaesthetist put him under with gas and at that stage I had to give him a tearful goodbye kiss, leaving him in the capable hands of the medical team while I returned to the ward to wait. The ENT surgeon came to see me at 9.50am to tell me he had seen Kyle and had stretched out the tissue that had gathered from the site where he had had his adenoids out a few years before. The surgeon assured me Kyle was fine and the gastrostomy was going ahead.

I was called down to recovery at about 11am where Kyle was still very drowsy and lying under a heated blanket to try and raise his temperature, which was presently 33 degrees. We got back to his ward just after midday once his temperature got to 35.5 degrees. Kyle slept on and off for the rest of the day and his temperature and blood pressure, which was also low, were both monitored closely. Kyle spent another three nights in hospital before getting home on Saturday just after lunch.

It's now a few months since Kyle's operation and we are coping well. Kyle at present is being fed 400mls over 10 hours during the night and also three bolus feeds during the day. A total intake of 800mls daily of Tentrini energy multi-fibre feed. We are aiming to gradually increase this to 1000mls.

We have learnt by our mistakes. Kyle doesn't sleep well and wriggles about in bed quite a bit and on our second night home we got most of Kyle's feed over the bed as one of the valves had come apart. Kyle also likes holding things in his hands, so we normally give him his teething rings to hold, preventing him from grabbing at his tummy tube.

As time has passed we have become confident in the management of Kyle's feeds and I've even amazed myself at how quickly we have adapted. Since the operation we have already seen an improvement in Kyle. He has put on weight and has had fewer seizures. Getting his essential nutrients and medications via his peg has made a big difference.



Speech, Language & Feeding Difficulties

in Sanfilippo Disease, MPS III

A Speech and Language Therapist will see children with Sanfilippo Disease to help with issues around communication skills and feeding difficulties. The following article has been written by Rebecca Howarth, Senior Specialist Speech and Language Therapist at Manchester Children's Hospital.

Communication

Children with MPS III will require Speech and Language Therapy input as their skills start to deteriorate. Usually this will become apparent in the second stage of the disease. This therapy is generally best provided as part of the child's education as this means the Speech and Language Therapist will be able to give staff who are involved with the child on a daily basis aims and strategies to help them communicate to their potential.

The children may benefit from a total communication approach to maintain their ability to communicate both verbally and non-verbally. Total communication can be described as a communication approach where any one of a number of communication methods is used in combination between the child, parents and teacher.

Such methods of communication may include speech gesture and sign language, on body signs, objects of reference, symbols and the use of communication aids.

As the child begins to find communication increasingly more difficult, it may be useful to employ an 'intensive interaction' approach with them. This is a practical approach to interacting with people who have difficulties learning, communicating and being social. It aims to help the child and their parent, teacher and carer relate better to each other and develop everyone's communication abilities in order to support the child and develop the child's confidence and competence as a communicator.

Feeding

Children with MPS III show initial difficulties with oral skills i.e. mouth movements. Once again, this will usually become apparent in the second stage of the disease although the severity of these difficulties can vary greatly from child to child.

Children will take longer to finish meals and parents and carers may find they have to avoid certain foods i.e. dry or sticky consistencies. It may help at this stage to adapt their meals to include lots of moist foods (generally achieved by adding extra sauces to their foods).

Some children will then go through a stage where their eating skills and appetite are very inconsistent. This may occur at the same time as they are having problems with sleep and concentration. This can be very frustrating for parents as their child's ability to eat foods seem to vary from day to day.

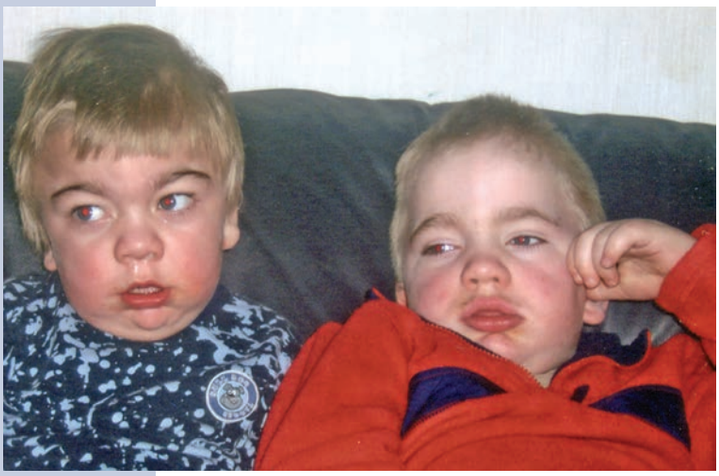
Parents may find that eventually it is easier for their child to eat very soft food. At this stage they may also begin to cough and splutter with drinks and, in some cases, it is useful to perform a videofluroscopy. This is a moving x-ray of the child's swallow and will show if there is any risk of food or drink going down the wrong way i.e. into the lungs. Drinks can be made safer by adding a thickening powder.

Sadly, some children will lose the ability to swallow safely even with thickened drinks and soft foods. In these cases a decision as to whether to feed them non-orally (usually by nasogastric tube or gastrostomy tube) will be discussed.

Do you have any stories, news or information that you would like to share with other readers?
If so, contact the MPS Society
on 01494 434156
or email us at
mps@mpssociety.co.uk

HOUSING ADAPTATIONS

We got there... in the end!



Our experience of achieving housing adaptations can only be described as a roller coaster… It took two and a half years to get the end result, a property all on one level which met the needs of my two sons, Connor and Craig who have Hunter disease. We are very pleased with the end result but would like to share our experience of the process we went through.

Our first challenge was getting the message across that the current accommodation we were living in was totally unsuitable and that the longer term needs of both Connor and Craig meant that we needed to move into something more suitable.

This was very difficult, no-one seemed to understand or even want to try to understand the needs of my two boys and were not prepared to listen to me. I found the support of the Advocacy Support Team invaluable in supporting me with this. They attended a meeting with the professionals involved, including housing and clearly outlined the long term needs of children with Hunter Disease and why this adaptation was necessary. To say these professionals were taken aback by this information was an understatement and I feel that this was the key turning point in things beginning to get done.

It was identified at this meeting that there were two empty properties available across the road from us but the main barrier in moving this forward appeared to be funding. I was faced with comments made like 'there are lots of other people on the housing list who have disabled children' and I had to hold onto what I felt was best for Connor and Craig to ensure that the situation was improved for them.

The Advocacy Support Team continually supported me throughout this process, looking at plans and arguing our case, giving reasons why we needed the adaptations and making sure that the professionals never lost sight of what was important in all of this, the quality of life of two of my children. I felt like everything was explained over and over again until they finally started work in February 2004. It took them 3 months to adapt the property and we finally moved in June 2004.

By Dawn McDougall

We are so happy with what has been achieved, my two older children benefited from the new house as they now have their own bedrooms and Connor and Craig no longer have to share. I feel that Connor and Craig enjoy the space in their new home, Connor now sleeps in bed in his own room whereas previously he would only sleep on the sofa.

The property is also safer, it enables me to contain the boys so that I know where they are and can supervise them, particularly as we now have a soft play room with a 'safe space'. Many a night I have to put up with them running up and down our long corridors when they are playing but feel that it gives them the opportunity to expend their energy so they will sleep at night! We also have a garden which is fenced in so I can allow them to play outside, again knowing that they are safe.

I am secure in the knowledge that this property meets Connor and Craig's needs now and for whatever the future holds. I had to keep shouting but eventually I was listened to and with the support of the Advocacy Support Team we got a positive end result. ■

Would you like support with a housing issue?

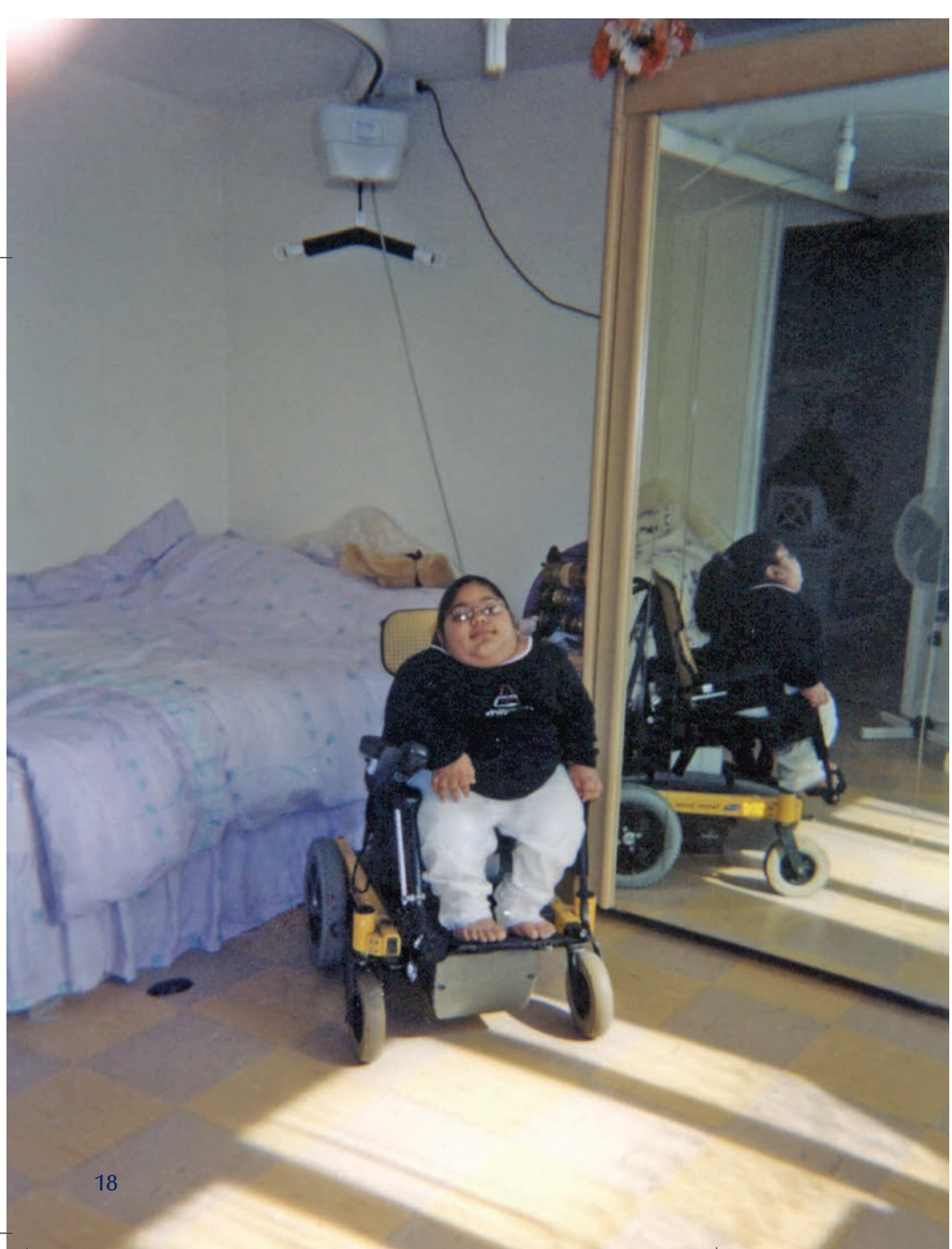
The Advocacy Support Team can provide our members with support on a wide variety of housing issues. This can include assistance at the outset in obtaining an assessment in relation to housing or providing a detailed report on the needs of a child or an adult's needs on an individual basis following consultation with the family or individual. This report would reflect their needs in relation to their MPS or related disease. We can also provide support throughout the process to ensure that the family's and individual's rights are upheld and that the process of any proposed adaptations or identification of an alternative property are needs-led and carried out in a timely fashion.

If you have any questions or feel that you would like the support of the Advocacy Support Team with this or any other issue then please do not hesitate to contact us. We are here for you to represent your needs.

Phone 01494 434156 or email advocacy@mpssociety.co.uk

An Advocacy Support Team Success Story

The Advocacy Support Team has supported Adikah, MPS IV, with a home adaptation and internal adaptations and furniture. This is Adikah's view on the final result…



MY BEDROOM
by Adikah Batool

My new bedroom is very big, and there's loads of space which makes it much easier to manoeuvre my wheelchair.

The walls have been painted (a creamy colour) but now I have decided I would like it painted in two different colours (mum and dad are not too pleased!) either baby blue and pink, or blue and lilac.

I really love my room as it gives me privacy and my friends come and sleep over.

My bathroom is really big too & has been decorated, but I'd quite like this tiled instead!

I would like to thank Christine and Sophie for all the help and support they have given to achieve this transformation.

CHANGES TO EDUCATION IN SCOTLAND

by Clare Cogan

A new framework for supporting children who require additional support for learning is due to come into effect in Autumn 2005. The Education (Additional Support for Learning) (Scotland) Act 2004 will give new rights to parents and place new duties on education authorities, health boards and other agencies specified by Scottish Ministers. It is important that parents who have a child who is educated in Scotland are aware of this Act and the possible changes which will occur when the Act becomes Law, at present this will be the summer of 2005.

Additional Support Needs?

The Act will mean that the term 'special educational needs' will no longer be used by professionals. Instead, the term 'additional support needs' will be used. Children who have additional support needs will be any who, for whatever reason, require additional support in the long or short term in order to work to their full potential. Additional support needs may result from a child's family circumstances, disability or health or from their learning environment.

Co-ordinated Support Plan?

For those who have enduring, complex or multiple barriers to learning and require support from a number of different agencies, including health and education, a Co-ordinated Support Plan (CSP) will be drawn up. These will replace the 'Records of Need' which are currently used to provide information about a child's needs and outline provision for their education in order to meet their special educational needs. The Education Authority will have responsibility for the Co-ordinated Support Plan and for the co-ordination of the support within it, which could involve areas outside of Education and aim to co-ordinate multi-agency services for a long term period for a child. However, other agencies, such as Health and Social Services have a duty to assist Education Authorities in implementing the plan.

Also under the new Act, an education authority must plan for post-school transition at least 12 months before a young person with additional support needs leaves school.

New Rights?

The Act is intended to enable parents to play a greater role in their child's education. Parents will have the right to ask the education authority to find out whether their child has additional support needs; whether they need a CSP, or need an existing CSP reviewed; and to request specific types of assessment to be undertaken. Parents views will also be taken into account and noted on CSP's.

How will it be introduced?

The existing special educational needs framework will remain in place until the Education (Additional Support for Learning) (Scotland) Act 2004 comes into force in autumn 2005. Any child or young person who has a Record of Needs (RON) when the Act comes into force will automatically have additional support needs.

Education Authorities will have to re-assess all children in their area who have a RON. Where a recorded child is not eligible for a Co-ordinated Support Plan (CSP), the authority must maintain existing levels of provision for up to two years unless a child's needs were to change significantly. If, when the child or young person is reassessed, they are eligible for a CSP, they will receive one. Until the Act comes into effect in Autumn 2005, any child or young person who is assessed as requiring a RON should receive one.

A Code of Practice has been devised and is currently being consulted on which provides guidance to all those who have duties under the new Act. This will include staff in education, health and social care as well as early years and childcare workers. It aims to clarify duties under the Act to ensure that a child and young person's needs are met and rights are upheld.

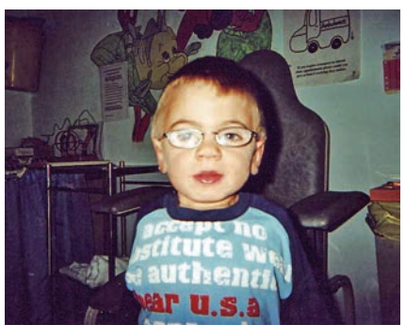
What if there are disagreements?

Dispute resolution is in the process of being developed. The aim of dispute resolution will be to help parents resolve disagreements about their child's additional support needs that cannot be dealt with. It will be for parents whose child has additional support needs, but does not have a CSP. Appeals relating to CSP's will be dealt with by the new Additional Support Needs Tribunal System.

It seems there is a way to go in developing and implementing this system, with those responsible for implementing the Act starting from the beginning and receiving training, information and guidance. All children in Scotland will be affected by these changes and any developments, as they arise we will endeavour to communicate to you in future newsletters.

If you would like to understand more about what is happening and keep track of developments there are several websites.

These are www.enquire.org.uk, www.parentzonesscotland.gov.uk/specialneeds or www.scotland.gov.uk which contains 'A guide for parents; the Additional Support for Learning Act'.



The Advocacy Team is currently supporting Jordan Mount's family with education prior to the new Act becoming law later this year. This has involved the writing of a report outlining Jordan's educational needs and providing as much information as possible to the professionals to ensure appropriate school provision is made available.

Our trip to Disneyland

by Claire Stevens

My name is Claire Stevens and my husband Bob and I have two sons, Oliver (5) and Samuel (3) who both suffer from Hunter Disease, MPS II.

Oliver was referred by his old nursery to 'Make a Wish Foundation' for a wish, and has wanted to go to Disney ever since he saw an advert on one of his videos, so, late January, courtesy of 'Make A Wish' and 'Give Kids the World' wish village in Florida, we began our trip. Oliver was very proud to wheel along the Buzz Lightyear suitcase he had been presented with, as was Samuel who was given a 'Nemo' one.

'Make A Wish' had sorted everything. We pulled up outside the airport and a man was waiting to park our car for us. We were given a letter to hand to the Virgin Atlantic cabin crew introducing Oliver and the purpose of his trip and before we had even sat down there were two hostesses making a fuss of the boys, taking us up to meet the captain and generally going above and beyond the call of duty to make sure we had a good flight. Before we landed, the captain made an announcement that we had two very special passengers on board called Oliver and Samuel. This is when the emotions kicked in!

We had someone to help us through the airport where we met our 'Greeter' for 'Give Kids the World' who took us to our car and we set off to 'The Village'.

The 'Give Kids the World' village is set up especially for Wish children. On arrival, the boys were given t-shirts and cuddly toys and we were shown to our villa. There was a basket of snacks waiting for us, more presents and cold drinks in the fridge.

Breakfast was at the Gingerbread House and a total fuss was made of us there too, especially Oliver. He had been given a big badge with his name on and the 'Give Kids' logo. When I tried to take my tray back after breakfast, it was taken from me and I was informed "your job is to eat…".

The 'Village' is fantastic. There is a carousel in the shape of a giant toadstool, a snoring tree, safari theatre for movies twice a day, Amberville houses lots of arcade games, model trains, etc. There is a Dinosaur mini golf, an ice cream parlour with ice cream on tap (Samuel's favourite). The Castle of Miracles was a fun theme and a giant beanstalk grows up the side. Inside, on a Thursday, Father and Mother Christmas are on their thrones to receive the children and give presents. It is somewhere for children to play, it has a wishing well which gives out a noise after each coin is tossed in – like a cow mooing!

There are lots of toys to play with and a slide with strange noises. There is a magic pillow-maker which all Wish children can use. A pillow is made for them while they press buttons to fill it with dreams and miracles (another emotional moment for me) whereas Oliver, totally oblivious to its significance, merrily pressed buttons and waited for his pillow!

Oliver was also given a glass star to write his name on. He had to put it in a box where the star fairy would collect it and place it on the ceiling in the castle, where it will stay forever and ever. Okay, cue the tears again, that really choked me – such a lovely idea and seeing the thousands of stars up on the ceiling…

There is also a Park of Dreams, a pool and water jets for relaxation and play. The whole village is governed by a 6 foot rabbit called Mayor Clayton. He does 'tuck-ins' and came to our villa on our last night to tuck Oliver in to bed. He has a great routine with lots of bouncing on the bed and Oliver was in hysterics.

We could have stayed at the village all week and not gone out, but with complimentary tickets it would be rude not to visit the theme parks! The first day was the most magical, the visit to Magic Kingdom to meet all their favourite characters. The boys were amazed to see so many including Mickey Mouse, Minnie, Pluto, Donald, the list was endless.

Buzz Lightyear was a big hit, and down in a saloon in Frontier Land, we found Woody and Jessie and Bullseye the horse (Toy Story for those who haven't seen it over 50 times!). Oliver's badge was incredible. Jessie spotted it and although there was a big queue, she took Oliver's and Samuel's hands and led them onto the stage for photos and autographs. The same with Mickey. All the characters had a 'human minder' who spotted Oliver's badge and we would be led to the front... "excuse me, we have a special child!" I waited for a parent to demand that their child was special too, but they seemed very tolerant.

Big hugs for Winnie the Pooh, Belle from Beauty and the Beast and so many photos and videos. The parade was fantastic where we could stand still and watch them all pass by.

Day Two saw Mickey and Minnie come to the village for official photos and we found Goofy and Pluto on the way to breakfast! We then went to Animal Kingdom and saw Robin Hood and Jungle Book characters, and the Lion King show where again we had special seating and the boys were taken on stage. Later we had Christmas at the village where the boys were given another present each.

Day Three we went to Universal Studios and Island of Adventures. The boys loved the Shrek 4D film where the chairs moved to make us feel we were moving along in the movie. They were also really pleased to find Shrek outside, and further on, Scooby Doo and Shaggy (big favourites) Crash Bandicoot and the Rugrats.

Over at Islands of Adventures there wasn't much for the boys to do as they were too little for many of the rides, but when a greeter spotted Oliver's badge, over at the Spiderman ride, we were told to go up the road, round the corner and wait, and we would have a private meet and greet with Spiderman. Now if you were 5 or 3, you would know how cool this is! Not only that, Spiderman, unlike so many characters, can talk and he spent about 10 minutes chatting to Oliver and Samuel who were a bit gob-smacked by the whole affair! After a major panic over losing our keys, we found they had been handed in so we made it home.

Day Four we played on the Dino putt and in Amberville games room and after lunch we went to the pool. The boys loved playing with the water jets. I just watched. They were freezing but the pool was a constant 80° so I was fine in there!

Day Five saw Seaworld. Oliver and especially Samuel love sharks and of course, Shamu the Whale was a fantastic show, we managed to avoid getting wet but it was a close call! Went to the pool again after lunch and Oliver didn't want to get out!

Later Oliver signed his star for the castle sky and made his pillow and we made a visit to the ice cream parlour. We managed to limit this to once a day to stop Samuel living on the stuff.

Day Six was MGM Studios where we saw the Muppet 3D Show and met The Incredibles, Mickey, Donald and Daisy and saw Beauty and the Beast show, a lovely show with great music, and Oliver and Samuel were invited afterwards to meet Belle in her beautiful golden gown. The boys were in their element. After Bear in the Big Blue House show, we retired to the pool for a while, and Mayor Clayton had a pool party and barbecue later on with a huge furry Shamu the Whale. Numerous ridges on the carousel were filmed for the new 'Give Kids the World' village video and it was time for bed with Oliver's 'tuck-in' by The Mayor.

Day Seven was our last day which involved checking out, lots of videoing, lots of ice cream, lots of emotion thinking we might never come back, and more emotion when someone told us we could come back and visit, have ice cream, ride the carousel and they would always find Oliver's star on the castle ceiling. I visited their chapel and wrote my thank-yous and hopes on the comments book and thanked them for the tissues they supply next to it!

Our amazing trip was nearly over, and it was a dream which money could never buy. Sure, we could do Disney again, but without a magic badge we would disappear in the crowds.

Make a Wish Foundation and Give Kids the World, and all the companies involved help make special children have special memories and we cannot thank them enough. ■

www.make-a-wish.org.uk

Make-A-Wish Foundation has one simple aim - to grant the wishes of children aged 3 to 18 living with life-threatening illnesses. Since 1986, thanks to Make-A-Wish Foundation, nearly 3,000 very special children have realised their greatest dream and experienced the hope, strength and joy each wish provides. A wish granted is true magic for the child and provides happy memories for all the family and respite from their normal routines of hospitals, doctors and treatment. To find out more phone **01276 24127** or check out their website.

National Institute for Clinical Excellence (NICE)

October 2004

The National Institute for Clinical Excellence (NICE) is part of the NHS. It is the independent organisation responsible for providing national guidance on treatments and care for people using the NHS in England and Wales. Their guidance is intended for healthcare professionals, patients and their carers to help them make decisions about treatment and healthcare. NICE was established as a Special Health Authority in April 1999, to promote clinical excellence and the effective use of resources within the NHS. NICE guidance is developed using the expertise of the NHS and wider healthcare community including NHS staff, healthcare professionals, patients and carers, industry and the academic community.

In October 2004 NICE held a meeting at the Royal College of Physicians to explore the issues of Expensive Therapies for Rare Diseases. There were presentations by the pharmaceutical industry, clinicians, health economists, NICE and the Department of Health. This meeting left one gaping question - What of the views of the Patients, their families and carers and the Patient Associations? The MPS Society and Gaucher Association made representations to NICE expressing concern that the people at the receiving and sharp end, the principal stakeholders, had not had a voice nor that voice heard.

NICE listened and as a result a meeting was held on Monday 7 March 2005, again at the Royal College of Physicians, enabling stakeholders to be heard. The meeting concentrated mainly on the appraisal of new expensive therapies for Ultra Orphan Diseases. Members of the MPS Society were invited to participate and Joanna Wilson spoke eloquently of her experience of Scheie disease and the impact on her life eleven months after starting on Enzyme Replacement Therapy. As Chief Executive I presented the struggles of achieving ERT for Fabry and MPS I patients and concerns over access to ERT treatment when licensed for individuals with MPS II and MPS VI. I begged the question of whether blanket refusals to treat small groups of patients with different ultra orphan diseases due to the very high costs of treating individual patients would amount to eugenics by rare disease.

JOANNA WILSON speaks at the Royal College of Physicians

Meetings

I was born a healthy seven pounds one ounce, lots of dark hair, a very loud cry and everything where it should be and as my parents told me later, their perfect baby.

For the first three years of my life I was progressing well, both physically and mentally as any child should. However, when I reached the age of three, my mother noticed that my hands had started to stiffen and my fingers had started to claw slightly. This concern led my parents to seek advice from our local GP, but nothing came from it. The condition being so slight we were told it would eventually correct itself as I continued to develop. Only a few months down the line, and another problem began to show itself, I had developed an umbilical hernia. Being four years of age, it seemed quite strange to my parents for me to be developing this hernia at this late stage. I was given corrective surgery for my hernia, but no further investigation or action was taken.

Not happy with the lack of diagnosis from any medical source, my parents looked elsewhere for the answer. We were introduced to a surgeon who thought the problem might have been that the bridge of cartilage in my finger was restricting my movement, and so causing the stiffness and clawing. So, he attempted, on my left hand wedding ring finger only, a trigger release, to try to reduce the stress on my finger. This surgical intervention actually ended up causing more harm than good, as after the operation my finger was even stiffer, and the clawing of it was much more substantial. Still no diagnosis, so I was continually passed from doctor to doctor, in the hope that someone would have a clue as to what was going on with my body. Meanwhile I had started to feel a lot of pain in various joints all over my body.

At about the age of seven, I started to get what I described to my parents as ‘sprinkling’, but now I know it was ‘pins and needles’. I felt it in short bursts at first, every week or so, but it then progressed to once every day and then eventually most of the time. It was joined by a dull ache in both wrists and by numbness in the tips of my fingers. About six months later I had carpel tunnel operations, one on each hand, which seemed to help at first as the pain had completely gone, but the pins and needles would come back now and then.

By this stage, both my parents and myself had almost given up hope of ever finding a diagnosis, until we met a doctor called Janet Gardener-Medwin. She took on my case, and spent months researching different areas that it could be in, but still nothing. When she turned round to me and asked if she could use my symptoms

to create a study case for her father's retirement party, where she would show a room full of doctors my case, including photographs of my hands and stomach. It was amazing, the moment she'd finished her talk about me; she asked if anybody in the room had any clue about what my condition might be. At that point, her father and a close colleague of his stood up and both said that I should have a urine sample and blood test to check for the condition called Mucopolysaccharide.

By now I was suffering varying degrees of pain over most areas of my body on most days. Some days I found it so hard to cope, my parents even said that I looked like an old granny trying to get up off the couch. It just wasn't right, I should have been out playing with my friends.

I am now a ten year old, trying to lead a normal ten year's old life; it wasn't easy. I couldn't dry or brush my own hair, it took me several seconds to get up out of a chair because I had become so stiff, I found it increasingly more difficult to participate in school activities, particularly those that needed use of my hands, but at least we could now put a name to it.

I am not sure if that was good or bad, because having joined and sought advice from the MPS Society, my parents and I now knew there was no cure for my condition and that I would continue to get progressively worse and nobody could tell me at what rate and how bad I would get!

However, the good news was that the medical profession were working on and reasonably close to a treatment that would slow down, or dare they say it, halt the progressive deterioration that was taking place in every part of my body.

Sometime before my fifteenth birthday, I heard that trials on the treatment for MPS I had begun in America. You can imagine I was beginning to get just a little excited, I couldn't believe that I may never get worse again. Then in December 1999, trials started in the UK but when I was told I wasn't to be included in those trials I was devastated, particularly when I realised I had to get a lot worse before anybody in the medical profession would help me.

The trials then seemed to go on forever and ever and I thought it would never be available for me. It seemed like eternity. June 2003 and patients in Europe were given the go-a-head for ERT treatment, but still nothing for me, I couldn't believe it!

Never mind, I would get it soon!

At last, in October 2003, I formally applied for funding for ERT from my local PCT, another high and full of expectations only once again to have them dashed. They had refused me – I couldn't believe it.

I was quite seriously in the depths of depression, not a good state of mind for someone due to take their GCSE's with aspirations of studying medicine at University. All I could think of at this stage was a life of disability and a premature death. I was now having increased difficulty walking, increased intolerance to exercise, more pain and lots of tablets to help me. How many seventeen year olds do you know that need their parents to do their hair, dress them, wash them etc? Finally and after another six months with an appeal to the PCT, they agreed to fund my ERT treatment. What a Christmas present, I can't tell you how wonderful I felt when I was given the news, hope at last.

Well here I am, stood in front of you today, eleven months since beginning treatment on Aldurazyme. A period over which I have been able to cram in, in spite of having the weekly infusions, two year's of A level studies and have a real chance of achieving my dream of going to medical school. In fact I have received an offer from Southampton.

How many NICE appraisals can measure my transformed quality of life both physically & mentally?

Life without constant pain, able to put my arms above my head and brush my own hair, climb the stairs without being constantly out of breath, complete a full day at school every school day and have enough energy left over to have a little social life. You would need to be my shadow to know just what a difference ERT for MPS I has made to my life.

My biggest fear now is that you may take it away from me! ■

Joanna Wilson and Sir Michael Rawlins, Chair of NICE

Ann Green, MPS Society Trustee, gives her views on the NICE Meeting...

On 7 March 2005, five people from the Society attended an important national conference at the Royal College of Physicians in London, the outcome of which may help determine whether the Department of Health continues to give its support to the funding of costly drugs for patients with diseases such as MPS.

The Conference was called 'How can NICE evaluate expensive drugs to satisfy the needs of patients with ultra-orphan diseases?' It was hosted by the National Institute for Clinical Excellence (NICE) and the Royal College of Physicians and it is testimony to the importance that these two organisations place on the subject in that the conference was introduced by Professor Carol Black, Chair of the Royal College of Physicians, and closed by Professor Sir Michael Rawlins, Chair of NICE.

In essence the event was a patient-led meeting to discuss the issues relating to how best to evaluate expensive drugs to satisfy the needs of patients with rare diseases. There were approximately 50 attendees, including two MPs, one of whom suffers from Gaucher's disease, and representatives from the Royal College of Paediatrics and Child Health, the Gauchers Association, the MPS Society and the Genetic Interest Group.

The first speaker, Professor David Barnett explained how NICE was structured and how it evaluated the drugs that were presented to them. Assessments are made, not exclusively on clinical or cost effectiveness, but also on the extension of life, versus the quality of life, by so-called 'QALYS'.

There followed a number of presentations by professional speakers, as well as a perspective on Gauchers disease by Susan Lewis, the Gauchers Association's Chief Executive, who is herself a sufferer from the disease.

The afternoon was very much led by people from the MPS Society. Christine Lavery spoke on the challenge of obtaining treatment for Fabry and MPS sufferers. Joanna Wilson followed and gave her own personal experience of the pain and heartbreak of suffering with an MPS disease and the struggle she and her family had experienced in order for her to receive treatment. Alison Millar, a Solicitor who regularly works with MPS sufferers, outlined the legal perspectives and the legal responsibilities towards patients with rare diseases.

The general feeling at the conclusion of the conference was that the needs of patients with very rare diseases were being acknowledged and it was stated that the key issues which were discussed at the meeting would form a report which would be included in NICE's guidance to the Department of Health. In addition, it was apparent to all present, that the Societies represented were strong and vociferous and would not hesitate to lobby and protest if their needs were not met or their voices heard.

Fabry Patient's Meeting by Sophie Denham

On Friday 11 February 2005, I was fortunate to be able to attend the Fabry patient's meeting, a meeting for patients and families with Fabry disease.

The meeting was held at the Sidney Sussex College in Cambridge and was a meeting organised jointly between Cambridge and the Royal Free Hospital, London. The location of the meeting was magnificent and afforded an opportunity to many of the professionals to share their knowledge of the college and the famous people who attended there.

After a brief history lesson we went forward with the programme for the day which offered us the opportunity to hear presentations from medical experts on Fabry Disease and learn about the latest developments and research. The programme was very comprehensive and included talks on NSCAG, FOS, Fabry Disease in children and Neuroradiological aspects of Fabry Disease.

My role at the meeting was to represent and promote the MPS Society to individuals and families and to be there to answer any questions that may arise. I was also there to be a part of the expert panel, although after being identified as a member of the MPS Society during the presentations, I was asked to explain the role of the Society and the support given to individuals, families and professionals.

This was my first time having to speak publicly to a room full of people and totally unprepared, something that was extremely exhilarating, scary and for those that know me, not like me at all. I'm very glad that I have done it though, as being a part of the expert panel at the end was not so daunting. It also provided the opportunity for individuals and families to know who I was and many who were not members then, are members now.

I would just like to say a big thank you to all those at Addenbrookes and the Royal Free for making me feel so welcome and it was a real honour to be a part of a very successful meeting. It was also wonderful to meet so many individuals at the meeting and to receive many of the completed membership forms given to those who were not already a member of the Society.

Second Symposium on Lysosomal Storage Disorders, advancing the understanding of lysosomal disorders

On 2 March, I was fortunate to attend the second symposium on Lysosomal Storage Disorders (LSD's) held in Athens. It was a 3 day symposium, which had a full and detailed programme, and professionals from around the world attended.

Unfortunately I did not arrive until late, due to delays in taking off from Heathrow. However, this did give me the opportunity to write numerous articles for this newsletter, as you will see. On arrival at Athens I was met by a hostess, who took me straight to the Metropolitan Hotel, where the conference was being held. Due to the two hour time difference and delay there was no time to freshen up before we were ushered into the dining room for what was the start of many banquets to come. After this, I retreated to my room for some much needed rest and relaxation, ready for the next day.

Thursday 3 March 2005

Although I had an early night, the 8.30am start (6.30am UK time) was a shock to the system and I needed a few cups of coffee before I felt able to take on the information for the day. The conference programme was held in the Auditorium Hall at the new digital planetarium, adjacent to the hotel.

Programmes were delivered from professionals across the world, looking particularly at Gaucher disease, Fabry disease, MPS I disease and Pompe disease. The morning saw presentations looking at early diagnosis in LSD's, cardiac manifestations of Fabry disease, new born screening, poster presentations and a new topic for many on neurological involvements across LSD's. This was presented by Dr Ashok Vellodi and for me it was extremely exciting to hear what could be achieved if they could break through the blood brain barrier.

After a small feast for lunch, the programme continued for a short period before people went off to join the workshop of their choice. There were seven in total and the one I opted for was Management of patients with MPS I disease.

After the workshops we were given the opportunity to see some of the sights of Athens (this was really essential as the evening dinner was being held after the social event and the coaches were departing from there). It was nice to be able to see some of the sights of Athens and to be able to spend time in the sunshine. We were taken to see the Acropolis and had a tour guide to give us some of the history as we walked around. After the tour we departed to a museum of contemporary and modern art by local people. This was where the dinner was being held.

The food was superb and plentiful and the company great as was the entertainment. We even got up and had a go at Greek dancing, which was very comical. After the entertainment finished we were taken back to the hotel.

Friday 4 March 2005

The following day was another full programme for the morning, with departures for the airport from lunchtime onwards.

I had a great time at Athens and met some great people, as well as promoting the Society and seeing how other professionals from different professions and countries support individuals with LSD's.

Thank you for this opportunity and I hope to see those who I met at future events.

by Sophie Denham

Update on the Children in India with Morquio who survived the Indian Ocean Tsunami

In the last newsletter, Winter 2004, we published a letter from Elizabeth Herridge, who first informed the MPS Society about the three children with Morquio Disease living in Chennai, two of whom Dr Uma Ramaswami and I visited when on holiday in India in January 2004. In her letter Elizabeth Herridge told how all the three children and their parents survived the Indian Ocean Tsunami.

Elizabeth has sent some recent photographs of Marimuthu, Radhika and Manikandan taken at Christmas. Marimuthu was interviewed on Christmas Day and asked what Christmas felt like to which he replied it gave him a warm feeling in his heart.

The children at the Cheshire home where Marimuthu, Radhika and Manikandan live spent Christmas day out on a treat sponsored by the Cookie Man in Chennai. Here is just one photograph of them enjoying Christmas.

Now they have their lives back

Pioneering treatment brings hope to sisters with incurable illness

Looking at them laughing and playing together, it is hard to imagine how much Terri and Jessie Hambly have been through in their short lives. Both girls were born with a rare genetic condition that has caused them excruciating pain and affected their sight and hearing. But thanks to revolutionary new treatment, they have now been offered fresh hope.

Seven years ago Terri, 12, was diagnosed with the incurable Hurler-Scheie Disease, and Jessie, six, was also born with the condition, which lets waste build up in the body. It is caused by faulty genes, which stop the body producing enzymes which break down the waste.

For their father Paul, of Bransholme, north Hull, it was a devastating blow. 'At first I did not believe it because it means they will go blind, deaf and crippled in every joint,' he said. 'It took a while to sink in, but this is what life has given us and you have to plod on.' They were not expected to live beyond their 20s, and a year ago Terri was in so much pain she just wanted to sit on the couch and watch television.

But Mr Hambly, 35, who gave up his job as fire safety officer to care for the girls, found out about a pioneering trial for Enzyme Replacement Therapy.

The family now travels to the Royal Manchester Children's Hospital once a week to take part in trials of a pioneering treatment, made possible by a grant by the Jeans for Genes charity - and their symptoms have improved dramatically.

It took all of the Mail photographer's persuasive powers to get them to sit still long enough to take their picture, and they list 'arguing with my sister' as one of their hobbies. 'They are just two bundles of energy and it is like living with two different girls,' said Mr Hambly. 'They have been given their lives back.'

Terri, who goes to Endeavour High School in Beverley Road, Hull, has also been helped by having part of her eye transplanted and Mr Hambly believes that without the treatments, she would no longer be in mainstream education.

Jessie is at Wheeler Primary School in Wheeler Street, Hull. Now Mr Hambly and his partner Helen Evans, 29, are offering their support to Jeans for Genes day on Friday, supported this year by pop band Blue. And for the charity's annual fundraising day, it is urging people to wear jeans for the day and donate money to the cause.

For more details, call 0800 980 4800 or visit www.jeansforgenes.com.

For an update on the Jeans for Genes campaign check out the MPS Fundraising newsletter.

Enzyme Replacement Therapy Trial

Sam and Jo Dacey

Sam and I would like to tell you about the ERT trial and how we feel it is helping…

Sam started the trial last January, and after the initial base line readings of height, weight, liver size and various occupational therapy and physio tests, we then met the nurse from the hospital running the trial.

Sam and I are very lucky as we only have to travel 12 miles for the treatment. Although we do have to travel to Manchester every 18 weeks to repeat all the initial tests and monitor any changes. Always good fun!

The trial is double blind which means that no one knows who is receiving the treatment. All we know is that the control group is split into three. One third of the boys will get the drug every week, one third every other week and one third will receive the placebo.

Obviously we don't know for sure what Sam is getting but we do know one thing, that there have been some changes.

Since January Sam has grown at least 4cm which is the same as he grew over the previous two years. His liver size has decreased and his pancreas is no longer palpable. So he can now choose between having an 'inny' or an 'outy' and do tricks with it!

The pictures on this page show Sam, at different ages and at school in July this year and we can see a difference in his features. As you can see from the pictures, he is happy and active and beat at least three of his classmates in the relay race on sports day! Although these are only small changes (and photographs can vary depending on the lighting etc.), I think what is really significant is that he hasn't deteriorated in any way.

The MPS Society's motto is 'Care Today, Hope Tomorrow'. For our Hunter boys I really believe that their tomorrow has come!

I really hope this will give encouragement and hope to all the rest of the Hunter boys and their families who are waiting for treatment.

To date, Sam has grown 5cm in one year and his liver and spleen have reduced in size by 7 and 8 cm respectively. I will report back further after May as Sam would have been on treatment for 4 months.

Genzyme Welcomes AWMSG Decision to Endorse the use of Aldurazyme® (Laronidase) for the Treatment of MPS I

Oxford, 16th March 2005… Genzyme Ltd today welcomed the decision of the All Wales Medicines Strategy Group (AWMSG) to endorse the use of Aldurazyme® (Laronidase) for the treatment of the rare genetic disease Mucopolysaccharidosis I (MPS I) www.wales.nhs.uk/awmsg.

Julie Kelly, Senior Director, Genzyme Specialty Therapeutics, Oxford said: 'MPS I is a devastating disease and can often be fatal. Early diagnosis and treatment of MPS I patients is critical and may help prevent irreversible damage. We are delighted that patients in Wales will now have access to therapy.'

MPS I is an inherited genetic disorder, which affects an enzyme called alpha-L-iduronidase. This enzyme is essential in breaking down long sugar molecules produced by cells. In MPS I, these sugar molecules accumulate in virtually all organs of the body to cause progressive and debilitating disease. Symptoms include enlargements of the liver and spleen and abnormally shaped bones. Hearing, vision, respiratory and cardiovascular functions are all affected and joint mobility is typically severely impaired.

Aldurazyme is currently the only treatment approved for MPS I in Europe and the US. Aldurazyme provides the body with a source of the missing enzyme alpha-L-iduronidase. MPS I is thought to affect 3,000 to 4,000 people worldwide.

Genzyme Ltd is the UK division of Genzyme Corporation, one of the world's leading biotechnology companies. Genzyme is dedicated to making a major positive impact on the lives of people with serious diseases. Founded in 1981, Genzyme has grown from a small start-up to a diversified enterprise with annual revenues exceeding $2 billion and nearly 7,000 employees in locations spanning the globe. With many established products and services helping patients in more than 80 countries, Genzyme is a leader in the effort to develop and apply the most advanced technologies in the life sciences. The company's products and services are focused on rare inherited disorders, kidney disease, orthopaedics, cancer, transplant and immune diseases, and diagnostic testing. Genzyme's commitment to innovation continues today with a substantial development program focused on these fields, as well as heart disease and other areas of unmet medical need.

For further information contact:
Citigate Dewe Rogerson
Chris Gardner
+44 (0)20 7638 9571

Genzyme Specialty Therapeutics
Malcolm Johnson/Julie Kelly
+44 (0)1865 405200

Enzyme Replacement Therapy and Clinical Trial Update

MPS I

Aldurazyme is administered once-weekly and has been approved in the European Union for long term enzyme replacement therapy in patients with a confirmed diagnosis of MPS I, to treat the non-neurological manifestations of the disease. As the first orphan drug approved for MPS I in the European Union, Aldurazyme has been granted ten years of market exclusivity. In 2004 some MPS I patients in the United States of America started to receive their once-weekly infusion at home. This has initiated considerable interest by UK MPS I ERT patients having to travel long distances to hospital and spending many hours in a hospital environment. The difficulty is that Aldurazyme was licensed in the European Union for hospital use only. The MPS Society is in dialogue with Genzyme Corporation to see if it is time now for the license to be expanded to include home treatment.

MPS II

Transkaryotic Therapies Inc (TKT) released research findings on 28 October 2004 at the American Society of Human Genetics 54th Annual Meeting held in Toronto, Canada, evaluating intrathecal delivery of iduronate-2-sulfatase (12S) in an animal model. The results, presented by Dr Justin Lamsa of TKT showed that repeated injections of 12S were well-tolerated and resulted in the accumulation of enzyme in various cells of the central nervous system (CNS). 'We are encouraged with these early research findings and intend to advance this programme further to better understand optimal dosing and methods to deliver enzyme into the central nervous system. Since many patients with Hunter disease will develop central nervous system involvement, we are committed to finding a way to safely administer this missing enzyme to the brain to compliment our product currently under clinical trial for non-CNS aspects of Hunter disease' stated Michael Heartlein, TKT's Vice President of Research.

MPS III

TKT, as part of their research to evaluate new approaches to the problem of treatment of the CNS, is hoping to move their MPS IIIA programme forward. If the trial to directly administer the enzyme into the CNS of individuals with MPS II is successful, they hope to expand their research initiatives to include MPS III A.

MPS IV

There are currently no programmes in place for developing treatment options for MPS IV. Breaking news is that the National MPS Society, with whom we collaborate closely is currently looking at funding interesting research into bone and MPS IV. We will give you more information on this as soon as it is available.

MPS VI

BioMarin Pharmaceutical Inc. announced 1 February 2005 that the US Food and Drug Administration (FDA) has accepted for filing and assigned six-month review to the Biologics License Application (BLA) for rhASB (galsulfase) the company's enzyme replacement therapy for MPS VI. A six-month review is typically granted to drugs that, if approved, would be a significant improvement in the safety or effectiveness of the treatment, diagnosis or prevention of a serious or life-threatening disease.

Mapping of MPS III C locus

MPS III C is the only mucopolysaccharide disease for which the gene code the deficient enzyme, acetyl coA:alpha-glycosaminide acetyltransferase (GNAT) has not been identified. Dr Pshezhetsky from Montreal, Canada, and his collaborators report their attempt to map the locus (the position where the gene occupies on a chromosome) for MPS III C. DNA was obtained from 44 patients and 18 unaffected parents and siblings from 31 families, in addition to 38 controls. Looking at regions of similarity on the chromosomes of individuals with MPS III C, the group identified the locus to be in the pericentromeric region (around the area that divides the chromosome into two arms) of chromosome 8. Linkage analysis was performed to support the informal analysis.

This interval spans 8.3 cM (centimorgans: unit of genetic distance on the genetic map) and contains 72 identified genes and open reading frames. These data will be used for identification of the gene coding for GNAT.

Exerpt from 'Localisation of a Gene for MPS III C to the Pericentrometric Region of Chromosome 8' by J Ausseil, JC Loredo-Osti, A Verner, C Darmond-Zwaig, I Maire, B Poorthuis, OP van Diggelen, TJ Hudson, TM Fujiwara, K Morgan, AV Pshezhetsky. (J. Med. Genet 2004; 41:941-944. doi:10.1136/jmg.2004.021501)

2005 MPS Grants To Research

To date a staggering £3,039,304 million gross has been raised from the 2004 Jeans for Genes Appeal to be shared between the four partner and four guest charities. In addition the MPS Society is the beneficiary of the 'Ollie G' Ball held in June 2004 of which at least £60,000 is designated for research into MPS diseases. £7,600 raised by Marina Foster who runs a charity shop is also supporting research into Sanfilippo Disease.

Development of Gene Expression

Targeted Isoflavone Therapy (GET IT) for Mucopolysaccharidosis Type III (Sanfilippo disease)

Lead Investigator	Professor Grzegorz Wegrzyn	University of Gdansk Poland
Co-Investigator	Dr Ed Wraith	Royal Manchester Children's Hospital

Lay Abstract

Mucopolysaccharidosis Type III, MPS III, Sanfilippo Disease, is a severe, progressive neurodegenerative disorder caused by four different enzyme deficiencies (MPS Types III A, B, C and D) involved in the metabolic breakdown of the glycosaminoglycan (GAG) heparan sulphate.

Currently, there is no effective treatment available for MPS III. Moreover, although enzyme replacement therapy (ERT) has recently been available for MPS I, and clinical trials are ongoing in ERT for MPS II and MPS VI, it is little chance that this kind of treatment may be effective for MPS III. This is due to severe neurological problems in MPS III and inefficient delivery of proteins, including those used in ERT, to central nervous system, because of the blood brain barrier.

Therefore, we aim to develop an alternative therapy for MPS III. We propose to consider a substrate deprivation therapy based on regulation of expression of genes coding for specific GAG synthetases, which is called by us 'gene expression-targeted isoflavone therapy' (GET IT). The etymology of this name is based on our finding that genistein, a naturally occurring isoflavone, inhibits synthesis of GAGs up to ten times in cultures of fibroblasts of MPS patients. Since the optimal genistein concentration, which blocked GAG synthesis effectively in cell cultures, was relatively low, this isoflavone may potentially be an effective drug for the treatment of MPS III. As a small molecule, it may potentially cross the blood-brain barrier. Nevertheless, we aim to construct a more lipophilic derivative that would cross the barrier even more efficiently and retain the ability to inhibit GAG synthesis.

Gene expression experiments are planned to learn more about the details of the mechanism(s) of genistein-mediated depression of GAG synthesis, with special emphasis on expression of genes coding for GAG synthetases and hydrolases. Finally, a mice model for MPS III B, which is currently available in one of collaborating laboratories, will be used to test a therapeutic potential of genistein and its derivatives in the Sanfilippo syndrome. If it is effective indeed, one might assume that GET IT can be effective not only in treatment of MPS III patients, but also of those with other MPS types in which central nervous system is affected.

Grant Awarded £160,000 over 3 years
Year 1 will be funded from the proceeds of the 'Ollie G Ball' organised by David Gosling of Country Wide Special Events
Years 2 & 3 will be funded from the Jeans for Genes Appeal & the proceeds of the Charity Shop

Details of further Research and Support Grants
will be given in the Summer 2005 MPS Newsletter

Therapeutic Targeting of Lysosomal Storage Glycoforms in Hunter and Fabry Disease

Lead Investigator Professor Timothy Cox University of Cambridge
Co-Investigator Dr Patrick Deegan University of Cambridge

Lay Abstract

There are two lysosomal storage disorders caused by genetic defects in one of the sex chromosomes; Hunter syndrome and Fabry disease. Differences between these two conditions in disease severity in females lie at the core of this proposal, which offers therapeutic benefit in several lysosomal storage disorders. If we were able to determine why women harbouring one copy of a defective gene for Hunter syndrome almost never develop the condition, whereas women carrying a single Fabry disease gene nearly always suffer from this disorder, we would be in a position greatly to enhance the action of treatments already available for patients with many lysosomal diseases.

Enzyme replacement therapy depends on the ability of cells to take up enzyme from the surrounding fluid and target it to the lysosome, where it has biological effect. Cells producing adequate amounts of enzyme usually export a proportion in the surrounding fluid. Every normal female is a mosaic; in some of her tissues maternal genes are active, while in others, paternal genes are active. Thus females with sex-linked conditions are made up of patches of tissue with normal production of enzyme and patches with impaired production. In Fabry disease and Hunter syndrome, females carrying the defective gene usually have detectable enzyme in body fluids. In Hunter syndrome, this enzyme appears to be taken up by the defective cells thus preventing expression of the disease, through a 'natural' enzyme replacement therapy. This process does not appear to work in Fabry disease, permitting the accumulation of storage material in affected tissues with resulting features of the disease in most females. This is all the more intriguing given that the pharmaceutical enzyme, supposedly similar in structure to the natural enzyme, clears storage and provides clinical benefit to female patients.

This research project will examine aspects of the structure and the distribution of the relevant enzyme in Fabry disease and Hunter syndrome. We will examine the question of whether the natural Fabry enzyme is modified in the circulation in a way that prevents its uptake into deficient cells. The uptake of natural, therapeutic and artificially-modified enzyme in diverse relevant cell-types will be explored. On the basis of this research, infusion of radiolabelled enzyme in patients will allow us to determine for the first time whether the structure of the enzyme can be optimised to permit uptake of the natural enzyme in females with Fabry disease and permit the development of enzyme treatments that are more effectively targeted to diseased tissue in a broad range of lysosomal storage diseases.

Grant Awarded £231,635
Years 1, 2 & 3 will be funded from the Jeans for Genes Appeal

Involvement of the Blood Brain Barrier in MPS III A and III B

Lead Investigator Professor Bryan Winchester Institute of Child Health, London
Co-Investigator Dr David Begley King's College, London

Lay Abstract

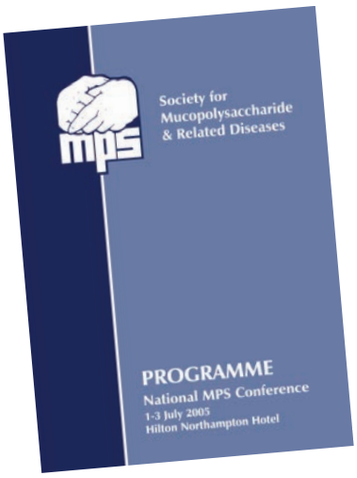
One of the most exciting developments in MPS recently has been the development of enzyme replacement therapy (ERT). It is now possible to treat some MPS disorders by infusing the 'missing' enzyme into the blood stream. The infused enzyme circulates the body modifying many aspects of the disease such as reducing joint stiffness. However, the enzyme does not enter the brain. This means that those MPS disorders with brain involvement such as Sanfilippo syndrome cannot be treated with ERT.

The difficulty of getting enzyme into the brain is due to the blood-brain barrier (BBB). This barrier exists to protect the brain from damage to circulating substances. In order to progress with ERT treatment for the brain, we need to understand more of the nature of the blood-brain barrier in the MPS disorders. This has not been previously studied. Our plan is to investigate this in a step-wise manner to answer these research questions:

Is the BBB normal or affected by MPS deposits?
Does ERT change the nature of the blood brain barrier?
If the BBB is normal how can we modify it or alter the ERT to allow brain entry?

Fortunately, we have mouse models of MPS IIIA and IIIB that we can use for this important research.

Grant Awarded £45,959 over 1 year
This research will be funded through a generous donation from the Shauna Gosling Trust



National MPS Conference

1-3 July 2005

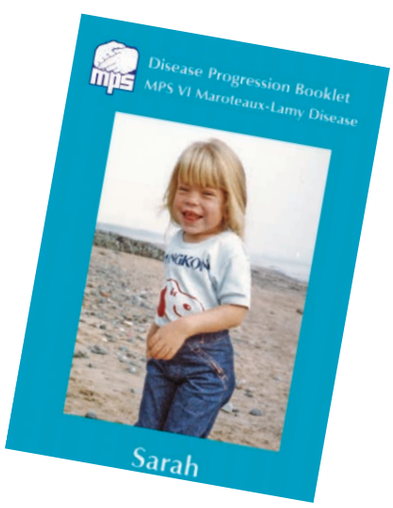
Hilton Hotel Northampton

The conference is almost full to capacity. However, there are still some single day conference packages available, particularly for delegates wanting to attend Conference C on Saturday 2 July 2005. This conference will provide the latest news on management and treatment of Fabry Disease.

Download a programme from our website or contact us now for more information and to book a place.

MPS VI Progression Booklet

A progression booklet for MPS VI, Maroteaux-Lamy Disease is being developed and will be available shortly. This booklet is primarily aimed at professionals working with children and adults affected by the disease. Through photographs of the same child at different ages, the booklet demonstrates the progressive nature of this disease. Check out our website or phone for more details.



MPS Awareness Bands

Silicone wristbands are the latest 'must-haves' and are in high demand all over the world. They are a trendy way to show your support to the MPS Society. Our blue 'Care Today, Hope Tomorrow' wristbands are available at £2 each or 5 for £8.

For more information on how to order your bands contact fundraising@mpssociety.co.uk or phone us now.

New Children's Publication!

Written for children with Hurler Disease, MPS I, these booklets are designed for children aged 5 years and over.

This colourful booklet uses large print to give age-appropriate information about bone marrow transplantation for MPS I. This is an invaluable tool for parents, siblings and teachers for use in schools to support children in understanding more about BMT.

To order your copy complete an order form enclosed, download one from our website or phone us now.

www.mpssociety.co.uk
01494 434156

A Chance To Live

The Society is delighted to be working with Richard Dunn, grandfather of Isaac who has MPS I and has undergone a Bone Marrow Transplant, on a second edition of his book 'A Chance to Live'. This book, introduced here, will be published shortly so check our website for availability. www.mpssociety.co.uk

I'm standing in the staff room and 150 pairs of eyes are fixed on me. It's the end of term and I know if I try to continue what I'm saying, I will begin to cry. It's been a difficult year. Living at home on my own as Kay works with Lou and Adam to save Isaac's life. Watching and feeling their despondency when the first bone marrow transplant had failed. Not having time to visit Sam, Harvey and Orlan and only talking to my mother on the phone.

I've had no one to talk to. At school it is always business as usual. By keeping everything brisk and formal I keep the emotions at arm's length. When you are the one in charge and everyone relies on you it's a lonely existence. Underneath the surface I feel near to tears but outwardly I have to appear in control, unflappable.

The one person who has supported me through this nightmare is Doreen my P.A. She asks about Isaac and listens intently when I need to talk. She relays my news on to those who are concerned but don't like to ask me directly. She has invited me to dinner with her and Ken and provided me with an oasis of calm and pleasantries.

So here I stand, unable to speak. I was doing so well until now. I've just thanked the staff for their efforts and successes. All I have left to do is thank Doreen but this is when my two worlds collide and the burning emotions of the man invade the persona of the Head Teacher.

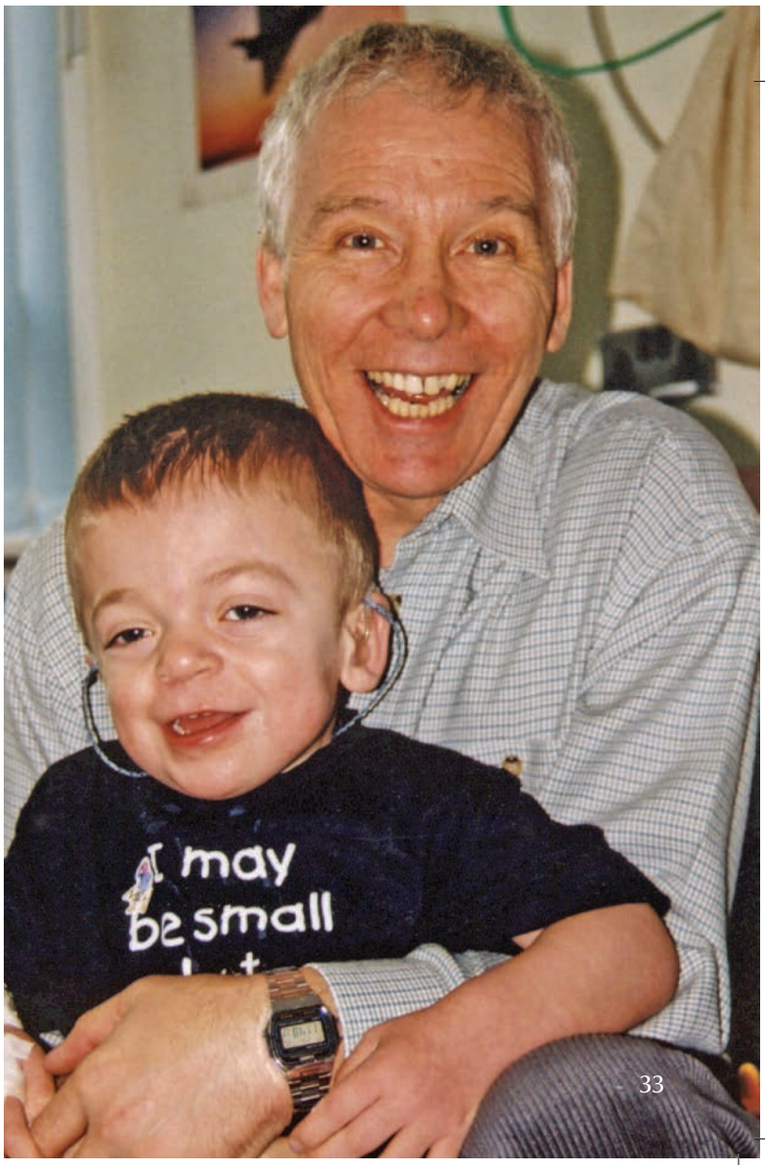
Everyone waits. I see eyes fixed on me, full of questions and concern. One two, three, the seconds tick by in this silent room. I choke back the emotions and try to compose myself. Finally, I thank Doreen and I'm ready to continue. I move back onto safer ground.

'We have the Ofsted inspection next term. I want to finish by saying that I have every faith in all of you. The school will get an outstanding report because you've earned it. The inspectors will be here for a few days and then we won't see them again. This is our school and our children. What we think of each other is more important than any report. Go home and have a relaxing holiday and next term we'll show these people just how good we are.'

I'm in role again, Head Teacher not grandfather. My colleagues stand and applaud. Maybe for a moment, without the need for words, they have shared the duality of my life. Tomorrow the Head Teacher will post the Ofsted forms and then the grandfather will drive to the children's hospital to spend his summer holidays waiting by the window of the bone marrow unit, in case he is needed.

After my grandson was diagnosed with MPS I my life and that of my family was torn apart. We all had to deal with raw emotions and thoughts almost too intense to bear. To lift ourselves time and again from setbacks and exhaustion: to find energy where none existed and hope at dark moments.

It is said that everyone has at least one book in them. When I retired I felt compelled to write about the pain and the courage shown by members of my family, as they struggled to save Isaac's life. I wrote about isolated episodes that had impacted on my life and feelings, as a way of releasing buried emotions. Slowly these disparate parts began to piece together like a jigsaw and 'A Chance to Live' became an account that I needed to share with the rest of the world. This is a story, seen through a grandfather's eyes, of one family's fight to save a young boy's life. This is Isaac's story.



Integrated Multi-agency Care Pathways for Children

with Life-threatening and Life-limiting Conditions

Palliative Care for Children?

As the need for palliative care has become increasingly acknowledged it has become important to define what is meant by the term. A definition promoted by ACT, the Association for Children with Life-Limiting or Terminal Conditions and their Families says:

'Palliative care is an active and total approach to care, embracing physical, emotional, social and spiritual elements. It focuses on enhancement of quality of life for the child and support for the family and includes the management of distressing symptoms, provision of respite, and care following death and bereavement. It is provided for children for whom curative treatment is no longer an option and may extend over many years.'

ACT/RCPCH 1st Edition 1997,
Development of Children's Palliative Care Services

What is a Care Pathway?

A care pathway framework and guidance has been developed by ACT (2004) in acknowledgement of the many and complex needs families have in caring for children with life-limiting or life-threatening conditions. This was funded by the Department of Health (England). These pathways can be a means of engaging with the child's and family's needs, which can be used to ensure that all pieces of the jigsaw are in place, so that families have access to the appropriate support at the appropriate time. The pathway aims to link children and families with community services, hospital based services, social services, education and the voluntary sector in one joined up planning process.

The document from ACT provides a broad generic template for developing an Integrated Care Pathway for children diagnosed or recognised as having a life-threatening or life-limiting condition. In this document, an Integrated Care Pathway is proposed as a broad outline of the key events that happen, or should happen, in the journey made by children and families.

The three key events in the child and family's journey include:

Diagnosis or recognition of a life-limiting condition

Ongoing care that may last for weeks, months, or years

End of the child's or young person's life, including **bereavement care**

The pathway will be guided by five essential standards. They represent the weakest points for many families in their patterns of care, points at which there are difficulties with communication and integrated working between professionals.

The five standards are:

The prognosis - breaking the bad news

Transfer and liaison between hospital and community services

Multi-disciplinary assessment of needs

Child and family care plan

End of life plan

The document will be of use to those striving to improve the provision and consistency of care and support to children and families. Families and children may find it helpful in the process of accessing support and information along their own individual journey and professionals should find it helpful in their aim to deliver integrated services meeting individual needs.

For a copy of this document, 'Framework for the Development of Integrated Multi-Agency Care Pathways for Life-Limited and Life-Threatened Children' (2004) please contact ACT.

0117 922 1556
info@act.org.uk

ACT
Promoting palliative care for children

INFORMATION EXCHANGE

The Benefits of Health Supplements

Louise Lewis shares her family's experience

I am just writing to share our experience of using a health supplement, for our daughter, Georgia, who has Sanfilippo Disease, Type A.

Since May 2004, we have been giving Georgia Equazen 'Eye Q' liquid. This is a combination of omega fish oils, and it is intended to improve brain function and concentration. We started giving it to her at a time when she went through a period of not sleeping (something which I am sure many of you can relate to!) as a friend of mine recommended it, saying it changed her little boy's behaviour quite radically (he suffered from hyperactivity and was obviously not sleeping). To be honest, at the time, we were totally exhausted and I thought anything was worth a try!

Within just three weeks, Georgia was sleeping all night and her school teachers commented on how much more aware she was and how much her eye contact and concentration had improved. They weren't aware that we were giving the 'Eye Q' liquid to Georgia! Also, at the time, family and friends commented that they had noticed a real difference in Georgia and recently somebody made a comment about how shiny her hair was! For anyone that has a child with Sanfilippo Disease you'll know that they can often have coarse hair, so maybe the fish oils have made a difference!

In the eight months that Georgia has been taking this supplement she has had only one cold, therefore just a brief period of her being unwell. I am convinced it is due to the fish oils (unless someone proves me wrong!), as normally Georgia would pick up all the bugs going!

The oil should be given daily and it is available from Boots or most health shops. It can be expensive (£9.99 per bottle), but we definitely think it is worth every penny, for the difference it has made to Georgia. I appreciate that this supplement is not a 'miracle cure' and may not suit everybody, but we just wanted to share our own personal experience.

The photograph below was taken in Minnie Mouse's house during our trip to Florida with the MPS Society. Georgia's eye contact never ceases to amaze me, it's magical!

Carers Break Vouchers

In England and Wales the Carers and Disabled Children Act 2000 contains provisions enabling (not requiring) Local Authorities to give vouchers to carers. Carers are defined as people caring for elderly, disabled adults and/or family members in their own home.

Carer Break Voucher Schemes are designed to provide a short-term break for people with long-term illnesses or disabilities. They can also give parents a break by providing care for a child if suitable, either with another family, or in a children's home. Care is provided for a specified period of time and may be within a residential home, a respite care service or within the person's own home.

A carer needs to be able to take a break from the responsibility of supervising or caring. A break needs to be long enough for them to be able to participate in the activity that will help to support them in their caring role. In some instances that could be as short as a couple of hours, or it could be overnight, or longer.

Care Breaks are mainly given to carers to take a break from looking after someone who needs a substantial amount of care or supervision.

The Voucher Scheme can only be used with approved service providers. It allows a carer to arrange the dates for an approved amount of 'respite'.

Vouchers will be issued on instruction from the Local Authority following a carer's assessment.

The value of the voucher will be expressed in money and indicate the type of care to be provided e.g. domiciliary care.

Vouchers can only be used with service providers that are approved or can apply to be approved with the Care Standards Commission.

The Local Authority will hold and maintain a list of service providers who wish to participate in the scheme.

NEW RARE DISEASES WEBSITE

Wellchild, the national charity working in the field of child health, has been very supportive of the many issues associated with rare disorders and has recently been awarded a grant from the Lincoln Group to set up a dedicated website for rare disorders. The aims of the website will be to:

Establish a **forum** for information exchange for individuals and groups involved in rare disorders

Promote **discussion** about current policy and major issues relating to rare disorders

Collect and collate UK wide **statistics** on rare disorders as part of a rare disorder mapping project

An independent advisory group made up of representatives from UK rare disorder support groups will work with Wellchild to oversee the development of the website.

New Carers (Equal Oppportunities) Act 2004

This legislation referred to in the last MPS newsletter when the Bill was awaiting approval by the House of Lords, has been passed and is now law.

This Act extends the duties of local authorities as detailed in the Carers and Disabled Children Act 2000. The Carers (Equal Opportunities) Act 2004, places duties on local authorities and health bodies in England and Wales in respect of carers. These include the duty to inform carers of their right to an assessment of their ability to provide and continue to provide care. This assessment must include consideration of whether the carer works or wishes to work and/or is undertaking or wishes to undertake education, training or any leisure activity.

The legislation also provides for co-operation between local authorities and other bodies, including housing, education and health in relation to the planning and provision of community care services that are relevant to carers.

Parental Leave Entitlement

The entitlement to parental leave is now available to parents of all disabled children (who receive DLA) while the children are under 18. This clarifies that if you have a child who is under 18 you will get parental leave (as long as you meet the service and notice requirements). It does not matter whether your child was under 5 at 15 December 1999 (this was previously unclear). If you have any queries please contact the Working Families helpline on the freephone number **0800 013 0313**.

Motability

Motability was set up by the government in 1977 and is a national charity which helps provide transport to people with disabilities. Before the introduction of the Mobility Allowance in 1976, only people with a disability who could drive themselves got government help with personal transport. Motability gives help regardless of the ability to drive.

How does the Motability Scheme work?

The Motability Scheme works by enabling people with a disability (and parents of a child with a disability) to use their disability benefit to lease or purchase a car, powered wheelchair or scooter, through one of three main schemes:

A three year Contract Hire Scheme to lease a new car

A Hire Purchase Scheme to buy a new or used car, over two to five years

A Hire Purchase Scheme to buy a powered wheelchair or scooter over one to three years.

Who can apply?

Applicants to the Motability Scheme must be in receipt of the high rate mobility component of Disability Living Allowance and the benefit award must be long enough to cover the hire period. Children over the age of three and other people on these allowances who do not drive, can apply for cars as passengers. Customers pay over part or all of their allowance to Motability for the period of the contract hire or hire purchase agreement. On some cars, larger and more expensive cars, an advance payment may also be required.

These schemes are administered on a contract basis by Motability Operations, a non-profit company, who run the car schemes and route2 mobility, who run the wheelchair and scooter scheme.

Contract Hire

This scheme offers a new car on a three year lease. All maintenance and servicing costs are included, together with comprehensive insurance and breakdown cover. At the end of the lease the customer hands back the car and the process can be started again.

Hire Purchase

This scheme enables a customer to buy their own car. This may be preferred when specialist adaptations are needed, which under the Contract Hire Scheme may need re-installing in another car after 3 years, or if the 45,000 mileage allowance under the Contract Hire Scheme is likely to be exceeded. Under the Hire Purchase Scheme customers are responsible for organising and paying for comprehensive insurance, breakdown recovery, servicing and repairs.

Hire Purchase Scheme for Powered Wheelchairs & Scooters

Important factors when considering a powered wheelchair or scooter are current and future needs. For instance, how far needs to be travelled, will the scooter/powered wheelchair need to be used on the roads and will it need to fit in the car. Every product on the Motability Scheme is covered by route2mobility's insurance policy.

Choosing a Car

Cars are supplied through a network of over 3,500 Motability dealerships across the UK. It is important that a customer chooses the right car. Considerations may include:
Comfort and suitability for the driver and passengers
Other people who may need to travel in the car at any time
Equipment such as wheelchair lifts which may help the person with a disability get in and out of the car
Space and easy loading for a wheelchair now or in the future
Special adaptations for individual needs
The cost of motoring – fuel consumption

Extra Financial Help

Motability provide extra financial help for customers who need this to pay for suitable cars, adaptations or driving lessons. A range of grants are available from Motability's own Charitable Fund and the government's Specialised Vehicles Fund, to help customers who could not otherwise afford the vehicle and adaptations needed.

For more information about Motability contact 0845 456 4566
or visit www.motability.co.uk

Dribble Bandanas

The Funky New Alternative to a Bib

Dribble Bandanas make age-appropriate alternatives to bibs. Perhaps you are the parent or carer of a young person or adult with special needs? Having problems finding an age-appropriate alternative to a bib? Dribble Bandanas produce handmade, funky bandanas that can be used instead of bibs. As youngsters with special needs grow up, they no longer wish to wear bibs that are traditionally associated with babies and toddlers. This alternative bib offers them the protection that they need while also acting as a fashion accessory.

Dribble Bandanas are only available to be ordered online and can be viewed at www.dribble-bandanas.co.uk

Specialist chairs from Huntleigh Renray

Huntleigh Renray Ltd has recently launched two new chair ranges specifically aimed at providing a welcoming, relaxing and comfortable environment for children with special needs.

The Huggle range of chairs has been designed for the profoundly disabled child who needs a comfortable environment in which to relax, whilst maintaining a degree of postural support.

The deep sides of the upper section provide excellent lateral support and the cocoon-like effect promotes a feeling of relaxation and well-being.

For more information check out
www.huntleigh-renray.com

Toytalk Ltd. produce resource material for therapy and education for people of all ages.

www.toytalkdirect.com

Are you a parent of a child with Sanfilippo Disease?

"I'm Meghan, the mother of Geoffrey who is 8 years old and has MPS III Type B. I'd like to be in touch with parents who have a similarly affected child and who are going through the same things as we are. Thank you." Email: goodmeg@hotmail.com

Can you help?

The Genetic Interest Group (GIG) have been contacted by a BBC journalist who is putting together an episode for the 'It's my Story' series on Radio 4. The programme will be broadcast in June this year.

Background

The programme is about the journalist's own personal journey of wanting to have children knowing that there is a 50/50 chance of her passing on her disability. The programme will be exploring the emotions of (potentially) being a parent to a child with a disability and the fact that the disability is bound up in the journalist's own identity. Candida will also be looking at what that says about how she feels about herself.

Candida Harris is therefore looking for people who are living with a genetic condition and are considering or trying for a family/are pregnant or have recently been pregnant or had a baby so that she can explore some of the experiences that others have gone through.

She would initially like to speak to people over the phone and then, if they are willing, she would like to interview them in person. She is happy to travel and do the interviews out of office hours. The amount of input that people would like to give is quite flexible, and if people wish to tell their story but remain anonymous then that is also OK. She is keen to get a variety of opinions and stories to put into this half hour documentary. She would like to begin interviewing as soon as possible.

Can you help?

Do you have a story that you could share with Candida which she can use for her research and background to the programme? Would you be interested in speaking to Candida? You don't have to appear in the radio programme.

If you are interested in finding out more contact Candida directly:
Candida Harris, Producer, BBC News and Current Affairs
Tel: 020 8576 1109 (Monday - Wednesday), Mobile: 07890 466584 (any other time)

KidsOut is a national Charity providing fun and happiness for disadvantaged and special needs children up to the age of 18. The Helpline offers help and support on many issues including parent support and self-help groups, grant aid, respite care facilities, special playschemes, as well as offering a listening ear when things get too much.

KidsOut grants are available to families, schools and groups that look after children with a wide variety of special needs. KidsOut helps children with physical disabilities; learning difficulties; those suffering from life limiting illnesses; abuse; and those from severely deprived or disadvantaged backgrounds. The grant aid could be for a holiday, to pay for a carer, respite care, play equipment or to go on a fun day out or activity break.

For more information on how to apply, please call the confidential KidsOut Helpline on 01525 385 232. Visit the KidsOut website - www.kidsout.org.uk - for more information about the charity. The National Charity for Children with Special Needs

For re-homing

Sleepsuits: Back double zip 3/4 opening
Sleepsuit 1 Body length 18 inch, leg length 11 inch
Sleepsuit 2 Body length 21 inch, leg length 15 inch
Dungarees: Body length 20 inch, leg length 18 inch

If you are interested please contact Andy & Kate Hall, Isaac's Mum & Dad on 01386 872743 or 0789 1412187.

For sale

Recaro car seat - Recaro Start Plus Special Needs 5 point harness, multi-recline, seat depth adjustment, built in speakers, swivel base for ease of lifting, washable covers, used for 3 months only, immaculate condition. Cost over £800. We are looking for offers of around £400. For ages up to 12 years (15-36kg). Please note the maximum weight for this seat is 5 stone 8 lbs. We will consider all reasonable offers.

Support Group Art Workshops

For those members with 'an artistic bent' Nowgen is running a series of art workshops for members of genetic support groups, their families and those in some way affected by genetic diseases. The workshops have been organised by GIG's Artist-in-Residence, Sayward Morley, and focus on personal identity, explore emotional issues, relationship development and communication. The workshops will be held at the Nowgen Centre, Grafton Street, Manchester, they are free and open to everyone and you don't need to have an artistic background. All children attending should be accompanied by an adult.

There have been two workshops already this year with the artwork produced so far apparently fantastic and everyone who attended has had a really good day out, both young and old! The workshops don't just cover art, but also music, puppetry, dance, poetry, textiles and a journal workshop.

If you are interested in attending these workshops, phone 0161 276 8943, fax 0161 276 4058, or email: sayward.morley@cmmc.nhs.uk.

Society for Mucopolysaccharide and Related Diseases

National Registered Charity No. 287034

Become a Friend of MPS

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

What are the benefits of becoming a Friend of MPS?

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

Phone 01494 434156
or download a form from
www.mpssociety.co.uk