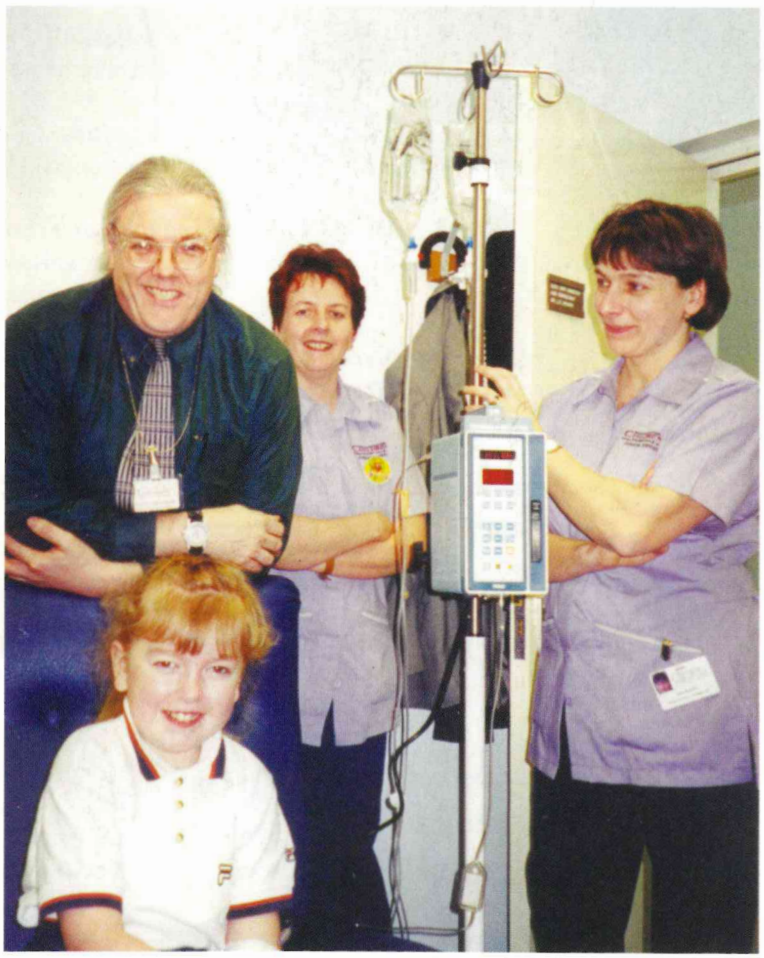
Newsletter

The Society for Mucopolysaccharide Diseases

National Registered Charity No.287034

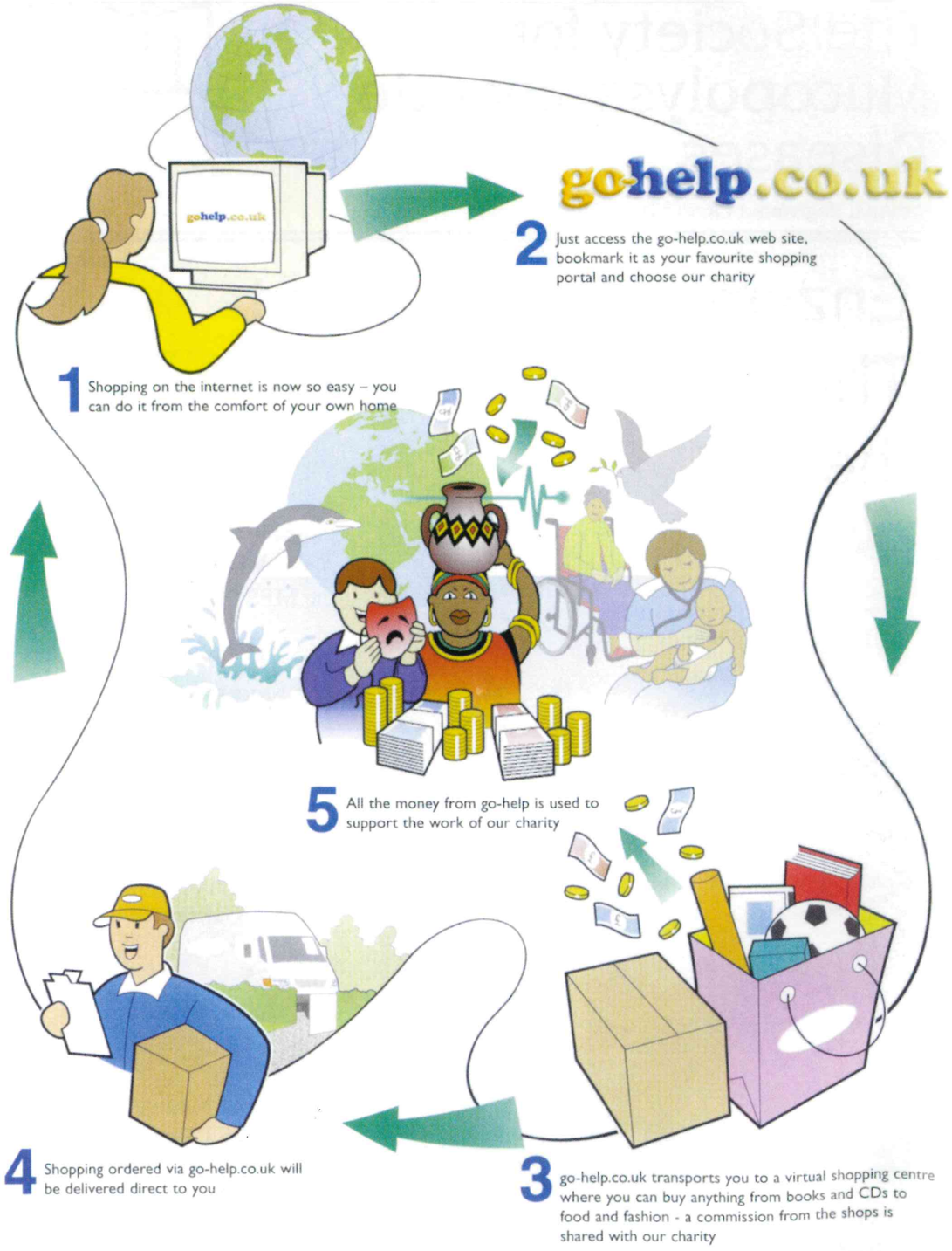
Spring 2001

Enzyme Replacement Therapy Trials Start in the UK



Shop on go-help.co.uk and raise money for

The Society for Mucopolysaccharide Diseases



'CARE TODAY, HOPE TOMORROW'

What is 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 Mucopolysaccharide and Related Lysosomal Diseases, their families, carers and professionals. It is a registered charity entirely supported by voluntary donations and fundraising. It is managed by the members themselves and its aims are as follows:-

- **To act as a Support Network for those affected by MPS diseases and related diseases**
- **To bring about more public awareness of MPS and related disease**
- **To promote and support research into MPS and related disease**

How does the MPS Society meet these Aims?

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

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

Regional Events
Social events held throughout the United Kingdom for mutual support

National Conference and Sibling Workshops
Held annually and offering families the opportunity to learn from professionals and each other

Information Resource
Publishes specialist disease booklets and other literature.

Quarterly Newsletter
Containing information on disease management, research and members' news. Sent to all MPS families free of charge.

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

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

CONTENTS

A WELCOME FROM THE VICE CHAIRMAN

A Welcome from our Vice Chairman, Angela Brown

Welcome to this edition of the MPS Newsletter. I would like to start with a little reminder to post your Conference booking forms as soon as possible. The Bone Marrow Conference and the Annual MPS Conference dates are drawing closer. The number of places available are limited and we don't want anyone to be disappointed.

As you will be aware we are heavily committed to the Jeans for Genes appeal. It may feel that we have only just finished the last one but please help to make this year's event on the 5th October the best ever. The sky's the limit, even if it is the kids schools or your local shops. Why not try something new? Enclosed with this Newsletter is a questionnaire so that you can provide the MPS Society with information that may help the Jeans for Genes appeal more than you may realise. Please complete the questions and return the form as soon as possible. We look forward to hearing from you.

The last Management Committee was particularly exciting. The Trustees met to consider the allocation of Jeans for Genes research funds. On behalf of the Trustees I am delighted to tell you that research grants totalling £ 236,000 were awarded. Full details of these grants can be seen below.

After what seems to have been such a long Winter, the worst for many years, particularly in Scotland, hopefully the weather man has got it right and we can look forward to an Indian Summer. On that note I bid you farewell and look forward to seeing as many of you as possible at the Conferences.

Grant Awarded to:
Royal Manchester Children's Hospital, Great Ormond Street Childrens Hospital, The MPS Society.

Project: To study the long term psychosocial outcome following Bone Marrow Transplant (BMT) for MPS diseases as a prerequisite to Enzyme Replacement and Gene Therapy

Funding: £50,000 (fIrst year)

It has been 20 years since the first bone marrow transplants for MPS 1 were carried out. There is little doubt that BMT has helped many patients by reducing the severity and extent of the various complications that they get. However, BMT is not a cure and patients continue to suffer from various problems to varying degrees. One area that has recently come to people's attention is the difficulty that older children and young adult survivors of BMT have in adjusting to society and the demands of everyday living. Often, they have difficulty in carrying out simple tasks such as shopping or opening a bank account. There may also be more deep-seated emotional issues, particularly those relating to sexual awareness and frustrations arising from physical disability.

It was felt it would be helpful if a survey to assess the nature and full extent of the problem were carried out. The earliest survivors are still only in their early twenties. It is therefore a good time to do such a study. It would be carried out over a period of two years by a nurse specialist and a psychologist working together.

Grant Awarded to:
Institute of Child Health London, Willink Genetics Unit Manchester

Funding: First year funding £186,000

Project 1: To establish the molecular basis of novel mutations found in patients with MPS I - Hurler disease and MPS III -Sanfilippo disease.

Project 2: To develop gene therapy for MPS diseases using herpes virus vectors.

Project 3: To develop gene therapy using bone marrow cells and to investigate how the patients tolerate these new cells.

Project 4: To produce antibodies for use in gene therapy.

The Mucopolysaccharidoses (MPS) result from genetic defects in enzymes involved in the turnover of large molecules called glycosaminoglycans (GAGs), which are important in maintaining the body's skeleton and in brain function. Recent research has indicated that it might be possible to replace the defective enzyme in MPS by putting a good copy of the gene that is responsible for the synthesis of the defective enzyme into a patient's cells - gene therapy. The major problems with gene therapy are rejection of the replacement protein by the immune system of the body, getting the replacement enzyme or gene into all tissues, particularly the brain and the short life of the replacement gene or enzyme. These projects seek to solve these problems.

FAMILY NEWS

Ashleigh Brown aged 9, interviews her Mother Angela

Q. Where were you born ?
A. Girvan in Scotland

Q. How old are you ?
A. 21 of course (not really, I wish, I'm 31)

Q. Where do you live ?
A. In sunny Livingston

Q. What do you like best about Livingston ?
A. Livingston is very central. We are only 15 minutes away from Edinburgh and 30 minutes away from Glasgow

Q. What is your favourite record ?
A. I love dance music, anything that I can dance along to.

Q. If you could be someone else who would be ?
A. This is a hard one. I'm not sure but probably a man because they get it a bit easier than us poor old women.

Q. What do you like most about yourself ?
A. I like being a mum especially when you are being good Ashleigh.

Q. Why did you want to be a Trustee of the MPS Society ?
A. Aiden had started nursery so I was at a bit of a loose end and wanted to do something new but did not want to take a job. I was voted on as Trustee in 1999 and have really enjoyed my time so far.

Q. What would people be most surprised to know about you ?
A. I am very nervous, honestly I really am.

Q. Where did you go for your holidays last year ?
A. Chester in our caravan. We had brilliant weather and the children behaved themselves.

Q. Have you got any pets ?
A. A dog called Amber and two Guinea Pigs, Cherry and Mischief

Prime Minister, Tony Blair meets Angela Brown and her Children Ashleigh and Aiden at St. John's Hospital, Livingston

NEWS FROM THE MANAGEMENT COMMITTEE

The Trustees met in January. Dr Ed Wraith and Professor Bryan Winchester joined the Trustees for the first part of the evening and heard their presentations in respect of their Jeans for Genes Programme Grant application.

Policies
Trustees agreed to the implementation of a Group Pension Scheme for all staff in accordance with the new Stakeholder Pension Legislation to become law on 1st April 2001.

Trustees approved the Financial Assistance Scheme form and agreed that requesting details of income is appropriate practice for grant making charities. It was agreed that applications for financial assistance to attend MPS events will be agreed through the Chairman. All other applications to be agreed by the Management Committee.

Staff Changes
The position of Senior Development Officer was reviewed. Following her interview Ellie Gunary has been appointed Assistant Director. It was agreed that the Chief Executive Officer, Christine Lavery should use the title Director when this was more appropriate. The title of Administration Officer (Finance) was reviewed. The Trustees agreed that Gina Page's post should be Finance Officer. The post of Project and Information Officer has been filled on an acting basis by Alex Roberts.

Health and Safety
Trustees agreed an occupational Health scheme run by the NHS for staff newly appointed and those with unsatisfactory sickness records. A declaration of convictions form was also agreed and to be signed by current and new staff.

MPS Regional Events
Trustees considered a letter of concern regarding the Society's charging policy for regional events. It was agreed that this policy which has been in place for a number of years should stand. This reiterated that where individual families find the subsidised cost is prohibitive, they may apply under the Financial Assistance Scheme.

Membership of the MPS Society
Trustees agreed the proposed membership form subject to one alteration. The Trustees agreed unanimously to include a statement on data protection on the reverse of the form. It was noted that membership is solely concerned with voting rights and not about receipt of MPS services including support and advocacy.

Enzyme Replacement Therapy (ERT)
The Director updated the Trustees on ERT Clinical trials for MPS I, MPS II and MPS VI.

MPS Conferences
Trustees were presented with the final programme for the MPS Bone Marrow Conference to be held on 8th June 2001 and the MPS Annual Conference to be held on 14th -16th September 2001. Both at the Hilton Northampton.

Jeans for Genes
The Director informed Trustees that the Cystic Fibrosis Trust had been offered a further year's partnership at 10% It was agreed to advertise the Jeans for Genes appeal on the MPS Society's Web Site.

Mark Beniston
Chairman

NEWS FROM THE MPS OFFICE

Christine Lavery
Director

It is with considerable sadness that we said goodbye to Hannah and Kate in February and Sue Taylor in March. Hannah had been with the MPS Society for five years, firstly as a student fundraiser and regular volunteer then as Assistant Development Officer before being promoted to Development Officer in 1999. She was much respected by the families she helped and will be missed. We wish Hannah, Kate and Sue every happiness for the future.

In February Antonia Crofts was appointed Assistant Development Officer. Antonia is the Society's first point of contact for Family Support and the advocacy service. She is also responsible for the MPS outreach clinics and all information enquiries. On 12th March Alison Britton joined the Society, funded by TKT, to work full time on the MPS European Registry (formerly known as database). Alison will be working with the British MPS families consolidating our data, as well as working with the European MPS Societies. Alison is also responsible for producing non-confidential data on the incidence and epidemiology of MPS and Related Diseases.

In April the Development Officer to succeed Hannah will take up post as will a second Development Officer to work with Ellie. Sarah and Andy will introduce themselves in the next newsletter.

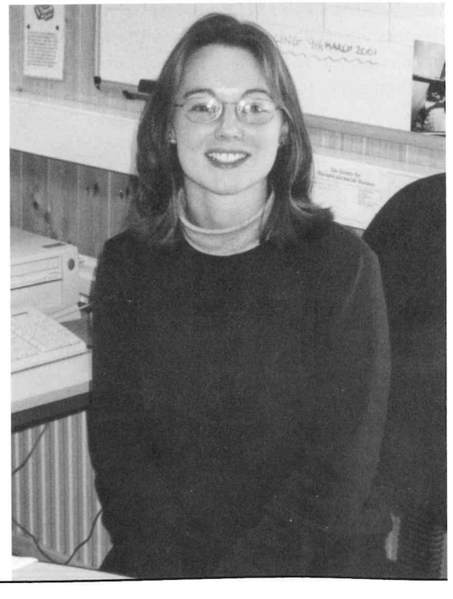
Since Gina took up post in November 1999 as Finance Officer taking over from Lynne Grandidge who was also Treasurer. Gina and Sue have worked together streamlining the financial administration. They have made such a good job of the task that the Trustees have accepted the recommendation not to appoint to Sue's post.

Alison Britton

I am a new recruit to the MPS team. I am currently working alongside Angela in maintaining and updating the Research Database for countries throughout Europe and beyond. I am enjoying this new role as I also know that it could possibly lead to a new treatment or therapy being discovered.

I look forward to meeting some of you soon so as I can relate what I have been learning on paper to real life. I hope I will measure up to the high standards of members currently in the team.

Antonia Crofts



I joined the MPS Society in the middle of February as Assistant Development Officer. Although I am still settling into my new post I am thoroughly enjoying the work.

Being the first point of contact on the telephone, organising the administration for the regional clinics and befriending scheme and maintaining the Society's information resources will all be important aspects of my work. These tasks should give me the opportunity to speak to many MPS families over the telephone and also, hopefully in person, as time goes on.

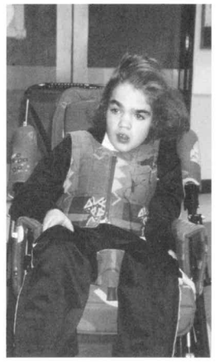
I am really enjoying being part of the Society, in particular the team spirit and find the work very rewarding. I hope that the MPS Society continues to be able to support as many families as possible and look forward to the future with new research and treatments.

A WEEK IN THE LIFE OF THE MPS SOCIETY

Monday

Monday starts as most do. A large weekend post bag, faxes, messages left on the answer machine and an increasing number of e-mails. Angela reports a quiet weekend on the out of hours mobile with calls from just one family. It is now Antonia's third week in post as Assistant Development Officer and things are moving along nicely for the team. Her priority today, apart from being the first point of call on the MPS phone is to put the finishing touches to the Bristol and Cardiff clinics as they are only ten days off, and get arrangements underway for the Northern Ireland Clinic in May.

The depth of the advocacy service sought by MPS families has increased considerably over the years. With so many local authorities supposedly strapped for cash it can require shifting Heaven and Earth to achieve the needs of MPS children and adults, their families and carers and sometimes Heaven and Earth can't be shifted. Today, there is the problem of the through floor lift installed for Thomas when he was no longer able to climb the stairs, but able to walk or be pushed in a light weight buggy. Now that Thomas can not walk at all and is pushed in a large buggy there is no room for a parent or carer to accompany him in the lift. The question everyone except the occupational therapist is asking is, is it safe and acceptable practice for Thomas (who is in the last stages of his disease) to travel in the lift alone, whilst an adult runs through the house up or down the staircase to meet the lift at the top or bottom? What happens if the lift breaks down (as it has three times in as many months) or the parent carer has an accident en route rushing to meet the lift? On behalf of the family and the Society I write to the Health and Safety Executive and now await their guidance.



Melanie Jones at the Cardiff Clinic

Tuesday

Tuesday, and I need to be treated delicately this morning. Last night very close friends in Germany told us that one of their twins, 17 year old Hendrick had died that morning. Both Hendrick and his sister, Emily, suffer from Sanfilippo disease. I felt pleased that only 10 days previously we had been able to spend a few days privately with the family. When I hugged and kissed Hendrick and Emily goodbye I knew that it was almost inevitably for the last time. But this had happened too quickly. An evening of decision. How we might get to Hendrick's Funeral on Friday? All the family want to attend but can we face another 800 mile drive? Can we get leave from work? But now back to today. An MPS mum calls in a distressed state. She's been denied a buggy for her 15 year old daughter who has Sanfilippo disease. Wheelchair services are only offering, to use a government phrase, 'bog standard' wheelchair model. Having had several attempts contacting the occupational therapist there is no alternative but to put pen to paper .

This afternoon, representatives of the company OmniVision are coming to see me. Trustees have agreed to commission a promotional video on Mucopolysaccharide and Related Diseases, how the diseases are caused, current and future treatments and work of the MPS Society. Christopher Morris (ex BBC journalist) will be leading the production team. The meeting went well and it is now down to me to pull all our ideas into a draft script BY FRIDAY!! There is no time to lose as we want to capture key events taking place in the Society this year and have the video ready for November 2001.

The Alvema Max

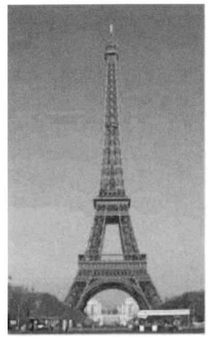
Wednesday

Today is eight weeks after the Conference booking forms for the MPS Annual Conference were sent out to families. Early indications are that we will be fully booked again this year. Another MPS Conference, '21 Years of Bone Marrow Transplantation', will take place in June. I meet with Alex to discuss possible contingency plans for the children's programmes in the light of the recent foot and mouth outbreak. Woburn Safari Park is currently closed.

A Development Team Meeting highlights the need to continue to prioritise the Society's advocacy support until the Development Officer to replace Hannah starts on the 17th April, and a second person starts in a similar post the following week. For years the MPS Society has supported the campaign to lower the age of eligibility to 3 years for receipt of the mobility element of the Disabled Living Allowance. With no great fanfare the disability lobby has won, and as of the 1 April 2001 all children receiving the higher rate care component of DLA are being invited to apply for the mobility element. Ellie reports that she and Antonia have resorted to desperate measures, becoming experts at completing three forms at a time for children with the same disease! Much needed with so many families asking for help in this area.

A WEEK IN THE LIFE OF THE MPS SOCIETY

Thursday

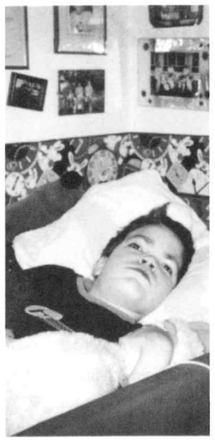


Thursday is a day of short meetings squeezed between advocacy support. I have been asked by one of the pharmaceutical companies to make a presentation about the voluntary sector in Europe at their business meeting in Paris next week. Angela and I meet to agree the preparatory work and the production of information packs and overheads. We only have until end of play next Tuesday to have everything ready as I shall leave for Bristol on Wednesday to carry out a Care Plan meeting with one family, and to give a talk to Fosseway School who have four Sanfilippo children as pupils, before running the Bristol clinic on Thursday and flying to Paris that evening.

The Society has received a grant towards funding the European Registry for MPS and Related Diseases. Alison Britton has been appointed to carry out this work and starts on Monday. Angela and I discuss Alison's induction and check that all personnel matters are in hand.

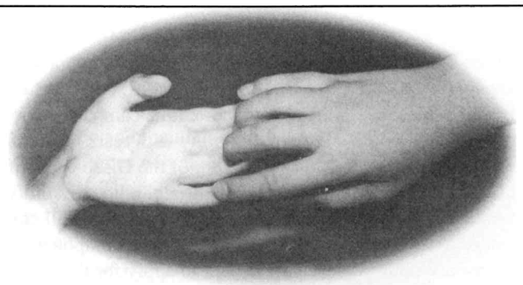
Gina and I then meet to discuss the draft accounts in the light of the Auditor's comments, and prepare the papers for the Trustees' Meeting in three weeks' time. Draft Accounts means Annual Report: another job for Alex, and I to add to the list.

Friday



Hendrick February 2001

Friday is a day's leave but the emphasis is still very much MPS. After consultation with Robin and the boys, Lucy (our 14 year old daughter) and I are on the early morning flight to Cologne to say goodbye to Hendrick. Friends and family have come from far and wide to gather in the small and charming town of Sinzig to give Hendrick the biggest and most individual funeral Sinzig has ever seen. German law does not allow burials in the afternoon, so Hendrick's parents, Richard and Liz, changed the order. First, we gathered at the little cemetery above the town for Hendrick's committal to the earth. Over three hundred filed passed and as Liz put it 'she shared a lot of, make up with a lot of people' as they hugged, cried and laughed. Mourners were given helium balloons with the name of the Children's Hospice, Baltazar, where Hendrick spent his days before the funeral with his sister Emily. The balloons were all let off in one go around Hendrick's grave by young and old alike.
It was then on to the church for a service of celebration, and afternoon tea, at the nearby Community Centre. Lucy having decided to stay on in Germany until Sunday with Richard, Liz and Hendrick's healthy siblings, Tilmann (13years) and Sophia (7years), I travelled home alone and exhausted. The one hour flight time was taken up reflecting not just upon the day's events, but our family's friendship with the Volk family. It started when Liz Volk contacted me in 1986, having seen an article about the Lavery Family in The Independent, and not long after Hendrick and Emily's Sanfilippo diagnosis. It was cemented at the MPS Conference in 1987 when Hendrick having been tucked up in bed, and supposedly asleep, found his way out, down in the lift and about to exit the hotel through the automatic doors when he was sighted and recognised by Robin, I safely returned him to his parents. Finally, my thoughts turned to all those who face the loss of a child, and what it must be like for Emily. Hendrick was her hero, and her constant companion through 17 years of Sanfilippo disease.



Hendrik Oliver Volk

* 30. März 1983 † 5. März 2001

Hero of his own life

Christine Lavery
Director

FAMILY NEWS

New Families

There have been eight new families seeking the support of the MPS Society since the last Newsletter. This includes the new families below who have given written permission for their details to be published.

Lisa and David Graver's daughter Lauren has recently been diagnosed with Hurler disease. Lauren is one year old. The family live in Cornwall.

Lisa Patrick and Mr Culverwell's son Reece has recently been diagnosed with Hunter disease. Reece is four years old. The family lives in Somerset.

Births

Adrian and Marrianne Stimpson have a new baby daughter, Jodie, who was born on 4th October 2000. A healthy sister for Dominic who suffers from Sanfilippo disease.

Simon and Nicola Hall have a new baby son, Corey who was born on 27th February 2001. Corey like his sister Jade has Hurler disease.

Robert McLean and Karen Calvert have a new baby son, James, who was born 14th November 2000. A healthy brother for Kirsty and Craig.

Corey Hall

Jade Hall

Congratulations

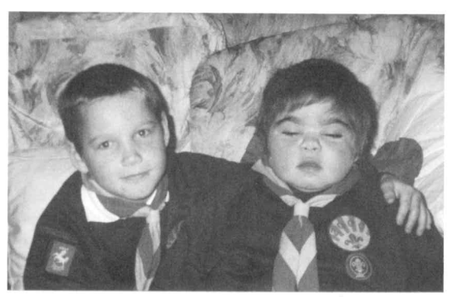
Wing Kim Yeung celebrated his 21st birthday on 1st March 2001. Wing has Maroteaux Lamy disease.
Louise Hale celebrated her 21st birthday on 7th April 2001. Louise had a BMT for Hurler disease.

Coronation Street Star

When Blaise Leslie, who suffers from Sanfilippo disease and his mum Terri-Ann visited hospital recently they were very surprised to meet Steve Mcdonald from Coronation Street. Terri-Ann has given us the photograph to publish.

Cub Scout Thomas Birch

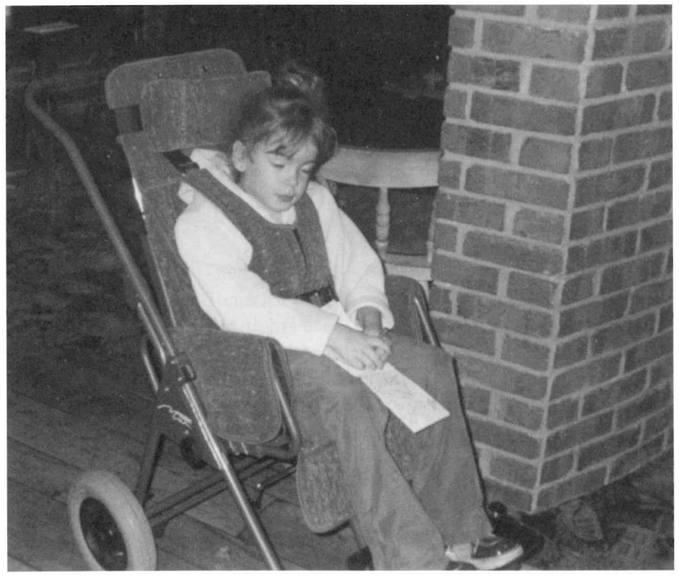
Thomas Joined the Demelza House Cub Pack in Summer 2000. He is pictured here with his cousin Christopher who is also a member of the same pack. Demelza House is the first Children's Hospice to have its own Cub Scout group.



FAMILY NEWS

What A Major Disaster

Hi I'm Paula Robjohns, Jordane's mum. As we all know, living with Sanfilippo Children can be quite challenging! More so if we have the great pleasure in taking them shopping or just out and about in the shops doing our chores. But when the pleasure becomes a pain is when the major buggy or flimsy buggy, I'm going to call it, becomes a little too flimsy for our kids as most of you are probably aware of. That's where our problem began!



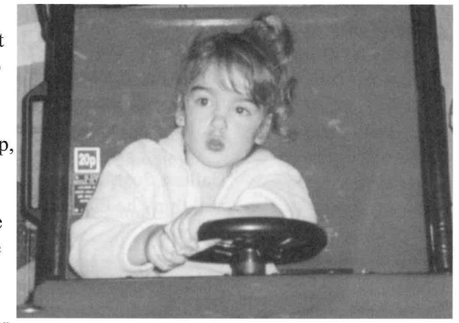
Jordane Started to climb out of the buggy, despite being strapped in. If that's what you can call it. The straps on the Major Buggy aren't that what you can call secure, safe, comfortable straps. You either strap it around their waist, their chest, and if you're lucky enough, as in our case, and got an extra third strap, you could fasten that and it would have fastened around Jordane's throat (HOW STUPID !!) She then started to tip the buggy backwards, and over she would go banging her head on the floor. That was the final straw. I telephoned Ellie at the office and after a lengthy chat we decided that the Alvema Max would probably suit Jordane the best.

The following day I rang my O.T as discussed with Ellie. I thought the O.T was there to help us and generally be in support of our children's needs. To my absolute horror how wrong could I have been? My O.T suggested that we have the NEW Major 2000 Buggy, with re-vamped super straps. So being as naive as I was then....Agreed!

Later that night my husband and I went onto the internet to check the major 2000 buggy out. To our surprise it's the same flimsy buggy, just different material and even more stupid straps. So the following morning I got onto the telephone to Ellie for yet another lengthy chat. Following further discussions with the O.T, we ended up going to the wheelchair clinic for an assessment.

Finally after weeks of waiting the day came. Myself and my sister, Jordane and her younger sister Brooke (oh and of course the buggy) went for what we thought to be a normal perhaps half an hour appointment. Again oh how wrong we were. We were there for 3 and a half hours. The appointment only lead to Jordane getting more and more upset in the buggy , my sister run ragged from chasing Jordane around when we let her out of her buggy, Brooke having a great time climbing in and out of all the different wheelchairs, and me in floods of tears, trying to explain why Jordane needed an Alvema wheelchair. The whole afternoon was a complete waste of time. The outcome being Jordane didn't need an Alvema and the Major 2000 was thought to be satisfactory. At that I voiced my opinion quite strongly and said if Jordane sustained any neck or head injury the blame would be on them. Almost immediately Jordane was given a Major 2000 to come home with on trial, as it was a little safer than the ordinary major buggy. But we were told we must keep hold of the handles at all times as the buggy could tip over. Even with them knowing this danger still they let us come away with something they knew was not safe. As you can imagine we came home not in the slightest bit satisfied.

The following day another phone call to EIlie. At this point I was too emotionally involved. Just thinking WHY! ...Our children go through so much. All I'm asking for is what she needs and deserves. At this point I was more determined than ever! Ellie got the ball rolling, letters and phone calls to people in high places, eventually reaching the director of Social Services. At last something was to be done! We were issued another wheelchair assessment at special seating. After speaking to Christine thankfully this assessment was cancelled, and as all that was needed was my O.T, the Alvema rep and Ellie to meet. Thankfully it was done at our home, where Jordane was more relaxed. We tried her in the Alvema Max and instantly I knew that it was the buggy for Jordane. And on that the O.T had to agree. From that meeting the wheelchair was ordered and delivered within a few weeks. So from a major disaster to almost a miracle. I must say a big thank you to Ellie and Christine, who gave me all the support I needed all the way through fighting for what Jordane needed. Not only did Ellie get the Alvema, she also at the same time fought to get Jordane a Kirton chair for home. Ellie then wrote off to a couple of charities and got funding for some accessories for the Alvema. The whole process took 6 months, but could have taken a hell of a lot longer if not for Ellie and Christine's magic letters and phone calls! As we all know the Alvema and similar wheelchairs don't solve all our problems. It just means that when we go shopping we can actually buy something, take it to the till and LET go of the handles of the buggy and pay for it! Without the risk of the Major buggies tipping over. Makes for an altogether a safer and stress free shop! Just to finish off by saying to everyone that is going through a similar situation. DON'T give up, fight for what our Children need and deserve. Thank you once again Ellie and Christine I'm not sure if I'd have had the strength to fight any longer if it wasn't for you.



The Alvema Max mentioned in Paula's article is one of a number of buggies the MPS Society recognises as suitable for children and adults suffering from MPS or a related disease.
Ellie Gunary

Paula Robjohns

FAMILY NEWS

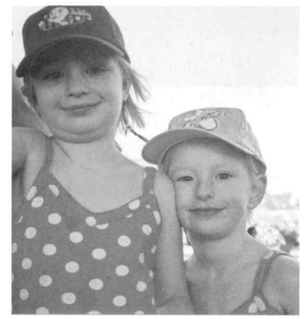
Dial-A-Dream

My name is Sue Stuart and my family consists of my hubbie Peter, my daughters Jessica (10), Hollie (6) and Annie (4). Jessica has Hurler Syndrome. I would like to tell you about our experiences with Dial-A-Dream.

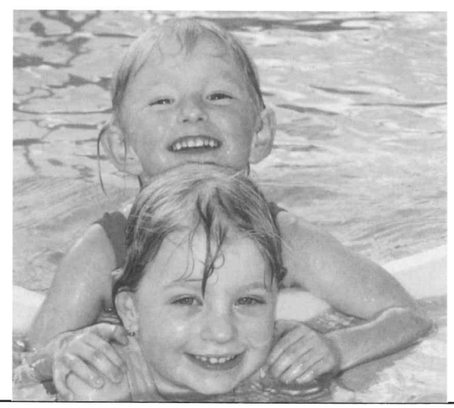
I first wrote an article about the Dial-A-Dream villa when my husband and I went to America with our daughters in October 1997 (Winter Newsletter 1997). We had such a lovely time that we wanted to go back. With the help of Dial-A-Dream we did go back in 2000 and 2001 but as Jessica was too ill to travel both times, we went without her. It was a hard decision to make but we felt that it was the right one for the other two children. Jessica stayed in respite care while we were away and loved every minute of it as she had 24-hour attention!

My contact at Dial-A-Dream is a lovely man called Bob Heath, I have spoken to him many a time on the phone asking questions about arranging a holiday to Florida. I was concerned that if we were staying in the villa, a dream family wouldn't be able to use it. Bob assured me that people like us, who use the villa, are doing the charity a service, as our fees go towards the cost of dream families staying, so everyone is welcome. With his help we were able to rent the Dream villa for two weeks, he also gave us advice on how to arrange the best available flights and car hire. To say that the dream villas are luxurious is an understatement. It was like staying in a 5 star hotel without the restrictions. I would recommend that if you are thinking of visiting Disney World in Florida, give Bob Heath at Dial-A-Dream a ring, as I'm sure he will be able to help you out.

Dial-A-Dream is a charity that grants wishes and dreams to children with life threatening illnesses, whether that be to meet Mickey Mouse or to fly in a hot air balloon or to visit a museum. It also owns two luxury villas in Florida ten minutes away from Disney World. The villas can be rented out to UK and USA families providing that a Dream family is not using them. You can find out more about the villas at www.dial-a-dream.co.uk.



Hollie and Annie enjoying their holiday



Sue Stuart

FAMILY NEWS

'Joshie'

Josh was born on the 23rd December 1996, normal birth two weeks early, a bit of a shock as we hadn't finished our christmas shopping!!

Callum, Josh's older brother was delighted, we still think he thought Santa brought him a playmate.

In Josh's early weeks, he had some physio on his head and left foot, both of these problems were rectified within weeks.

In the first year of his life, Josh had constant nose bleeds, and also eleven ear infections. Just before he turned one he was referred to ENT at Ormskirk Hospital, where gromets were suggested. Comments were also made regarding the size of Josh's head.

This played on my mind and on another visit to the doctors (yet another ear infection) I asked if he thought his head was too big. He measured it, and it wasn't dramatically larger than the norm for his age, but said he would arrange a brain scan to ease my worry.

That proved okay. While at the hospital the doctor felt Josh's tummy, his liver and spleen were both enlarged, but she was not concerned because Josh had a heavy cold at the time.

A few weeks later Josh was back at the hospital having gromets fitted, the doctor who had done the brain scan, just happened to be on the ward. She had a feel of his tummy, liver and spleen again. They were still enlarged. She ordered some samples to be taken of blood and urine.

Three weeks later we received a phone call asking us to go to Ormskirk Hospital as Josh's tests had come back and that there was a problem. Ormskirk were very vague about the problem. Four days later we met with Dr Ed Wraith at Manchester Children's Hospital. Josh had been diagnosed with a very rare genetic disorder called Mannosidosis. He was thirteen months old.

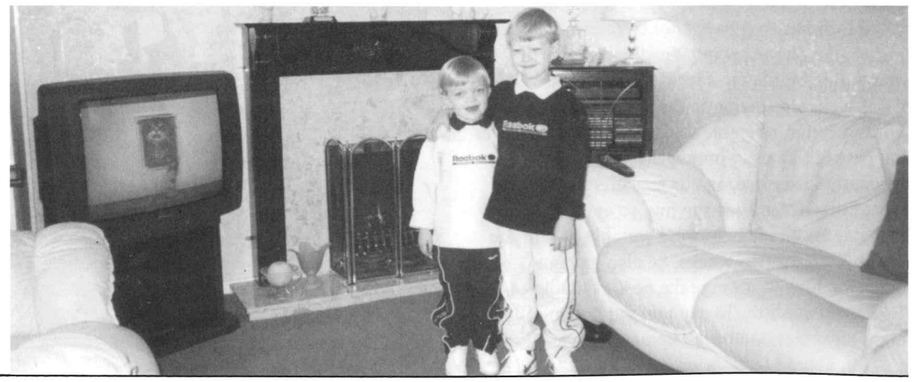
It all really unfolded like a bad dream. Still now when I watch Josh playing with his brother, I still find it hard to believe and think if I had never been to the doctors and mentioned my concerns about Josh's head none of this would have been discovered for at least a few more years.

But we do know now and enjoy him even more. He is the most adorable little boy. Everybody who knows him loves him. I haven't just made this up to compensate for what my son has been dealt. We think someone has given us a little extra bit of 'special' in Josh to help us through the low's.

Josh was four in December. He's doing really well. He wears hearing aids now. The gromets never really worked, and then we have discovered he is slightly deaf anyway. He is so sweet, and he likes his hearing aids. He has support at nursery school, he doesn't eat or sleep very well at all, although he drinks pints and pints of milk. I've never seen a child enjoy a bottle like our Joshie.

Things do get very difficult at times. Lack of sleep being the main problem. But Josh gives us so much pleasure. Everything new he does like starting to talk properly is such a big bonus, even to Callum, they can argue and understand one another now! Although we wish Josh didn't have what he's got we just thank God we have got him at all.

Joshie with big brother Callum.



Beverley Westhead

OVERSEAS

TJ's Story (continued from Winter 2000 Newsletter)

He enjoyed the aeroplanes and loved flying. He was also fond of trains, but he loved it best in his own home where he could forward and rewind his favourite Irish music videos. Over the years we had some wonderful times with TJ, watching the moon at night time with my neighbour's binoculars and feeding the birds early in the morning. Everybody knew when TJ was around because he loved to whistle.

At the age of 7, TJ stopped talking but we could still communicate using his own sign language. He could tell us when he was hungry or thirsty. Shortly after that, TJ stopped walking. For me this was a very difficult time. I tried to understand why. Connie and I had to watch him become less and less able as each day passed; watch him deteriorate from being a very active happy little boy to being totally dependant upon us for his care. TJ could no longer talk. He could not even tell us if he was cold or hungry and he could not enjoy the things he had loved doing.

Connie and I had to get used to our 'new quiet' little boy. I sometimes felt as if somebody had put him in a little box, a safe place where he could stay and live on his memories, away from the people and reality. Perhaps nature's cocoon, preparing my little boy for bigger things? He looked at his photographs over and over, which I had collected for him over the years. Everyone who spent time with TJ was touched by him. He was generous with his love.

TJ couldn't hold his own glass anymore and he had problems swallowing. Gradually he stopped feeding himself and at the age of seven and a half I started to liquidise his food. He still got excited when people came to visit us. I bought TJ a wheelchair and he really loved it. On weekends, I took him to the zoo to see his favourite animals.

At the age of eight and a half, TJ started to get epileptic fits and I had to take him to hospital more and more often, where he stayed for two to three days at a time. He got pneumonia very quickly and he had problems with his teeth. His hands were sometimes sore and he got headaches very often. It took me a very long time to get used to the fits. I always felt so helpless because I knew that there wasn't really anything I could do to help him or ease the pain. All I could do was to make him as comfortable as I possibly could.

On TJ's 9th Birthday he couldn't sit up straight any more and we had to support him when we took some pictures. He never wanted to be left alone in the room and his eyes followed you everywhere. I noticed that he couldn't cry anymore. He would hold on to your hand tightly while he was watching his favourite videos. As TJ was lying on his bed or on the couch in the living room most of the day, he started to get bedsores. I tried all the creams and treatments that were available and luckily the 'tea tree oil' came to our rescue!

On the 14th September 1999, when I came home from work, I sat with him, kissed him and talked to him. When I looked at him, it was as if he was looking past me, not really recognising me. I took him to hospital and he stayed there overnight. I could see that he was tired and I knew that the time had come for me to let him go. I couldn't have asked God for more time with TJ. He had given me so much already! So many memories, so many stories to tell, so much to remember! When I left the hospital, TJ gave me his last brave smile - the following day at 13h10, TJ passed away at the tender age of 9 years.

Through TJ I have met some wonderful people over the world, been to some amazing places, and learnt more about life than I ever would have believed possible. Without TJ, I would not be the person who I am today. He has taught me about loving, about sharing, about understanding and I have learnt to take each day as it comes. To live for today and to fill each day with beautiful memories. The strength and bravery of TJ's soul when his body was getting weaker, puts the trivialities of life into perspective and this was an invaluable lesson.

I know that this might be a very sad story, but this is TJ's story. I want parents with MPS children to know how special they are. That God has chosen THEM to look after these angels for a while. In the end, only you realise how special!

Lynn Pienaar

MPS CLINICS

A Beginner's Guide to the MPS Regional Clinics

2001 Clinics

Northern Ireland
Hilton
Templepatrick
Friday 11 May

Scotland
tbc
Thursday 7 June

Birmingham
Children's
Hospital
Friday 6 July

Bristol
Frenchay Hospital
Thursday 18 October

Wales
University
Hospital of Wales
Friday 19 October

Ellie Gunary
Assistant Director

Where are they held?
Norwich, Birmingham, Newcastle, Bristol, Cardiff, Belfast and Scotland (shared between Edinburgh and Glasgow).

How often are they held?
Norwich, Newcastle, Belfast and Scotland are held annually. Due to a larger number of patients to be seen in the areas of Birmingham, Bristol and Cardiff these clinics are held twice a year. Each child or adult with MPS is offered one appointment a year unless there is a clinical need for an additional appointment.

Which doctors attend the clinic?
A local consultant sees patients with either Dr Ed Wraith or Dr Maureen Cleary from the Royal Manchester Children's Hospital. More often than not these consultants are joined by other local doctors interested in learning more about MPS.

Is a referral needed to be seen at the Regional Clinics?
Each patient to be seen needs a referral from their local doctor to the consultant who is hosting the clinic at the regional hospital. This is needed even if a patient is seen by Dr Ed Wraith or Dr Maureen Cleary because they are being seen within the regional consultants health authority.

Why hold Regional MPS Clinics?
The aims of the regional MPS Clinics are to:-

Enable families and adults affected by MPS to access a consultation with an MPS specialist consultant without having to travel long distances to the clinical centre of excellence.
Increase expertise in the regional centres on MPS diseases and clinical management of those affected.

What is the role of the MPS Society at Regional Clinics?
The MPS Society enables these clinics to happen by funding the MPS specialists attendance. The Development Team liaise closely with the local consultant and the MPS specialist consultant in arranging appointments. Depending on the clinic either the MPS Development Team or the regional hospital staff send out the appointments. There is always at least one member of the MPS Development Team at each clinic to meet families and individuals face to face and offer support with advocacy issues. If a family or individual attending the clinic has a specific issue they know they wish to discuss beforehand it is helpful to let the Development Team know so any relevant information can be taken to the clinic.

Making an appointment at a Regional Clinic
It is essential that when each family or individual receive either a clinic appointment or a request form for a clinic appointment at a Regional Clinic that the confirmation slip is returned. Unless this is received back at the MPS office the appointment will be offered to someone else. Likewise if an appointment is received by a family or individual they do not wish to attend or even at the last minute are unable to attend it is important that the MPS Society is informed as soon as possible as another family may be able to benefit. This arrangement does not apply to Cardiff who arrange their own appointments.

Evaluation of the Regional MPS Clinics
After each clinic an evaluation form is sent to each family or individual who attended. These are collated a few weeks after the clinic and the feedback is taken into account when planning the next clinic. There are of course some things that cannot be changed quickly! Parking, the size of the waiting area if that is all the hospital has to offer and available public transport for example. Continuous feedback of experience of these factors remains valuable however as when a hospital moves or a consultant moves on these factors can be considered when looking for new contacts or locations.

Thank you Ed!

As we have been travelling to support these clinics, so too has Dr Ed Wraith to whom we are very grateful for his continued support of the MPS Regional Clinic Programme.

MPS CLINICS

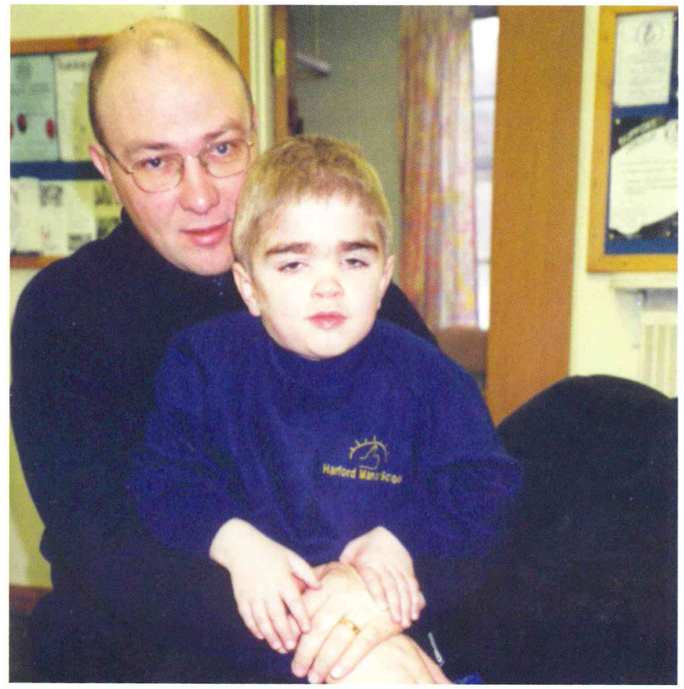
East Anglia MPS Clinic- Friday 19 January 2001

Christine and I attended the morning of this clinic and in the afternoon while Christine held an MPS information session at a local school I continued to see families.

The night before we had been concerned as to whether we would actually make it to the Norfolk and Norwich Hospital as snow and bad weather threatened. We were also relieved that when we left at the end of the clinic this promised bad weather had not yet settled in and it looked as though we would get home.

For me it was first opportunity to meet many of the MPS families and individuals who live in the East Anglia region. Several local professionals attended the clinic with the families they were supporting. This provided an ideal opportunity to share first hand expertise and address issues around housing and equipment as needed.

Dr Ed Wraith along with Dr Chris Upton and Dr Uma Ramaswami who had travelled over from Addenbrookes Hospital, Cambridge, saw 9 children in total, some of whom are featured in these photographs.



Dominic Stimpson with Dad

Birmingham MPS Clinic - Friday 26 January 2001

A week after the East Anglia clinic I was on the road again, this time travelling to the Birmingham Children's Hospital. At this clinic 14 MPS children were seen. This is always a busy clinic and was made even busier by the presence of TV cameras filming two families attending for a BBC television documentary to be broadcast later this year. Angela Ratcliffe and I being camera shy were very happy to stay out of the way of filming though we did catch some of the children on our own camera.

Beryl Holmes, Clinical Nurse specialist at Birmingham Children's Hospital ran the appointments while Angela and I met with all the families attending.

Bethany Allen and Dad

MPS CLINICS

Newcastle MPS Clinic- Tuesday 6 February 2001

Daniel Muers with his mum and new baby sister at the Newcastle Clinic

It hardly seemed as though I had completed the follow up to the Birmingham clinic when I was setting off again for another regional clinic, this time in Newcastle. The weather nearly did hinder this clinic taking place with frantic phonecalls on the day before checking whether the weather reports of local heavy snow were going to prevent MPS families and adults getting to the Royal Victoria Infirmary.

In the event the weather proved no hindrance at all, other than to delay my journey back to London. This was due however to high winds in London rather than poor weather in Newcastle. Being the most economical means of travelling I had flown up for the day.

The clinic ran very smoothly with Dr Andrew Morris, Senior Lecturer in Paediatric Metabolic Medicine, at the Royal Victoria Infirmary seeing 11 patients with Dr Ed Wraith. I, as were many of the families I spoke with, to learn that Dr Andrew Morris is planning to move on from Newcastle in the near future. Dr Morris has been instrumental in establishing the Newcastle MPS Clinic and we are very grateful for his support in getting this annual clinic underway. We extend our thanks to Dr Andrew Morris and wish him well in his new post. In the meantime we are working with Dr Morris and Dr Ed Wraith to identify another local consultant to host this clinic.

Ellie Gunary
Assistant Director

Bristol MPS Clinic - Thursday 15 March 2001

Christian Natrella with his father at the Bristol Clinic

This, I am told, is always a busy clinic and Thursday 15th March was no exception! Due to circumstances beyond our control with British Rail, Dr Ed Wraith was some what delayed in arriving in Bristol. Unfortunately we started a little late with the first clinic appointment and became gradually later with each successive appointment. Generous credit should be given to Dr Wraith and Dr Philip Jardine who together saw fifteen families in total and ensured each family received ample time for discussion.

However, the waiting area itself was very busy with a number of MPS families waiting to be seen. This time allowed families to chat over a cup of tea or coffee, talk to MPS Society staff about points of concern and raise any issues with which support was needed. It is a very good opportunity for us to meet parents and children and we hope that the families feel it is just as worthwhile.

We would like to say a big thank you to Dr Wraith and Dr Jardine and all those who were involved in the clinic arrangements. This was the first clinic I have attended and I can now understand why they have become so vital. Thank you to everyone who attended.

MPS CLINICS

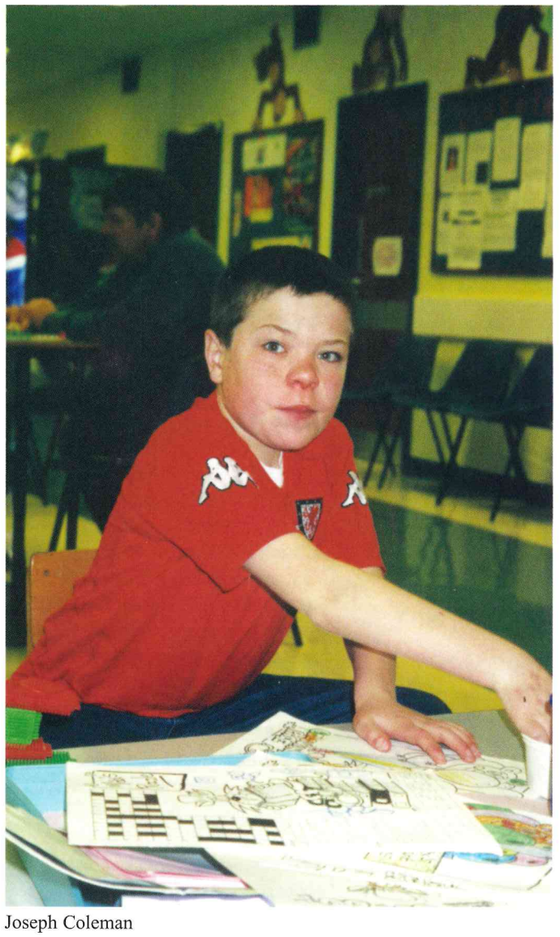
Cardiff MPS Clinic - Friday 16 March 2001

Following on the next day from the Bristol Clinic, Ellie and I travelled to Cardiff to attend the Welsh MPS Clinic. Although slightly less frantic than Bristol, Dr Ed Wraith and Dr Graham Shortland saw nine MPS families. MPS Society staff, meanwhile, sat in the waiting room amidst toys, drawing crayons, pictures of Winnie the Pooh and got covered by food as we looked after some MPS children whilst their parents talked to the doctors. All in a day's work!

We do hope, however, that this clinic enabled families to meet, and provided the opportunity to discuss problems with the doctors and ask any questions. The clinic also gave families a chance to meet with MPS Society staff and raise any issues with which support was needed.

We hope those of you who attended found the appointments beneficial and we would like to thank Dr Shortland and Dr Wraith and all those who were involved in facilitating the clinic and ensuring it ran so smoothly. It was very nice to meet the families and have a chance to talk.

Sarah McKnight



Joseph Coleman

Antonia Crofts
Assistant Development Officer

IN REMEMBRANCE

Deaths

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

Micheal Hoather who suffered from Hunter Disease 13.09.1988 - 14.03.2001

Zain Ilyas Hussain who suffered from Sanfilippo Disease 18.02.1989 - 08.02.2001

Natasha Macintyre who suffered from Sanfilippo Disease 05.06.1979 - 16.01.2001

We also extend our sympathies to Bob and Rhian Mcknight whose baby daughter, Alexandra, born 11th December 2000 who passed away on 19th January 2001. Our thoughts are also with Sarah and Rhoswen Alexandra's sisters.

Ann Canton whose husband who died suddenly on 11th March 2001. Our thoughts are with Ann at this time.

Remembrance day in the Childhood Wood

It is with regret that the Trustees have decided to cancel the 2001 Remembrance Celebration in the Childhood Wood planned for July. Although thankfully as yet Sherwood Forest is free from Foot and Mouth Disease, there is no guarantee that the situation will remain the same. Sherwood Pines has reopened this week but may have to close again if Foot and Mouth comes within 3 miles of the forest.

We had planned a big event this year with marquee, teddy bears picnic and balloon release to bring to an end the Society's 18th birthday celebrations. Recognising that many families living in rural areas have little to celebrate at this time the event has been postponed until July 2002.

Childhood Wood Poem

I am a 72 year old pensioner and l often go walking in the Nottingham Sherwood Pines Forest And just sit and rest on the seats at the Childhood Wood, and think about all the children that are remembered there. I am a father and grandfather and sitting there makes me feel more grateful for what I have, but at the same time it makes me feel very sad for those children and their loved ones. Whilst sitting there recently I wrote the following poem and I hope it will make the loved ones of those children feel just a little bit better.

When you sit in this garden just stop and pray
for these lovely children that have passed away,
They were born into this world to be loved by all
until God with His wisdom gave them a call,
So don't be sad when you rest here a while;
because with God they are all happy
So give them a smile.

They were just children all happy and gay
but God made His choice and took them away,
He made them His angels in heaven above
now they have no sorrow because of his love,
So please don't sit here with tears in your eyes
because with god they are all smiling,
Up there in the skies.

We know your world ended when the time came to part
but God picked them all to be next to his heart,
He chose your children from all of the rest
to be up in heaven for eternal rest,
Their time came early and you don't feel it's right,
But in Heaven they will be sleeping
And he will have kissed them goodnight

Mr R. Johnston

IN REMEMBRANCE

Katie Louise Martin 18th May 1989 - 8th January 2001

.....In loving memory of our dear daughter who was always a star on earth and who is now a star in heaven.

Everyone here has their own special memories of Katie, be they at home, at play or in school. We at Springwater have known Katie for many years and over that time have built up lots of memories of her.

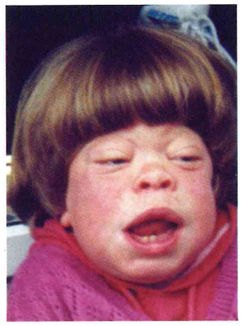
All our children are special, but Katie had a number of extra qualities. She was certainly bossy. How many times have we heard her say, "Come hear and sit down " -and that was just to the staff .We did have a discussion about employing her as a supply teacher as she most definitely had a way of getting people to do exactly as she wanted. Lee and Ross would do anything for her - and she would let them!

She had that special spark which allowed her to make friends in every age range. Every visitor would respond to her greeting of "Hello! What's your name?" and it even worked out of school. We as a class got to talk to many people who knew Katie and who came to befriend us all. So much for don't talk to strangers! Perhaps people picked up on her caring nature and real concern for others. "Are you alright Sue," wasn't just a phrase, it was said with real feeling.

Apart from talking, what did Katie like to do best? That's an easy one. She liked to sing, dance, and party. Take her to a disco and she was never short of a partner. Whilst she loved any music, she did have her favourite songs and she had ways of ensuring that we sang Baa, baa black sheep at every available opportunity-in the pool, in the park, and even on the back of a horse!

What a sense of humour! And what a filthy laugh! Once she started we all joined in, even when she laughed at something she wasn't supposed to hear and gave herself away. She got a lot of fun from hearing and repeating gossip--but honestly, Ray and Sandra, we only believed half of it! School is going to be quieter and we'll all miss Katie, but just for once,

I've had the last word!

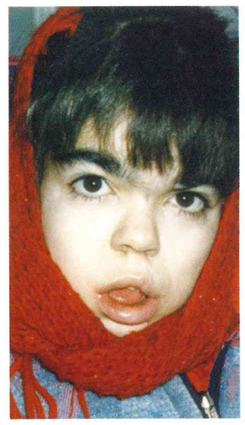


This piece was written by Katie's class teacher Mrs. Sue Thompson and she read it out at her funeral, and we feel it was so apt of our little Katie, who had Mucolipidosis Type II

Natasha Ebdon Macintyre 5 March 1979 - 16 January 2001

Our dear Natasha left us to go on to better things on 16 January. Her courage and determination was an inspiration to all who met her, or loved her. She never complained at all life threw at her, which was certainly a lot over the years, and all she ever asked for was a cuddle, a smile and to be cherished. She had the capacity to magic people to fall in love with her, and gave back all that love with a gentle smile or a poke or a tweak of the nose. Natasha knew no malice, or bad temper all of her life - which I am sure is an example that many of us cannot honestly own up to!

In the early days she used to run around like a plump human cannonball leaving a trail of destruction in her wake! This was never in frustration or anger but with gusto, joie de vie and the exuberance for life. Things that went crash or flying through the air did so with a joyful shout of glee, or shoulder shaking laughter! Many a time she would make her way to an available lap and "plonk" herself on it for a cuddle whilst swinging her legs with joy - severely bruising your shins with her callipers!
She taught us the important fundamental things in life. Through her, our lives have been enriched by the people we have met, the things she taught us and them and the pure perfection of a "true spirit". She also taught us that trick of pulling a tablecloth from a table and leaving the items intact on the table does not work!! We will remember Aunty Heathers' reassurance that it really didn't matter all the new jars of pickle, mayo and plates were smashed on the floor.



When she suddenly became so very ill it was a natural progression, that seemed destined, for us to all be around her at Helen House. We could not have managed without them. Not only did they fulfil 100 per cent of her every need, but they did the same for us. Through their loving support we managed to ensure that Tasha's final days, hours and minutes were full of warmth, security, comfort and love surrounded by her family. Her passing was serene and beautiful.

We will never forget her and feel warmth and a smile whenever we see her photo or think or talk of her. We know we'll be reunited one day. We know she is in heaven but are not sure what side of her personality is reigning - is she a peaceful gentle little Angel or is she flying around like a tornado and pulling everyone's halos!

FUNDRAISING

Sharesave Charities Event 2001

The MPS Society was delighted to learn that it had been selected as one of twelve charities to benefit from the maturity of the tenth and twelfth operation of BT's empoyee Sharesave scheme.

At a reception held on 25 January 2001 Christine Lavery and Ellie Gunary representing the Society were presented with a cheque for £10,500 by former Welsh International rugby player Ieuan Evans. Thank you to Gordon Rowe for putting the Society forward to benefit.

Marathon Runner

My New Years resolution for the year 2000 was to run a marathon for the MPS Society. I decided as I needed lots of training the Dublin Marathon which was run on October 30th 2000 would be my best bet. Along with James my brother who has Hunter syndrome and the rest of my family I collected £1200 which I was extreamely proud of. The training for the run and the actual 26.6 miles on the day was the hardest but most fulfilling thing I have done in my life and I felt a brilliant achievement.

Marie Stewart

MPS Coffee Morning

Three years ago when we decided to hold a coffee morning to make funds for the Mucopolysaccharide Society, it had to be delayed because the event coincided with the funeral of Princess Diana. This year we thought the weather would stop people coming out but we were lucky. It was the only fine day in several weeks of wet and windy weather and our friends did us proud. It is quite daunting to organise such an event within the family and hope people will support you for 'something that nobody has ever heard of and that they do not understand.' It is a measure of our friendship and their concerns for our granddaughter, Samantha, Hurler Sheie, that they all turned up or sent donations.

Samantha, and her sister Daniella were so excited at the prospect. They had spent several hours decorating very large fir cones ready for Christmas and sold the lot. On the day Samantha, aged 9, was in charge of the Tombola and Daniella, nearly 5, collected the money for the fir cones. Their mother helped sell other things and friends sold raffle tickets and did the coffee. My husband and I fitted round everybody else and worked very hard. We raised £521.45 which we were delighted with considering it was in our own home and only our own friends and acquaintances were invited.

We hope that the money will be put to good use.

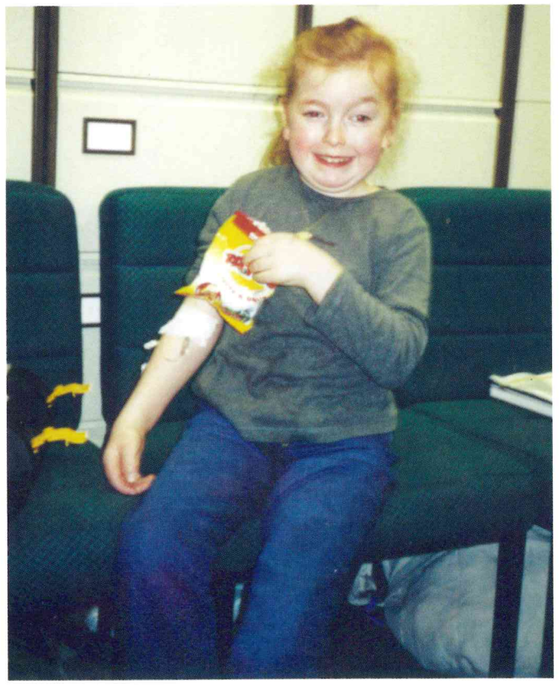
Samantha is about to start on clinical trials for the Enzyme Replacement Therapy in Manchester which means that we shall spend a great deal of the next six months travelling up and down the dreaded M6.

Samantha Brockie's Grandparents

Fundraising for MPS

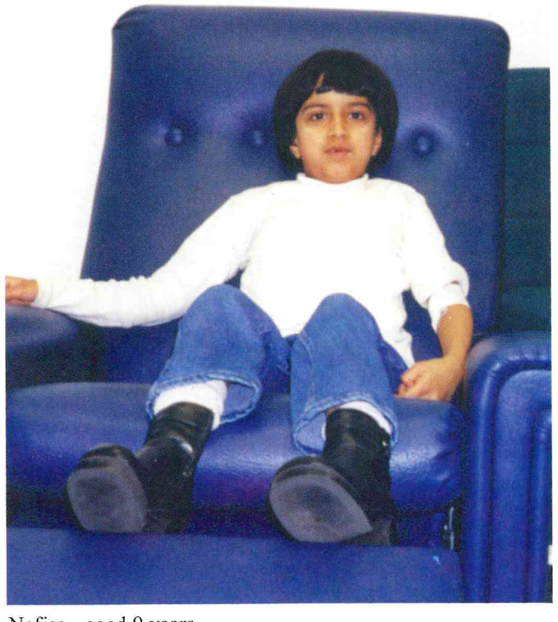
Thank you to everybody, MPS members their families and friends who are busy raising money for the MPS Society. The Society relies on its substantial income from fundraising to fund its support and advocacy services including the publication of the Newsletter and Booklets. If you can help the MPS Society by raising funds this would be most appreciated.

ENZYME REPLACEMENT THERAPY GALLERY

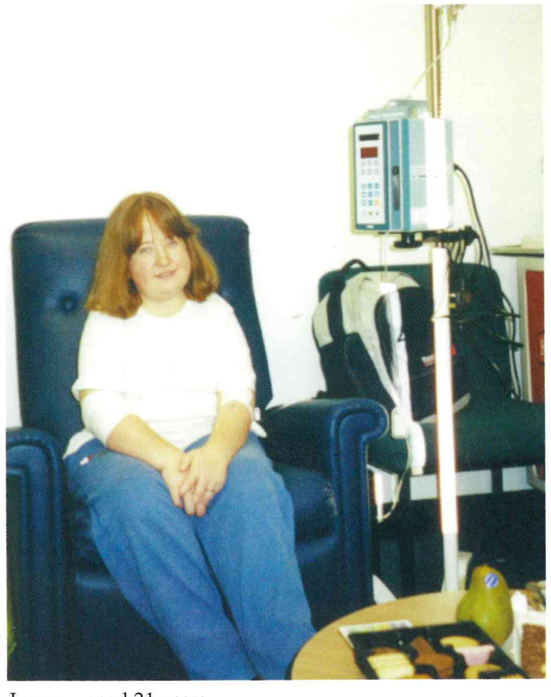


Deborah - aged 12 years

ERT Nurses Jean and Jane

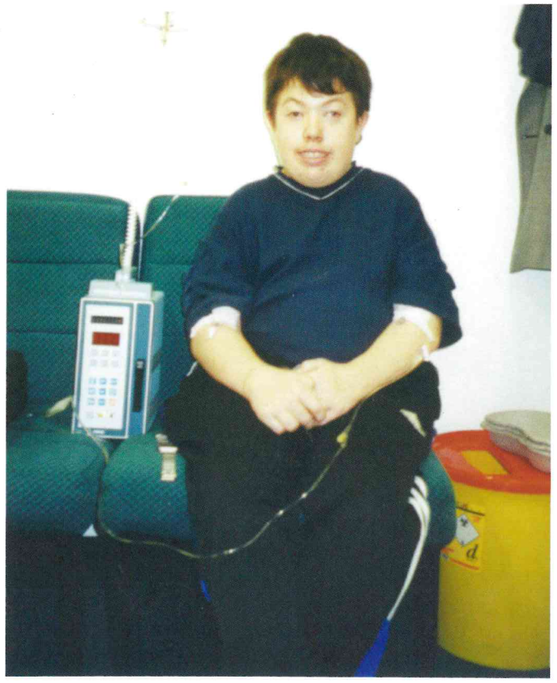


Nefisa - aged 9 years

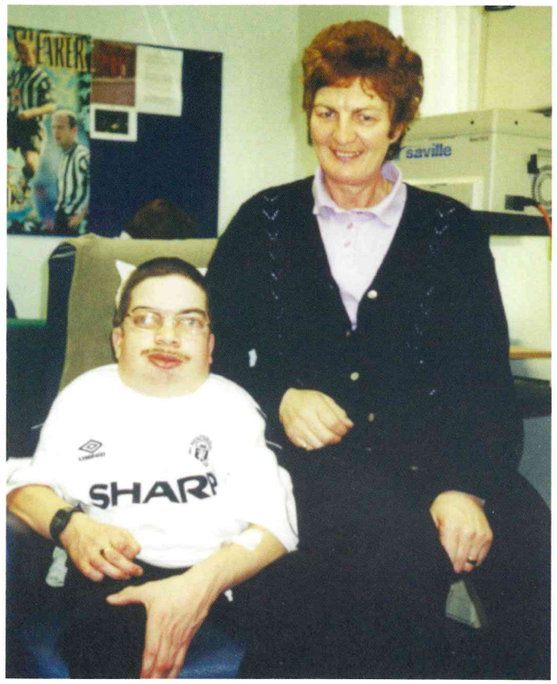


Joanne - aged 21 years

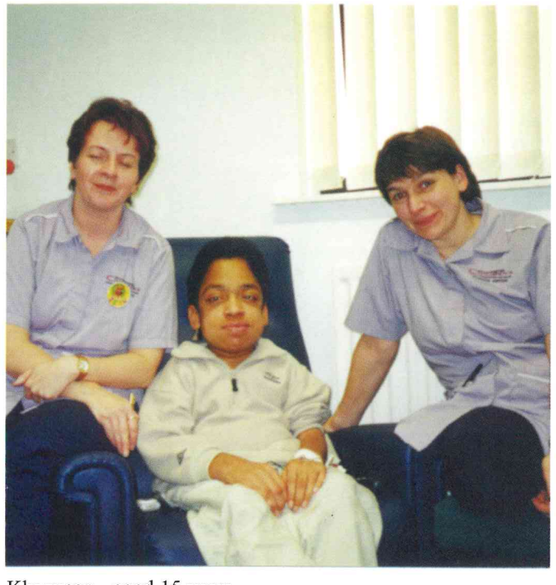
ENZYME REPLACEMENT THERAPY GALLERY



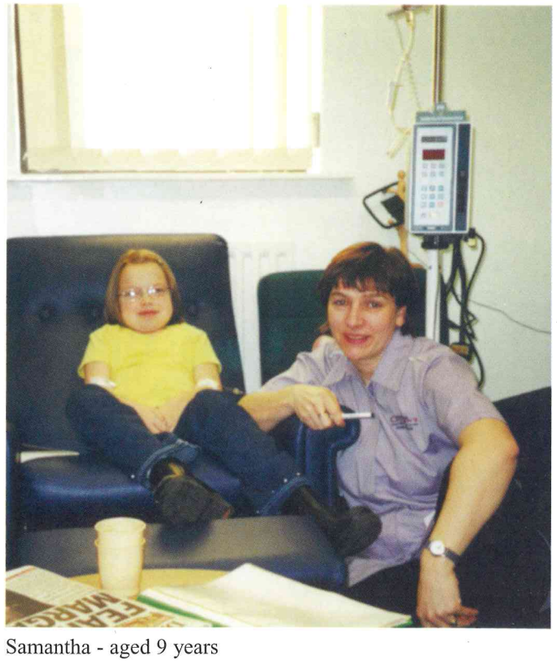
Myles - aged 20 years



William - aged 21 years and his mum Rachael



Khaquaan - aged 15 years



Samantha - aged 9 years

RESEARCH AND THERAPY

Clinical Trials for MPS I

We are now twelve weeks into the Enzyme Replacement Therapy Clinical Trial for MPS I Hurler Scheie and Scheie disease. There has been some disappointment that the trial planned for Great Ormond Street Children's Hospital London did not get under way. Happily two of the patients selected have transferred to Manchester.

The Society has been able to provide regular support at the Willink Genetics Unit to those on the trial and their families. All the trial candidates and their families have settled well into the weekly routine commuting to Manchester. As one mum put it, 'You've just about unpacked, done the washing, planned the meals for the next week and you're bags are packed and we're off again.'

Clinical Trials for MPS II

A Phase I clinical trial for MPS II, Hunter is due underway at Chapel Hill, North Caroline under Dr Joseph Muenzer within the next few weeks. This is a safety trial and alongside this, as we did for MPS I, we will be collecting clinical, psychological and social data from willing MPS II patients and their families to support this trial. If you would like to help please do contact Angela Ratcliffe or Alison Britton who will explain the information required, how it can help and who has access to it.

Clinical Trials for MPS VI

In October 2000, Biomarin began a Phase I Clinical Trial for rhSAB (recombinant human arylsulfatase B) in enzyme replacement therapy in MPS VI (Maroteaux Lamy Disease). The Phase I Trial follows successful completion of preclinical studies of rhASB that were conducted in a naturally occurring cat model of MPS VI. In the initial Clinical Trial, Biomarin is testing six patients at two dose levels during a 24 week patient evaluation period. The primary objective of the trial is to measure the safety rhASB. In addition, the trial will evaluate the effects of the treatment based on several physical, biochemical, and functional parameters known to be severely effected by the disease.

Enzyme Replacement Therapy for Fabry Disease

Two pharmaceutical companies Genzyme General and Transkarayotic Therapies (TKT) have both learnt that their respective Fabry disease treatments have been recommended for European approval by the Committee for Proprietary Medicinal Products (CPMP).

Fabry, a rare genetic disease closely related to the Mucopolysaccharidoses is characterised by a deficiency in the enzyme alpha-glactosidiase, which normally breaks down certain glycolipids. The deficiency allows the glycolipids to accumulate in the lining of the blood vessels within the kidneys, the heart, the skin and other organs, which can ultimately lead to kidney failure, stroke, cardiovascular disease, severe pain and numbness.

If the European Agency for the Evaluation of Medicinal Products (EMEA) follows the advice of the CPMP, which it usually does, it will clear the the drugs, Fabrazyme (agalsidase beta) produced by Genzyme and Replagal (agalsidase alpha) produced by TKT, within the next few months.

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The Use of Medication in the MPS Disorders

Alexander the Great is one of my heroes. He really was a great man. He had conquered half the world by the age of 21 and was also quite sensible when it came to medical care and this is one of his famous quotes.

"I'm dying with the aid of too many physicians"
- Alexander the Great (356BC-323BC)

Alexander the Great had it just about right; doctors can be very dangerous people. The majority of doctors are very kind and helpful, and are doing the best for your child but there will occasional times when things will go wrong.

Before treating any patient with medication a doctor must decide:

- In a person who has a rather complex disorder, should the doctor interfere with the patient at all? Is what he/she is proposing to do going to be to the patient's benefit?
- What alteration in the patient's state he/she hopes to achieve.
- That the drug being considered is capable of achieving that change.
- What other effects the drug may have and whether these may be harmful to the patient.
- Whether the likelihood of benefit out-weighs the likelihood of damage.

There are some specific problems when dealing with MPS children.

- In many situations it is very difficult to tell whether treatment is working, at least initially.
- The dosages often used in MPS disorders are not standard dosages. They are much higher, in some circumstances, to get the same effect.
- In some children who have learning difficulties, particularly if those learning difficulties are severe. When adverse events occur it is often very difficult to work out what is disease and what is side effects of the medication.
- With MPS Disorders there is very little literature to help one decide upon a rational approach to therapy. Often we are using medications by trial and error.

Most pediatricans are conservative in their prescribing habits. Most children's doctors think about medication very carefully, and are less likely to prescribe medication at first contact. This is important. It means there is less risk that the child is going to be given something unpleasant but, as a group, we often deny patients access to newer therapies. For example, most pediatricians don't use some of the newer anticonvulsants that are available, as they tend to use tried and trusted medications rather than trying something new.

There are also some problems with parents. This is a two-way thing; it is not just doctors who have problems. Some parents may cause problems because:

- Often parents expect an immediate result from medication. They go to the doctor, explain the problem, the doctor says we should try this medication and when it isn't working twelve hours later or the following day, they get upset. They often expect results of an over-ambitious nature. it is very unusual for you to go to a doctor with a problem, the doctor prescribes a drug and the problem goes away. The only exception would be an infection and antibiotics. It would be very unusual for this to happen with other, more complex, medical problems.

- The other problem is that many parents are actually non-compliant. I'm sure that you are all surprised to hear this, but there are many parents who actually don't give their children the medication. The commonest things that parents don't comply with are things like asthma treatments and epilepsy treatments. If we had an indicator test which allowed us to take a sample of urine and detect all the different drugs in the urine, I think we'd find that many of the children for whom we think we're prescribing medication, aren't actually getting the medication.

I'd like to home in on three particular areas which cause problems in MPS disorders and talk about the treatment of these with medication.

1. the management of challenging behaviour
2. sleep disturbance
3. epilepsy

Challenging behaviour
What do we mean by challenging behaviour? Challenging behaviour is:

- Hyperactive, "motor driven" behaviour where you see children on the go with a very short attention span, flitting from object to object, subject to subject, often with no apparent purpose and non-stop.

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- This is often associated in some patients with some element of physical aggression.
- Occasionally with property destruction
- Often with the children being non-compliant with instructions and with no sense of danger.

This is a fairly typical pattern of behaviour, for instance, for many of our Sanfilippo children and some of the other children with MPS. It's a very difficult problem for parents to deal with and a difficult problem for doctors to treat.

In the management of challenging behaviour, drugs are only part of the management strategy.

- Firstly, it is important to get some sort of behavioural assessment performed. Our psychology colleagues can have a look at the situation both inside and outside the home, make some form of assessment and suggest some strategies that parents can try .Although we have Challenging Behaviour Teams in the UK, their success rate with our patients is actually quite small. There are some children who do respond very well to a very carefully structured psychological approach to their behaviour management, but in my experience the numbers of those patients are actually very small.
- There are of course other important things you can do such as environmental modifications. This is a long-winded way of saying put your television out of reach, or your video on a top shelf and don't have your breakable ornaments within reach etc.
- Equally important is to try to get a break from this type of very wearying behaviour by making sure that you do get adequate respite.
- When you have thought about all of these, you can think about whether drugs are indicated and if so which drugs should be tried?

There are many different drugs that can be tried in the treatment of challenging behaviour and they come under different categories.

1. narcoleptics
2. anxiolytics
3. anti-depressants
4. psychostimulants
5. miscellaneous

When you see lists like this with lots of different types of drugs you know that's because not one works well. If this was a list with one drug on it you'd know that there was a wonderful treatment for challenging behaviours. Parents often find it difficult to accept that, in some circumstances, doctors (even those with the best will in the world) are not able to successfully treat the child's challenging behaviour. It's just too difficult to treat.

1. A **Narcoleptic** medication is a medication, which produces quieting. If you are given this medication as a normal individual it makes you indifferent to your surroundings, oblivious to your surroundings, and causes slowing of your thought processes and slowing of your movements. If children have too much of this type of medication, it's when parents say, "you've turned my child into a zombie!" There are a large number of narcoleptic medications and the following are the proper names for the ones we use in the UK.

Phenothiazines :
- Chlorpromazine (*largactil*)
- Thioridazine (*melleril*)

Butyrophenones :
- Halperidon (*serenace*)

Without a doubt the most prescribed for this kind of challenging behaviour in the UK is *melleril*. The reason for this is that *melleril* has a long history of use in this situation and, compared with the others, it's pretty safe even in high dosages. It's a good medication, quite old fashioned, but a good medication nonetheless. It's certainly better than, *halperidol* although *halperidol* is the second most common drug used in this group.

It is important to remember when you use this type of medication in children that the incidence of side-effects is very common.

General side-effects:
- The most common side-effect you see is drowsiness. A common complaint by parents is that I gave him this medication and you couldn't do anything with him, he was just too quiet.
- More worrying are the movement disorders, which you' sometimes see occurring as a side-effect of this type of medication. If you see children develop uncontrolled excessive motor activity then you have to stop the medication.

Less common side-effects:
- There are some other very rare complications. For instance, high dosages of *melleril* over a long period of time can damage the retina at the back of the eye, so visual loss can occur. This is not usual in the dosages that are used for MPS.
- *Largactil* has a problem in that it can damage the liver and you need to keep a check with liver function tests from time to time if you use the

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medication.

2. The second group of drugs which are used quite commonly in patients with MPS are drugs called **anxiolytics**. These are the valium-type drugs, the benzodiazepines. Anxiolytic drugs are drugs, which suppress aggression, they sedate patients and reduce anxiety. They are also quite useful because they have anti-convulsant and muscle-relaxing properties. So, in children who have a lot of excessive motor activity, muscle spasms or twitching, this group of drugs can be very good. By far the most commonly used drug of the anxiolytic type is diazepam (valium), because it is the one that doctors have the most experience with.

Side-effects:

- One of the major side-effects is the increased secretion which occurs in many patients. This can be a big problem, particularly in those children who have very poor swallowing. The secretion build-up which occurs can actually limit the use of this type of medication.
- Again, drowsiness and confusion are the commonest side-effects seen in clinical practice.
- This is a group of drugs where you get tolerance. You tend, once you start on this preparation, to have to increase the dosage over time to get the same effect. That's not addiction; it's tolerance. The children aren't addicted to the drug, they just metabolize it faster and need a larger amount of drug to get the same effect.
- It's also important to remember if you have been on them for a long time, you must withdraw them slowly. If you stop them suddenly you put the child a risk of developing seizures. So you have to reduce this group of medications very slowly.
- Respiratory depression is another problem to watch out for.

3. The other drugs, which are used, are Anti-depressants. **Anti-depressants** are drugs which are aimed at lightening your mood but can be useful in some children who have mood disturbance. It must be avoided in patients with difficult epilepsy. There are two common groups of anti-depressants, which are used widely for children with MPS disorders.

- Of the tricyclic anti-depressants, imipramine hydrochloried (*tofranil*) is very useful and is the treatment of choice for a particular form of epilepsy known as gelastic epilepsy. Gelastic epilepsy is epilepsy induced by laughing. So children often laugh and laugh uncontrollably and then lose their muscle tone and either drop to the ground or slump. They can injure themselves or hit their head when they do this. Gelastic epilepsy does not respond to normal anti-convulsants and is treated best by using *tofranil* or *imipramine*.

- The other group of drugs which I use quite a bit now are the serotonin re-uptake inhibitors such a fluoxetine (*prozac*).

Prozac is very good in children with mood disturbance. Some children with MPS II and III have quite wildly fluctuating mood - they'll cry for no reason, laugh for no reason, and it will swing almost minute to minute in some children. *Prozac* can smooth that out and can generally be mood lightening in patients, with this type of disorder. It's very difficult to get over to parents that when children cry they're not crying because they are in pain. They laugh and there's no reason for them to laugh. Because laughter is a sound we associate with very positive thoughts, we ignore it but when children cry, and the stimulus to do both is the same, we get upset and very anxious and tense and want it stopped. This fluctuation can often be treated quite successfully with *prozac*.

Side-effects:

- You have to be very careful. In children who have difficult epilepsy you shouldn't use *prozac*. There is quite a body of literature which suggests that *prozac* can make the control of epilepsy worse. It is not a big problem in a child who has well-controlled epilepsy on anti-convulsants but if you have a child who has difficult to control epilepsy, or multiple anti-convulsants you ought to avoid *prozac* because it can make that treatment process even more difficult.

There are some newer serotonin re-uptake inhibitors on the market and I have no experience of those preparations at all because I am a conservative pediatrician who denies my patients new therapies!

4. The other drugs, which come in and out of fashion very frequently, in the management of hyperactivity, challenging behaviour or Attention Deficit Disorder, are the **psychostimulants**. The most commonly prescribed amphetamine-related psychostimulant in the UK is methylphenidate (*Ritalin*). This is generally used as a therapy in children who have motor-driven attention deficit disorder or hyper-activity and is probably a very over prescribed preparation in most countries. The thing to remember about *Ritalin* is that its effective dose range is extremely wide. There

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isn't one fixed dose, which will work in any one child.

Side-effects

- It can cause alopecia or baldness. It is a side-effect which occurs almost immediately after starting treatment so you can be reassured if your child has been on this treatment for a long time.

5. The other drug, which is very useful, is carbamazepine (*tegretol*). It is a very highly effective anti-convulsant as well as a very good drug for treating challenging behaviour. It is useful in treating pain, which is neurological in origin. It is the treatment of choice in several conditions where you get neuralgia or pain originating in nerves. However, you must start with a very low dosage of *tegretol* and increase it slowly or side-effects are very common.

Side-effects:

- Causes dizziness and confusion, and ataxia with the child becoming unsteady, if children are put on too high a dosage, too quickly.
- It can also affect white blood cell count and this needs to be checked from time to time.

Sleep disturbance

Sleep disturbance is another important issue for many parents. There are different types of sleep disturbance or insomnia and I am only going to talk about two types today. Insomnia is when:

- you have difficulty in getting off to sleep but when you sleep you actually sleep all night. You wake refreshed in the morning
- The other type of sleep disturbance is characterized by frequent wakening. This type of sleep is not refreshing.
- There are some children who exhibit a combination of both types.

Parents often exaggerate sleep disturbance. When you have been deprived of sleep it is a terrible thing. If you get parents to keep a proper diary, like an anti-convulsant diary, you can go through it with the parents and see if the medications you are using are actually working.

Sleep diaries are essential:

- Sleep diaries should be kept at least 1 week prior to starting treatment and for at least 2 weeks on the treatment. Then the treatment can be assessed.
- Often when you keep a diary, you find surprisingly that the amount of sleep the child has is usually normal in a 24 hour period but it is just very fragmented. The child sleeps when he is tired. As soon as he is refreshed he wakes up, falls asleep again, wakes up. So the amount of sleep over a 24 hour clock might be normal for the child's age, except that a lot of that sleep is not occurring at a time when the parents are asleep. This causes the problem. Children with neuro-degenerative disorders often have sleep diaries like children who are blind. The reason why we fall asleep is because of the light-dark cycle.

There is a long list of medications, which are used to treat sleep disturbance. There is not one which will work on every child. The commonest groups are the benzodiazepine and cyclopyroline.

Sedatives:

Benzodiazepine:

- Nitrazepam (*mogodon*)
- Temazepam (*temazepam*)
- Lormetazepam (*lormetazepam*)

Cyclopyrroline:

- Xopiclone (*zimovane*)

All these have side-effects including tolerance and hangover the next day. There was great hope that Xopiclone would be the wonder drug in these cases. It works well for elderly people but not as well for our children. The next group are the hypnotics and chloral hydrates. I haven't put any dosages here because you start with the standard dosage and work up.

Hypnotics:

Chloral and derivatives:

- Chloral hydrate (*welldorm*)
- Triclofos (*triclofos*)

Avoid barbiturate and anti-histamines because of very high incidence of side-effect.

I never use barbiturates and antihistamines to treat sleep disturbances. In the past we have often used quite high doses of vallergan which can be very effective, but in my patients it seems to produce such a high incidence of side-effects, particularly the following day, that I don't use them at all.

Melatonin:

Most of my patients are on Melatonin to treat their sleep disturbance. Melatonin is a naturally occurring chemical synthesized from serotonin in the pineal gland in our brain and is secreted in response to the light dark cycle important in the initiation of sleep. When it becomes dark, our pineal gland starts to secrete serotonin and this activates that part of our brain, which sends us to sleep.

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It is now the drug of first choice for treating sleep disturbance in neurologically impaired children in the UK and can be highly effective in about three quarters of the children.

There are some important things about Melatonin which you need to know if you are going to use, or it won't work.

- Strict bedtime regime, with same sleep time every night .
- Given 30 minutes before normal sleep time or 12 hours before the child is supposed to wake
- Darkened room
- Effect within three days
- Three month trial
- Dose 2-5 mg/night - increase to 10mg if no effect in 7 days

Epilepsy
Some general points about epilepsy:
- Not all paroxysmal activity is epileptic
- There are many different seizure types and some children will have many different types of seizures
- Many different drugs used in treatment

No consistent treatment protocols

Study results in epilepsy in MPS in the UK
I'd like to share a small study that we did in the UK. We surveyed by questionnaire 237 patients with various types MPS:
- 148 returned (62%)
- information on 147 children

By Doctor Ed Wraith from a presentation at the 8th National Family MPS Conference in Australia

The reassuring thing is that the majority of children didn't have any seizures. Seizures occur mainly in children with MPS II or III. No children surveyed with MPS I or MPS IV had seizures. Of 36 children with Hunter syndrome, a number had a history of epilepsy. Of 56 children with Sanfilippo syndrome, 21 had epilepsy. Some of the children with Sanfilippo were quite young and may develop seizures later.

Seizure type:
- primary generalized or mixed - 75% - which responds well to treatment
- partial seizures (absent or tonic) - 15%
- gelastic -2 patients (1.5%)
- unsure - 8.5%

Drug Treatment:
- sodium valproate (epilim) alone - 60%
- carbamzepine (tegretol) alone - 20%
- together - 15%
- various including lamotrigine (lamictal), vigabatrin (sabril), imipramine (tofranil)

In conclusion, medication is going to be necessary for many, many patients. Generally speaking, medicines are safe as long as they are used with caution. Both doctor and parent should be realistic in their expectations from the use of medications. There is very little literature about the use of specific drugs in MPS patients. It is important to remember that what is effective in one patient may not work in the next. Each child is an individual and should be treated as such.

Morquio Disease - A Dental Perspective

An eight-year-old male patient presented for a routine oral examination and prophylaxis.

A comprehensive medical history obtained from the patient and his mother indicated that the child had been diagnosed with Morquio syndrome (Mucopolysaccharidosis IV). The medical history also included partial conductive and sensorineural deafness, and spin.al fusion of CI-C2 vertebrae. The patient routinely has echocardiograms performed to rule out valvular damage.However, no history of heart murmur or valvular pathology has been reported.

The patient exhibited several facial features consistent with Morquio syndrome. He has a flat nasal bridge and flared alae nasae, and a prominent lower third of the face notable for a square jaw and short neck.

An oral and radiographic examination was completed. Several significant findings were observed. The patient is a severe mouth breather, making dental examination and treatment difficult. The patient presented with a Class I molar occlusion. The maxillary anterior teeth are widely spaced. These multiple diastemata are consistent with the patient's broad flat palate.

The posterior teeth had a noticeable taper, with the cusp tips markedly pointed on the molars and canines.

The enamel of the teeth appeared to be of normal hardness but the surfaces were significantly pitted, allowing for accelerated staining. No caries were detected either clinically or radiographically. Radiographically, the enamel appeared less than one-quarter of its normal thickness but could be distinguished from its adjacent dentin. The dentin, pulp chambers, roots and root canal systems appeared within normal limits.

The patient receives frequent dental prophylaxis and fluoride treatments. This has minimised dental staining and prevented the development of dental caries. Also, because of the patient's difficulty breathing, his visits are kept short. Medical consultation is obtained frequently to determine if any valvular defects have developed. If such defects are detected, then antibiotic prophylaxis would be indicated following the guidelines of the American Heart Association.

Discussion

Specific dental findings often include flaring of the anterior teeth with pointed cusps and enamel pitting. The radiographic dental findings generally include decreased enamel thickness in both the primary and secondary dentition. The dentin, pulp chambers and root canal systems of all the teeth usually are normal. These patients may be more prone to dental caries because of enamel thinning and should have more frequent examinations and cleanings.

With improved medical and surgical treatment, patients affected with Morquio disease are living longer, more productive lives.

As dentists we are likely to be called upon to deliver care to this population of patients. Thus it is important that we fully understand all their special problems so that we can administer proper dental care.

Dr. Fitzgerald, general dentist USA
Dr. Verveniotis, dental surgeon USA

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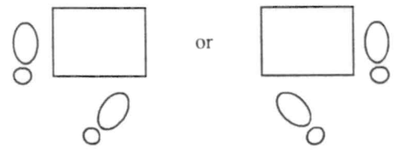
The Principles of Safe Lifting

Raising an MPS child involves frequent lifting - lifting your child in and out of cars, baths, chairs, from the floor and danger etc. etc. A back injury will not only make your life miserable, it will limit your ability to care for your child and will increase the workload of your partner/helpers.

By following these principals whenever moving your child, or any load the stresses on your spinal muscles, discs, joints and ligaments will be reduced, thus decreasing your risk of injury .

1. Plan the lift -determine the best technique for the task, being sure to avoid reaching, bending or twisting of your spine, if possible. Check the travel path is free of obstacles and spills. Take note of any change in ground elevation.
2. Position feet correctly -Feet should be a hip width apart, with one foot ahead of the other, in the direction of travel.



3. Get as close to the person/load as possible- this keeps your back from acting as a fulcrum and reduces stress. Avoid jerky movements.
4. Bend the knees and keep back straight - bend at knees and squat to lift. Never bend from the waist to pick up an object. no matter how small. By keeping the back in its normal arched position forces are more evenly distributed.
5. Keep head and shoulders up as lift begins - this helps to maintain the natural arch in the lower back.
6. Tighten stomach muscles as lift begins -this causes the abdominal cavity to become a weight bearing structure thus unloading spinal stress.
7. Lift with legs- use the powerful muscles of your legs to straighten the hips and knees. Perform all movements smoothly.
8. Move the feet if a change of direction is required -this eliminates the need to twist at the waist - a real no no !
9. Control lowing of the person/load- bend the knees and keep back straight.
10. If two people are performing the lift communication is vital- discuss the lifting route. lift together on a prearranged count.

INFORMATION EXCHANGE

Helping You to Stay Independent

A guide to long-term care services and benefits for people who live at home

One of the key commitments of the Modernising Government White Paper was to improve access to public services and make them more responsive to the needs of customers, Five groups, called Service Action Teams, were set up to address the problems facing individuals in trying to access services. One of these was the Service Action Team on long-term care at home.

The team identified the need for a guide for users and carers that 'sign-posted' the key long-term care and support services that they might need to call on to help them continue living at home.

Copies of this guide are now available and can be requested by email at doh@prologistics.co.uk or by post from the Department of Health, PO Box 777 London SE1 6XH. The guide is also on the Department of Health's website: www.doh.gov.uk/scg/sat.htm

Disability Rights Commission

The Disability Rights Commission (DRC) has been established by an Act of Parliament as an independent body to eliminate discrimination against disabled people and to secure equal opportunities for them across the UK. To achieve this they have set themselves the goal of: "A society where all disabled people can participate fully as equal citizens."

The services they provide are as follows:

- A helpline
- Legal advice and appropriate support for individuals
- Policy advice to government and others

They will also be working to:

- Establish a comprehensive information and advice service, through the DRC helpline, website and case working service
- Prepare and consult on codes of practice on provision for disabled people in education
- Take forward an action plan on key policy areas with particular priority for employment, education and better health and social care services for disabled people.
- Introduce a new conciliation service for disabled people and those selling goods or providing other services
- Establish authoritative baseline data on disability issues.

For further information or advice, contact: DRC Helpline Tel. 08457 622633 Textphone 08457622644 e-mail: enquiry@drc-gb.org Website: www.drc-gb.org

A Vote for Carers

National Association Need your Help!
Within the next few months the Government will be calling a general election. The build up to the election is a perfect time to raise awareness about the needs and rights of carers. That is why Carers National Association is producing a Carers Manifesto, setting out the main concerns of carers and proposing action that a future government should take to help the UK's six million carers.

At the last election, all major parties included pledges to support carers. Carers National Association wants to ensure that carers are included in forthcoming manifestoes. Carers National Association is contacting thousands of carers to ensure they have an opportunity to contribute their views to the manifesto. They want you to tell us the main issues that a future government should be tackling.

Everyone who helps them draft the manifesto will receive a copy of the finished product, along with advice on how it can be used to influence your local candidates. They will also be sending it to all political parties asking them to support its aims. Finally, they will launch the manifesto in the press. They hope the publicity will encourage 'hidden carers' to come forward and get involved.

Please write to them with your five main issues to be included in the manifesto and tell them what your own personal message to the next government would be. We understand how precious your time is but this will really help make an impact on the election.

Send your letters to: Carers National Association, A Vote for Carers, FREEPOST NWW 4556A, Salford M6 9DJ.

INFORMATION EXCHANGE

Naidex 2001 joins forces with Medtrade

Naidex, the UK premier event for those with disabilities and their carers, has teamed up with the organisers of Medtrade in the USA to run the first Medtrade UK at the NEC, Birmingham.

Running from l5th to 17th May 2001, these two events together make this UK's largest exhibition for disability, home healthcare and rehabilitation equipment.

There will be a full programme of free seminars over the three days covering topical issues such as sexuality, parents and family issues, accessible holidays and positive thinking. The professional programme targeting occupational therapists and other healthcare professionals will again be supported by Unison.

With hundreds of new products to see and lots of fun things to do please call Touchstone Exhibitions, Tel. (020) 8332 0044 to get your free tickets.

Charter of Children's Rights

To coincide with the lOth Anniversary of the UN Convention on the Rights of the Child, Mencap have produced a charter of children's rights. It states that children with learning disabilities have the same rights as all children. These rights included:

- The right to be valued and respected as a child first and foremost
- The right to services that support an ordinary life
- The right to be protected from harm
- The right to be protected from discrimination
- The right to personal privacy

For a full copy of Mencap's charter contact Mencap Public Liaison Unit, 123 Golden Lane, London EC1Y ORT Tel. (020) 7696 5593

Free RNIB Toy Catalogue

A Free '2000/2001 Toy Catalogue' has been produced by the Royal National Institute for the Blind, which includes toys which have been specially selected for their suitability for blind and partially sighted children. The toys featured are available in many high street shops and can be enjoyed by all children, so brothers, sisters and friends can enjoy them too. To order copies of the catalogue please contact:

Customer Services, RNIB, 224 Great Portland Street, London W1N 6AA Tel. 0845 702 3153

Carers and Disabled Children Act

Since the last MPS Newsletter, the Carers and Disabled Children Bill has been given royal assent and is now an Act of Parliament. The Act will come in to force in England in April 2001. We understand that it may be slightly delayed in Wales but probably by no more than a few months. The new Act will give carers, including parent carers, an enhanced right to an assessment of their own needs, as opposed to those of the person they care for. For the first time, Social Services departments will be able to provide direct services to carers including direct payments and vouchers for respite care. Young disabled people aged 16 and 17 will be eligible for direct payments to enable them to purchase their own services.

The Society for Mucopolysaccharide Diseases broadly welcomed the Act. However, the thorny issue of charging for services is still not resolved. Unfortunately, as law and practice currently stand, Social Services departments will be entitled to charge carers for any services they receive. Many carers feel that this is very unfair. After all, carers often give up work to provide care and are forced to live on low incomes. Caring for a child or adult at home saves the statutory services huge sums of money they would otherwise have to spend on home care and residential services. Why should carers be expected to pay for their own services too?

DONATIONS

Celebrating Holland - I'm Home

Carol Anthony writes: Many readers will, at some time, have read "Welcome to Holland" by Emily Perl Kingsley, who, as the founder of the USA Downs Syndrome Association, likened having a child with special needs to going on holiday and arriving in a different country to the one you had planned to visit. Here Cathy Anthony, a parent, advocate and presently the executive director of the Family Support Institute in Vancouver , offers her follow-up to the original.

" I have been in Holland for over a decade now and it has become home. I have had time to catch my breath, to settle and ad just, to accept something different than I'd planned. I reflect back on when I first landed in Holland. I remember clearly my shock, my fear , my anger, and the pain and uncertainty. In those first few years I tried to get back to Italy, my planned destination, but Holland was where I was to stay. Today, I can say how far I have come on this unexpected journey. I have learned so much more, but this too, has been a journey of time.

I worked hard; I bought new guidebooks; I learned a new language, and I slowly found my way around this new land. I have met others whose plans changed, like mine, and who could share my experience. We supported one another and some have become very special friends.

Some of these fellow travellers had been in Holland longer than I and were seasoned guides, assisting me along the way. Many encouraged me; many taught me to open my eyes to the wonder and gifts to behold in this new land. I discovered a community of caring -Holland wasn't so bad!

I think that Holland is used to wayward travellers like me and grew to become a land of hospitality, reaching out to welcome, assist and support newcomers. Over the years, I have wondered what life would have been like if I had landed in Italy as planned. Would life have been easier? Would it have been as rewarding? Would I have learned some of the important lessons I hold today?

Sure, this journey has been more challenging, and at times, I would (and still do) stomp my feet and cry out in frustration and protest. Yes, Holland is slower paced than Italy and less flashy than Italy, but this too has been an unexpected gift. I have learned to slow down in ways too, and look closer at things, with a new appreciation for the remarkable beauty of Holland with its tulips, windmills and Rembrandts. I have come to love Holland and call it Home.

I have become a world traveller and discovered that it doesn't matter where you land; what is more important is what you make of your journey and how you see and enjoy the very special, the very lovely things that Holland, or any land, has to offer. Yes, over a decade ago I landed in a place I hadn't planned yet I am thankful, for this destination has been richer than I could have imagined!

Charity Presses for Child Chair Provision

The charity Whizz-Kidz held a conference in November to press for the provision of powered wheelchairs for children under five years.

Jan Furumusu, a physiotherapist from the Rancho Los Amigos Rehab Centre in Los Angeles, said that children could benefit from the chairs if they were assessed to have spatial awareness and an appreciation of danger.
Ros Ham, director of children's services at Whizz-Kidz, said "The NHS is not providing. But the evidence is there that the right children with the right equipment can benefit from as early as 18 months.

"It can help their whole lifestyle. It avoids making them passive individuals by seven to nine years old when the NHS decides to provide. It allows them to behave like normal kids."
www.whizz-kidz.org.uk

In Car Safety Centre

The In Car Safety Centre specialises in all types of car seating. They publish a booklet entitled, 'car and bus seating for children with special needs'. They also have a showroom where all seats are displayed.
In Car Safety Centre, Unit 5, The Auto Centre, Stacey Bushes, Milton Keynes MK12 6HS. Tel. 01908 220909

FUNDRAISING

Homeopathy

Due to interest in Jacquie Hendry's article, 'Homeopathy for Victoria', here are two addresses for further enquiries:

The Homeopathical Medical Association
6 Livingstone Road
Graves End
Kent DA12 5DZ
Tel. 01474 560336

The Society for Homeopathy
4a Artisan Road
Northampton
NN1 4HU
Tel. 01604 621400

Sleep Scotland

Sleep Scotland is a charity to help the parents and carers of children with special needs who also have severe sleep problems. Its offers the following services:

- running Sleep Clinics which help families change their child's sleep pattern
- training Sleep Counsellors in specific techniques which are helpful for children with, for example, poor memory skills
- working with parent support groups across Scotland
- letting the public, professionals and politicians know the scale and consequences of this problem, so that services to families can be improved
- provide a daytime listening and advice service (0131 651 1392).
- A nightline (0845 603 1212)

Getting Britain Giving

The changes set out in the Getting Britain Giving package, announced by the Government in November 1999, came into force in April this year. The changes will make it easier for people to give and charities to receive. Some of the important changes are listed below. Make sure you bring them to the attention of anyone making a donation to your group so that you get the maximum benefit from their gift.

People who wish to make a donation to a charity simply have to confirm that they are a taxpayer. They can do this either verbally or in writing. Then tax can be reclaimed on their donation. For people giving a donation of more than £10, charities simply need to provide them with a Gift Aid Donation Form which they can hold in case the taxman asks for proof that they have made the donation.

Some donors may wish to open a Charity Account Card. This way, they can give more to the charity of their choice. Tax is recovered at basic rate and added to the person's account. Those who pay tax at the higher rate can claim the difference between the higher and lower rates back for themselves.

Give as You Earn is another way of ensuring charities receive more. Payments are taken from contributors before tax, so each £10 given only costs the donor £7.80. For higher tax payers, it will only cost £6 to give £10. And from now until April 2003, the Government has agreed to add an extra 10% to every donation made through Give as You Earn.

People who give shares to a charity as a donation are not only exempt from Capital Gains Tax but can also claim relief from Income Tax (up to 40%) on the market value of the shares they have given.

The changes announced in November 1999 included VAT exemption for fundraising events. Following consultation with charities. the types of events covered has been widened and include participative events and internet events. Also up to fifteen events of anyone type in anyone location can be exempt per year.

INFORMATION EXCHANGE

Mobility Roadshow 2001

What is it?
The world`s largest outdoor mobility event and essentially the Motor Show for disabled people. The Mobility Roadshow aims to give anyone with a mobility problem - drivers, passengers, adults or children - the chance to see what is available to help solve that problem and most importantly to try out and evaluate the options in a `no pressure` environment.

When is it?
Friday 15th, Saturday 16th and Sunday 17th June 2001. Open daily from 10.00 a.m.

Where does it take place?
The Transport Research Laboratory, Crowthorne, Berkshire (where they test the cars on BBC`s Top Gear). On site parking is available and accessible buses will be running from Reading Station to the show site. All on site facilities are fully accessible.

How much does it cost to get in?
Admission is free.

What sort of products will I see?
Latest cars and converted vehicles fitted with adaptations; infra red switching systems; lightweight, sports, power and manual wheelchairs; hand controls; hoists; car seats; commercial vehicles; battery chargers; trikes and scooters; walkers; rotating car seats; specialist wheels and tyres; suspension systems; power steps; ramps; seat clamps; wheelchair restraints; floor tracking; hi-tech mirrors; lifts and numerous gizmos and gadgets to make mobility easier for disabled people and their carers.

Independent Living

The Independent Living events are the best place to see the latest products in the market, update your knowledge and find out about changes in the industry.
- See 100's of products from leading brands
- Sample free lifestyle therapies
- Attend free seminars or talks

With so much to do and see, a visit to Independent Living is not to be missed.

If you specify, purchase, recommend or use rehabilitation products or services, then visit the Independent Living series of events.

Order your free tickets today:
0870 429 4372
www.independentlivingevents.co.uk

Action on Entitlement Campaign - New Code on Special Education


Lauren Cawthorne

Alarm Call to MP's
In the information exchange section of the last two newsletters we have included articles about protests at the proposed new SEN code. Protesters from the umbrella group "Action on Entitlement" gathered at 10 Downing Street on 13 October to hand a letter to Prime Minister-Tony Blair claiming the revised code which advises local education authorities on SEN provision is misleading.

Following this action and further pressure by voluntary organisations including the MPS Society the Government has conceded that they will keep the word "specify" instead of "set out" in the regulations. Although this is good news the MPS Society understands from the Action on Entitlement Campaign that a draft of the additional new guidance in the code is not available and that it would appear likely that the guidance will say special educational provision should only be "quantified as necessary" and that it "may often" need to be expressed in terms of hours, equipment or personnel. There will be no necessity to put hours or specific details in statements.

The MPS Society's Development Team through supporting families has already experienced first hand the Action on Entitlement campaign's concern that many LEA's will take advantage of any wording in the code that allows them to avoid quantifying provision. This is before this has become Law!

All MPS parents who registered to support this campaign last year will have already received further correspondence advising on how to support the next stage of the campaign. If you would like to register to support this campaign and receive further details please contact the MPS office.

Ellie Gunary
Assistant Director

DONATIONS & FUNDRAISING

Donations

The Society is grateful to the following who made a donation to the Society

Union of Catholic Mothers
Cytogenetic DNA Services
Benham Charitable Settlement
D Hayward & C Hayward
Miss Joy Weston
Fiona Thompson
Gordon Fraser Charitable Trust
Mrs J Russell-Cook
Mrs Shaw
Hallmark Cards
R F Walker
De Clermont Charitable Co. Ltd
British Telecom
Mr Wilford
Mr & Mrs Holderness
Cliff Richard Charitable Trust
Three Star Engineering Ltd
Dixons Group plc
3M United Kingdom plc
Jessica Stuart
Mr & Mrs Summerton
Mars Electronics International
Mary Barralet
Damian Lavery
Spiffing Stationery
Charity Flowers
Greater Bristol Foundation
Clydebridge Steel Works
Mr N Atkinson
Mr & Mrs McTiffin
Mrs Dorothy Duckett
Egremont & District Sunday Football League

COLLECTION BOX
Trull School of Dancing – Taunton
Books Etc – Queensway
Roland Avenue Post Office

Fundraising

The Society is grateful to the following who held fundraising events

Chris & Dawn Jones– Charity Fair
Clwyd Compound – Xmas Raffle
Coronation Dental Practice – Dinner Dance Raffle
Tracy Stokoe – Webb Ivory Catalogue
St Georges Hill – Golfing Day
Charlene Murray – Coffee Morning
Haddenham mummers – Performance of play
A Trevor – Run
Pete Robjohns – Nottingham Full Marathon/Gt North run
Marie Stewart – Dublin Marathon
Karen and Andrew Weedall – Webb Ivory Catalogue
Towersey Morris Men – Performance of Mummers Play
Holt Town Trust – Collection at Carol Concert in Village Hall
Mr & Mrs Thompson – Junior Youth Club
Peggy Darper/Amanda Stuart – Car Boot Sale
Gryffe High School – Xmas Dance
Lyn Longhorn – Skittles Evening
Marina and Dave – Car Boot Sales
Clyde & Co – Tennis Tournament
The McKerr Family – Sponsored Drink/Music Evening
Viscountess Cowdray – Tennis Tournament
Alan & Fiona Byrne – MPS Dance
Mr & Mrs A Del Tufo – Amersham Museum
Thornbury (shamley) Ltd – Tennis Tournament

KINDLY DONATED TO THE MPS SOCIETY IN MEMORY OF
Shaun Darren Osment
Katie Louise Martin
Edward Nowell
Katie Devine

STAMPS/FOREIGN COINS
Mrs F McConnell
Miss L Ricketts
Lucy Lavery

Donate Online

Donations to the Society for Mucopolysaccharide Diseases can now be made online through our secure server at

www.mpssociety.co.uk

MANAGEMENT COMMITTEE

NEWSLETTER DEADLINES

SPRING
31 March 2001

SUMMER
30 June 2001

AUTUMN
31 September 2001

WINTER
17 December 2001

Do let us have your family stories and any helpful hints you would like to share with our newsletter readers. if you have a question that you would like to see answered in a future edition of the newsletter, please do write to us.

To submit information to the newsletter please send materials (preferably via e-mail for text) and mail photos to the address below.

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.

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MPS CONFERENCES

THE SOCIETY FOR
MUCOPOLYSACCHARIDE DISEASES

21 years of Bone Marrow Transplants for MPS and Related Diseases

This conference will address the ongoing medical and social needs of children and young adults who have undergone bone marrow transplant for a mucopolysaccharide or related disease

Hilton Hotel
Northampton

8th June 2001

1980 - 2001

Friday 8 June 2001
9.45am - 4.30pm

Chair
Professor Bryan Winchester

Welcome *Professor Bryan Winchester*	**Head of Biochemistry** *Institute of Child Health London*
A Historical View of BMT *Dr Charles Peters*	**Associate Professor of Paediatrics** *University of Minnesota USA*
Spinal Surgery – Development & Outcome *Mr Brad Williamson*	**Consultant Paediatric Orthopaedic Surgeon** *Royal Manchester Children's Hospital*
Managing MPS Necks *Mr Richard Cowie*	**Consultant Paediatric Neurosugeon** *Royal Manchester Children's Hospital*
Lower Limb Surgery – Hips & Knees *Mr Esmond Foggarty*	**Consultant Paediatric Orthopaedic Surgeon** *Our Lady's Hospital for Sick Children Dublin*
Corneal Transplants for MPS I *Professor Anthony Bron*	**Professor of Ophthalmology** *Nuffield Laboratory of Ophthalmology Oxford*

Lunch

Chair
Dr Fiona Stewart - Belfast City Hospital

Endocrinology and the role of Growth Hormones *Dr Uma Ramaswami*	**Consultant Paediatrician** *Addenbrooke Hospital Cambridge*
Addressing the Psychological aspects of BMT *To be confirmed*	
A Parents Perspective *Angela Brown*	**Mother of Aiden** *Edinburgh Scotland*
Education – Reaching Full Potential *Christine Lavery*	**Chief Executive MPS Society** *Amersham England*
The Future of BMT *Dr Rob Wynn*	**Consultant Paediatric Haematologist** *Royal Manchester Children's Hospital*

Panel Discussion Chaired by
Dr Ed Wraith - Royal Manchester Children's Hospital
Dr Ashok Vellodi - Great Ormond Street Children's Hospital

Professor Anthony Bron	*Dr Fiona Stewart*
Mr Richard Cowi	*Dr Ashok Vellodi*
Mr Esmond Foggarty	*Dr Rob Wynn*
Dr Charles Peters	*Mr Brad Willamson*
Dr Uma Ramaswami	*Professor Bryan Winchester*

THE SOCIETY FOR
MUCOPOLYSACCHARIDE DISEASES

19th National Conference on Mucopolysaccharide and Related Diseases

Hilton Hotel
Northampton

14th - 16th September 2001

Saturday 15th September 9.15 am - 12.45 pm

Welcome from the Chair
Dr Charles Pennock
Consultant Paediatric Chemical Pathologist (retired)

Genetics for Beginners *Professor Bryan Winchester*	**Head of Biochemistry** *Institutute of Child Health London*
A Personal Experience of Pre-Natal Diagnosis *Jackie Edwards*	**James' Mother** *Cardiff Wales*
Managing Ear Nose and Throat problems *Mr Mike Rothera*	**Consultant Paediatric ENT Surgeon** *Royal Manchester Children's Hospital*
Feeding Difficulties *Martina Ryan/Elizabeth Singhi*	**Speech and LanguageTherapist** *Great Ormond Street Children's Hopital*
After 16 - What next? *Alison Cowan*	**Information Manager** *Family Fund York*
A Grief Support Programme for Children *Allyson McCulloch*	**Senior Practitioner** *Winston's Wish*
We Are Siblings *Emma Hill and Claire Moraid*	**Claire's sister** *Lancashire* **Richard's sister** *London*

Satellite Seminars

Home Adaptations		
Overview	*Dr Fiona Stewart*	*Belfast City Hospital*
Homes fit for children	*Ginnie Shaw*	*HODIS*
Alex's Story	*Ellie Gunary*	*MPS Society*
Other Therapies		
Aromatherapy	*Sarah Horswell & Debbie Crocker*	*Naomi House Children's Hospice*
Play Therapy	*Ailsa Foster*	*Rainbow Trust Children's Charity*
Physiotherapy	*Pauline Hensman*	*Royal Manchester Children's Hospital*

Sunday 16th September 9.30 am - 11.30 pm

Palliative Care		
Overview	*Dr Ed Wraith*	*Royal Manchester Children's Hospital*
Facing Loss	*Jenni Thomas*	*Child Bereavement Trust*
Developing a Care Plan	*tbc*	*MPS Society*
Looking after me	*tbc*	
Child Bereavment Trust		
Child Bereavement Trust	*Angela Ratcliffe*	*MPS Society*
Jeans for Genes Research	*Dr Guy Besley*	*Royal Manchester Children's Hospital*
	Dr Maureen Cleary	*Royal Manchester Children's Hospital*
	Dr Robert Coffin	*University College London*
Preimplantation Diagnosis	*Professor Joy Delhanty*	*University College London*
Personal Experience of Enzyme Replacement Therapy	*Steve & Amy Holland*	*MPS Society USA*
Enzyme Replacement Therapy Update	*Dr Ed Wraith* *Dr Ashok Vellodi*	*Royal Manchester Children's Hospital* *Great Ormond Street Children's Hosp.*

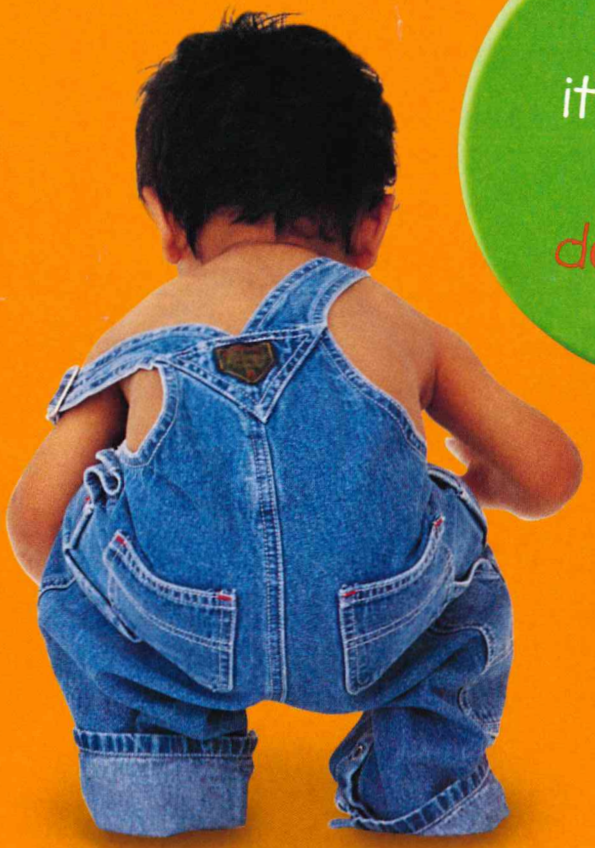
Jeans for Genes Day
5th October 2001

I'm ready...

Get ready to throw out the usual dress rules and jump into your jeans on **Friday 5th October** – this year's national **Jeans for Genes Day**!

It's great fun to join in and so easy to organise – simply wear jeans at home, work or school and donate just £1 to the appeal. Ask your friends, family and colleagues to join in too.

This year's appeal aims to raise at least £2.5 million for research into genetic disorders and to provide support services for families across the UK.



It's fun, it's easy, it's a great cause... don't miss out!

To find out more, simply register for a FREE fundraising pack TODAY!

Call freephone

0800 980 4800

or visit

www.jeansforgenes.com

...are you?

A national appeal to support the Society for Mucopolysaccharide Diseases and four other national charities.

Net proceeds from the Jeans for Genes Campaign will be distributed among the five charities.