Newsletter

The Society for Mucopolysaccharide Diseases

National Registered Charity No. 287034

Summer 2003

CONTENTS

Outside Cover: Barry Wilson and Ellie Gunary

'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 Storage 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 and related diseases**
*** To bring about more public awareness of MPS and related diseases**
*** To promote and support research into MPS and related diseases**

How does the 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

National and International Conferences
Held annually and offering individuals and families the opportunity to learn from professionals and each other

Sibling Workshops
Specialist activities for siblings who live with or have lived with a brother or sister suffering from MPS or a related disease

Information Resource
Publishes specialist disease booklets and other resources including a video

Quarterly Newsletter
Containing information on disease management, research and members' news

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.

Director's Report

Christine Lavery

I write this report having just returned from the Society's most sucessful National MPS conference ever. Over two hundred and fifty members, and children as well as one hundred professionals participated in a weekend that offered something for everyone. For children a fun packed sibling workshop followed by outings to Drayton Manor Park, Northampton Megabowl and Gulliver's Land. Young 16+ adults enjoyed dinner on a barge on the Grand Union Canal. The three conferences on Saturday offered an insight into all aspects of palliative care, independent living and Fabry disease as well as on Sunday an update on research and treatment. At a reception prior to the gala dinner the Society's Chairman, Barry Wilson spoke warmly of the achievement by Biomarin Pharmaceuticals and Genzyme corporation in bringing the first enzyme replacement treatment for MPS to the marketplace. Alduazyme™ an Enzyme Replacement Therapy for MPS I was approved in June by the European Medicine Evaluation Agency for its use throughout Europe. Julie Kelly representing Genzyme thanked the investigators, patients, their familes and the Society for all their help in this wonderful achievment.

This occasion also afforded the Society an opportunity to show our appreciation to individuals who over the years have contributed in the most exceptional way to those suffering from MPS, their families and the Society. Each was presented with a blue rose (or as near as you can get to blue) 'Rose of the Year' for 2003.

Some two weeks prior to the conference one hundred and fifty MPS members and invited guests gathered in the members' dining room of the House of Commons to celebrate the Society's 21st Birthday year. A series of speakers spoke glowingly on the Society's work and reflected on the achievments of the past 21 years as well as looking into the future. We are grateful to British Airways for donating three pods allowing seventy members and guests to finish the day with a ride on the London Eye.

Finally, this bumper newsletter is packed with information and I would particulary like to highlight the Society's publications which we hope will benefit not just our members but the many professionals working with children and adults with MPS and other related diseases.

Introducing Forthcoming Research on BMT

Cheryl Pitt

On April 1st I started work at the Society as Advocacy Support Researcher. Over the next three years I will be conducting a variety of research projects, which will explore the psychological outcomes of bone marrow transplant, enzyme replacement therapy, and clinical trials. Those of you who have had experience of BMT will already have heard from me, as the first piece of research I have embarked upon, explores the social and psychological issues associated with bone marrow transplant. Those of you who are receiving ERT or who are about to take part in clinical trials, will also hear from me in the not too distant future. So, I look forward to meeting as many of you as possible over the next couple of years, and I hope to encourage you to give up a little bit of your time to talk to me, so that I can carry out this very important research to the best of my ability.

My background is in psychology and health psychology, and I have worked in family research for the past six years. However, this has mostly been in academic settings, like hospital research units, or universities. So, this is my first time working at a charity, which is a very exciting prospect, as it gives me the opportunity to follow-up the research projects with on-going family support. Although the research I am conducting for the MPS Society will address some issues relating to surgical procedures, medical interventions, and physical disability post-BMT, the main focus of the research will explore the social, emotional and psychological issues that surround such experiences. This will include the decision-making process pre-treatment, your expectations of the treatment, and subsequent outcomes of treatment. As a result, it is hoped that we will have a greater understanding of the needs of families affected by MPS diseases, especially those families that undergo therapeutic interventions. Furthermore, since new therapies, such as enzyme replacement therapy and gene therapy are virtually upon us, it is imperative that we find out about their psychological impact, so that we can better meet needs at the decision-making stage, throughout treatment, post-treatment, and throughout life.

My time at the MPS Society has, so far, been a huge learning experience for me. I was fortunate enough to attend the Clinical Management of MPS I National Symposium in Naples only ten days after I had started work at the Society, which in my case, turned out to be more of a crash course in mucopolysaccharide diseases than a conference. Suffice to say, I learnt a lot, I met some key figures in the field, and I ate a lot of mozzarella cheese! I am now making some headway with the research and I will be in touch with many of you in the near future. I look forward to meeting you.

News From The Management Committee

Christine Lavery

The Trustees met in January and May and considered the following matters:

Jeans for Genes Research

In January 2003 Dr Rob Wynn and Dr Ed Wraith from the Royal Manchester Children's Hospital and Dr Claire Beesley from the Institute of Child Health, London made presentations to the Trustees on the progress made in the second year of their research programme grant awarded by the Society from money raised by Jeans for Genes. A research update by Claire Beesley can be found on page 45.

Policies

Over the last six months the Trustees have agreed and reviewed the following policies: Financial Controls Policy, Data Protection Policy, Moral and Ethical Policy, Embryo Research Policy, Working Away from the Office Policy, Child Protection Policy, Moving and Handling Policy, Volunteer Conduct Policy, Financial Assistance Scheme and Reserves Policy.

Annual Year End Accounts

In May the Trustees adopted the Society's year end accounts prior to being presented at the Society's Annual General Meeting.

Support and Advocacy

The Trustees received reports from the Assistant Director and Director on the support and advocacy work being undertaken. Trustees also reviewed updates on the MPS Annual conference programme and the planned Remembrance Day in the Childhood Wood. A proposal to hold an Expert Meeting on Orthopaedic Management of Mucopolysaccharide and Related Diseases funded by Jeans for Genes and jointly hosted by Dr Ed Wraith and Dr Charles Peters from Manchester, and University of Minneapolis respectively was also agreed.

Wilma Robins and Lynne Grandidge being recognised for their service to the Society

Annual General Meeting

Christine Lavery

The Annual General Meeting of the Society took place at the Alton Towers Hotel on 10 May 2003 at 09.30. Forty one members attended. No apologies received.

The minutes of the 2002 AGM meeting were distributed in advance to those members present. The minutes were accepted as true and accurate.

The Chairman, Barry Wilson, presented the Trustees' Report. This is published in the Society's Annual Report for the year ending 31 October 2002.

The Treasurer Judith Evans, presented the statement of accounts for the financial year 31 October 2002, the details of which are also to be found in the Annual Report. Judith thanked the members for their support in fundraising and encouraged the members to help the Society in any way they can. Trustee Chris Holroyd, showed the breakdown of charitable expenditure, sources of income and levels of direct charitable and indirect charitable expenditure for the financial year. It was acknowledged that the Society's indirect charitable expenditure was less than 6% of its total income in that financial year.

It was proposed and seconded that the auditors, McLintock and Partners, Chester, be appointed the Society's auditors for the financial year ending 31 October 2003.

During the Election of Officers, the Chairman, set out the objectives of the Society and the role of Trustees stating that co-option gives the individual an opportunity to get to know the work and the time involved before putting themselves forward for election. Ann Green, Wilma Robins, Bob Devine and Judith Evans stood for election and were duly elected as Trustees.

As there was no other business the Chairman, Barry Wilson closed the meeting at 09.50 and wished everyone a wonderful day out at Alton Towers.

Lauren Cawthorne meets the Tweenies at Alton Towers

MPS Birthday at the Houses of Parliament

Antonia Crofts

This year, in 2003, the MPS Society is celebrating its 21st Birthday Year with a number of events to mark this special occasion.

On Wednesday 4th June 2003 the Society organised Afternoon Tea at the House of Commons. Hosted by Andy King MP, Member of Parliament for Rugby and Kenilworth, the Tea was held in the Members' Dining Room at the House of Commons.

In the months and weeks leading up to the day the Society had compiled a guest list of approximately 120 people. By all accounts this was not meant to be an exhaustive list but was simply designed to merely represent a cross section of the vast number of people who have contributed to the work of the MPS Society over the last 21 years. Given the limited space available we could not possibly include all those who we would have liked to invite to share in this special day.

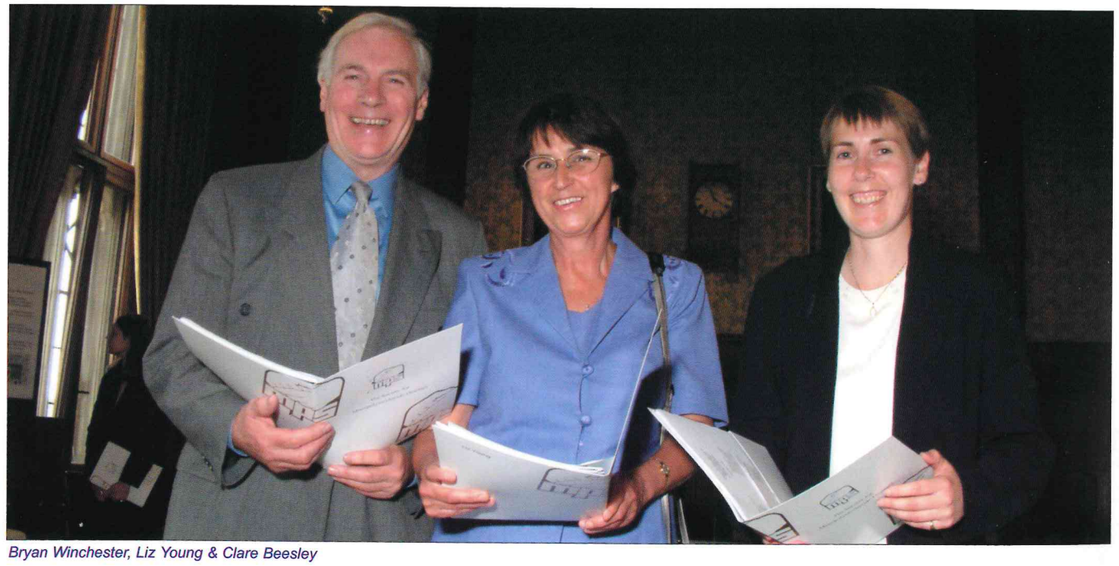
Amongst those who were invited were many doctors and nurses specialising in the field of MPS and Related Diseases and who have close links with the Society. Other guests included fundraisers who spend time and effort raising money for the Society, staff from the Jeans for Genes campaign, researchers, representatives from the pharmaceutical companies with whom the Society works very closely, volunteers who generously give up their spare time to take part in the children's outings at our MPS conferences and of course, the MPS individuals and families for whom the Society provides support. As befitted our venue, the Society also asked each MPS family to extend an invitation for the afternoon to their local MP, a number of whom did manage to drop by for a cup of tea and a chat with their constituents!

Commencing at 4pm the guests were firstly welcomed by Andy King MP. Barry Wilson, Chairman of the Society's Trustees, introduced the Society followed by another long-standing Trustee, Wilma Robins, who talked on the historical perspective of the Society. Professor Bryan Winchester from the Institute of Child Health spoke of the Society's role in medical research followed by Dr Ed Wraith who talked about clinical management in partnership with patient advocacy. Drawing the speeches to a close Sue Peach, Trustee, spoke on facing the future and Andy King gave the closing remarks and toast.

Joanne Evans who has Morquio disease cut the fabulous cake made especially for the occasion with a 21st Birthday logo emblazoned on the top while guests were able to begin their cream teas in earnest!

The Society was very privileged to have the use of the Members' Dining Room for Afternoon Tea and from the feedback we have received from the guests who attended we believe it was a wonderful backdrop in which to celebrate. As always, events such as these give people the opportunity to chat, exchange information, catch up with old friends and acquaintances and to reflect on the Society, its evolution, its future and the numerous people who have been involved and who hopefully continue to be involved along the way without whose hard work, commitment and dedication the Society would not have reached 21.

As the afternoon drew to a close in the early evening we said goodbye and thank you to all those who had attended. Several guests had taken up the opportunity of places on the London Eye which the Society had been lucky enough to receive free of charge from British Airways London Eye. We extend our gratitude to the London Eye for supporting the Society and helping us celebrate our 21st Birthday in style!



Bryan Winchester, Liz Young & Clare Beesley

Top: Andy King MP with Sue Peach & Guest Bottom: Helen Patterson (MPS volunteer)with Joanne Evans

Top: Christine Lavery with Dafydd and Elinor Wigley Bottom: Wilma Robins

MPS 21ST BIRTHDAY

Top: MPS Staff team at the House of Commons Bottom: MPS guests in the London Eye Pod

New Members

The Society has recently been contacted by Margaret Cessford who has Fabry disease. She lives in Scotland.

Michael and Nichola's daughter Evie has recently been diagnosed with Mucolipidosis. Evie is seven months old. The family live in the North East of England.

The Society has recently been contacted by Sue Jenkins who has Fabry disease. She lives in East Anglia.

Catherine and Andy O'Neil's daughter Keira, has been diagnosed with Hurler disease. Keira is 3 years old and had a bone marrow transplant 18 months ago. The family live in West Yorkshire.

The Society has recently been incontact with Tina and Padraic whose daughter Sophie has recently been diagnosed with Hurler disease. Sophie is 1 years old and lives in the South of England.

The Society has recently been contacted by Ashley Rigg who has Fabry disease. He lives in Lancashire.

Births

Deby & Mark McAfee had a little girl Faith Jade McAfee born on the 5th May 2003

Gordon and Norma Broadley had a little girl Megan Broadley born on the 27th May 2003

Mr Muhammad and Mrs Zadjai had a little girl born on the 30th May 2003

Louise and Adam Turner had a little girl Eliza Turner born on the 14th June 2003

Deaths

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

Albert Brown who suffered from Hurler disease 20 August 1994 - 18 April 2003

Lucia Dawson who suffered from Hurler disease 2 June 1990 - 22 April 2003

Shazia Begum who suffered from Morquio disease 6 April 2000 - 2 June 2003

Andrew Wilson

It is with considerable sadness that we remember Andrew Wilson, son of our Chairman Barry Wilson and his wife Babara and brother to Joanna. Andrew was one of the British servicemen killed in the Gulf when two Royal Navy Sea King helicopters collided. Andrew Wilson aged 36 was from 849 squadron based at the Royal Naval Air Station Culdrose, near Heston, Cornwall and was serving as a Royal Navy Pilot on the Ark Royal. Andrew left a wife, Sarah who is expecting their first baby in August. Our thoughts are with Barry, Barbara, Joanna and Sarah at this time.

As we remember Andrew our thoughts are also with other service families in the MPS Society and members with family and friends living in Iraq.

Bringing Issac Home

Richard Dunn (to be continued in the Autumn Newsletter)

I feel like an astronaut sitting by a sealed porthole gazing out at the universe. "I love him for who he is now but wait in anticipation to see the man he will become."

Bandaged heads; jerking limbs. Stifling heat. The smell of disinfectant. Metal cots that rattle when you touch them. Oxygen masks hanging limply on the walls. Heywood, the neurological ward. Kay and I have been here for a week. Isaac our grandson has hydrocephalus and needs urgent surgery. Sleeping in chairs, Louise our daughter and her husband Adam work together to care for their son. Kay and I live out of a suitcase. Offering support. Being in the only place we could be.

"I don't suppose they'll ever know why he's had all these problems," Adam speculates. "Everything's going well now, you and Kay could go home. Lou and I are seeing the paediatrician tomorrow. I don't expect she'll have anything new to tell us". We go home.

The morning's going well. Last day of term, "Thanks Pete, Sue. Excellent work this term". "Have a great holiday. Crete! Very nice". "Oh yes he's doing well. The operation was a success. Thanks for asking".

Back to my office. Still smiling. "Hi Doreen."
"It's Kay, Dick. I'll put her through to your room".
No hint but she already knows. Sun shining. Feeling good. "Hi love, this is a pleasant surprise"
"Sit down Dick, I've got some bad news".
Sit down! Bit melodramatic. "What's up love?"
A moment's silence. A mere hesitation that lasts an eternity. Gripping the phone tighter now. Waiting, waiting. Now in a rush. "It's Isaac, the hospital says he has a terrible illness and there's no cure". "Surely they can do something?" "Well Adam was a bit vague. Sounded shocked. He said something about a bone marrow transplant but it wouldn't cure him".
Keep calm Dick; be business like.
"Just wait there. Louise and Adam will need us. I'm coming now". Replace phone. Sweaty palms.
No. No. Coat. No. No. Briefcase. NO. NO. Glasses. Please no. Car keys.
Doreen. Standing where I'd left her. Expectant. Serious.
"Just go Dick. I'll take care of everything".

We trooped into the doctor's room. Single file, formal. On edge but still smiling, we try to maintain some vaguely expected normality. "Nice people bearing up". It had been almost a week since we had heard the diagnosis. The shock of the totally unexpected, like plunging into arctic water, had left me breathless and numb.
Now we were to look at the options. Face the reality. Understand the details.

The paediatrician, speaking in quiet tones began to outline Isaac's condition. Slow, patient, systematic explanations. So matter of fact. Like a newsreader or a teacher helping small children to understand something they found difficult.
I wanted to bellow, "this is my grandson you are talking about", but I didn't. Everyone catches the mood. We all seem detached. Like a group of travellers in an English railway carriage.
Words and phrases. Just words and phrases. "Hurler's syndrome. Genetic disease. Mucopolysaccarides. Slow decline. Dementia.
He's got beautifully expressive eyes and such a happy smile. So innocent and trusting. I listen impassively as my grandson's life is torn apart by words and phrases. "Life expectancy. Physical deformity. Heart failure."
This can't be happening.
"Hoping for a break through soon."
What! What was that she said? Break through? Soon? How soon? What break through? What can we do? We need to do something. Anything!

The signs had all been there of course. His breathing difficulties at birth. His back beginning to bow. The deafness. Each new condition a blow that knocks my daughter to the floor. I watch Louise as, like a felled prize-fighter, she gathers her energy, clears her head and gets ready to stand and continue the fight. I am both spectator and participant in this cascade of blows.

Each of us has to deal with the punches in his or her own way. Adam silently taking the punishment but as yet no bruises showing. Kay fighting back. Always upright and alert. Only letting her guard down during long quiet nights. Wet pillow and heaving chest give testimony to love that for the moment is distant grey and black, wrapped round a sinking sun. I can't believe this is happening. My way has always been to take action. To make things happen. That's what I do best. We are in the hands of others. We have to believe in the doctors. There is hope. I already feel exhausted but through the fatigue I can see there is a poem to be written.

Like a small child I learn to wash my hands. Soap and hot water carefully rubbed into palms, back of hands and between fingers. This ritual is a necessity, a medical procedure but also an act of devotion. We need to protect Isaac at all cost. This is the bone marrow unit. This is their confinement, their prison. This is the space pod that will keep Isaac safe while Lou, Adam and Kay navigated him through a hostile world. This is the casino where we will gamble on his life. Until now reality was suspended. Soon I, we, will know the outcome. The dice that we elected to throw earlier that year will tumble to a halt and we will all know and we will move forward. We might save Isaac's life. We might see him die during the next few weeks or months or we might see him through the trauma of a bone marrow transplant only to fail. We are in orbit, waiting to see if we can bring Isaac back to earth.

Hope for the Future

Paul Hambly

Jessica and Terri

Jessica age 5 years started on the Enzyme Replacement Trial at Manchester Children's hospital in January 2003. This was a little after the other children. Jessica had to have her appendix taken out and this pushed the start date back a few weeks. She is getting along fine now and is into her tenth week she screams to high heaven at the start and finish of each infusion when she sees the needle and the IV drip but, like the rest of the children, is fine within a few minutes and soon settles down to playing and singing until she falls asleep with the effects of the pre-med.

I have been informed that the Enzyme Replacement Therapy has become available on the NHS and hopefully I will be able to get Terri this treatment as well, although it will be at the discretion of the Hull Health Authority. As you know it is a very expensive treatment and the cost will be doubled as I have two children with Hurler Scheie Disease. Nevertheless I am at present trying to get appointments with the relevant people.

Otherwise Jessica and Terri are well and happy. Jessica has finally started back at school now that the Enzyme Replacement Therapy has become a regular occurrence in our week, and like Terri is doing well in her work. Terri now has an insatiable appetite for information and asks questions about everything. She has become a firm believer in God and Jesus and loves to discuss this subject but especially with Helen who is a catholic. Terri has also become an accomplished chess player and loves to give me a run for my money. We still have all the usual arguments that come with children at any age although Terri and Jessica can even argue about who is stealing whose water when they share a bath!

Terri is 11 years old this year and is moving into secondary school this September. It is a newly built school and is fully modernised with many adaptations to cater for Terri. Terri is a little scared of the move but I know that she wili adapt quickly and learn a lot when she gets to know the teachers and pupils.

Bath time

Jessica and Terri at Christmas

Keegan Danny Lovick

Melissa Al Qadi

Hi, my name is Melissa Al Qadi and I'd like to tell you about my beautiful son, Keegan Danny Lovick. Keegan was born on 19th November 1994 at 8.06pm weighing 7 lbs 1oz. He was gorgeous with big dark eyes and lots of dark brown hair.

Keegan looked and seemed quite healthy and normal although he did get a few chest infections and he had a few episodes of staring blankly. The GP referred him to the neurologist at the John Radcliffe Hospital in Oxford where Keegan underwent many tests.

After a few weeks we were given the bad news that Keegan had Hurler disease. As the doctor explained what it meant and what lay in store for Keegan we were devastated. He would have both physical and learning disabilities and many more problems as well. On top of this we were told that Keegan wouldn't live beyond 7-10 years. What a blow!

Over the years we managed to cope somehow. Keegan made it easier for us. He was such a lovely little boy, full of smiles and laughter. Keegan went through a lot. He had grommets inserted at 2 years old but he had many ear infections. So, when the grommets fell out he had hearing aids instead which he wore really well.

We took Keegan for regular check-ups to see Dr Ed Wraith in Manchester. On one visit when Keegan was three, Ed put Keegan on permanent antibiotics to prevent chest infections, immodium for regular bowel problems and Ibuprofen for Keegan's joint stiffness.

Keegan's development started to slow down at around the age of 4 but he loved activities any other child enjoyed such as kicking a football around the garden and playing hide and seek. He really enjoyed chasing the pigeons in the market square in town.

He enjoyed swimming and horse-riding with his school too. Keegan loved school so much and it was at school that he learned to use Makaton sign language and the computer. He used to enjoy taking the class register back to the office and had a wicked sense of humour, spitting on the naughty children to the teachers and watching as they got told off!

Keegan had sleep apnoea so in June 2001 Mr Rothera removed Keegan's tonsils and adenoids in Manchester. For a few months afterwards his food would come out of his nose after swallowing it. Quite a sight!

Keegan was a little chatter-box. He could not put a sentence together but he had a variety of words and everyone who knew Keegan could understand what he was talking about.

Keegan began to get croup a lot and pneumonia so he spent a few times in hospital. He had a supply of oxygen and nebulised adrenalin at home too. By July 2002 Keegan's breathing had got much worse. I called Royal Manchester Children's Hospital for advice and they told me to bring him in. Mr Rothera monitored

Keegan with his step dad

Keegan for a couple of weeks and it seemed that Keegan was on oxygen constantly so Mr Rothera took Keegan into theatre and looked down Keegan's throat whilst Keegan was under general anaesthetic.

Mr Rothera told us that Keegan's throat was in a bit of a state, swollen in some parts, narrow in others and also floppy. He told us he could do a tracheostomy or leave him and send him home on constant oxygen. Either way, Keegan didn't have much more than a year left. In the end we decided against a tracheostomy as there seemed to be so many disadvantages against it.

However, the next morning Keegan was having big problems with his breathing. He was on 15 litres of oxygen but his sats were dropping to 75%. He was taken to the Intensive Care Unit but by then he had been given nebulised adrenalin and steroids but nothing was working. Mr Rothera told me if Keegan didn't have a tracheostomy there and then he would slip into a coma and wouldn't make it through the night.

I was so scared but I told Mr Rothera to do it. I had to try it. I couldn't lose him! When Keegan came back to ICU he was on a ventilator. It was breathing for him and he was on drips to feed him and morphine for the pain so he didn't wake up for a couple of days.

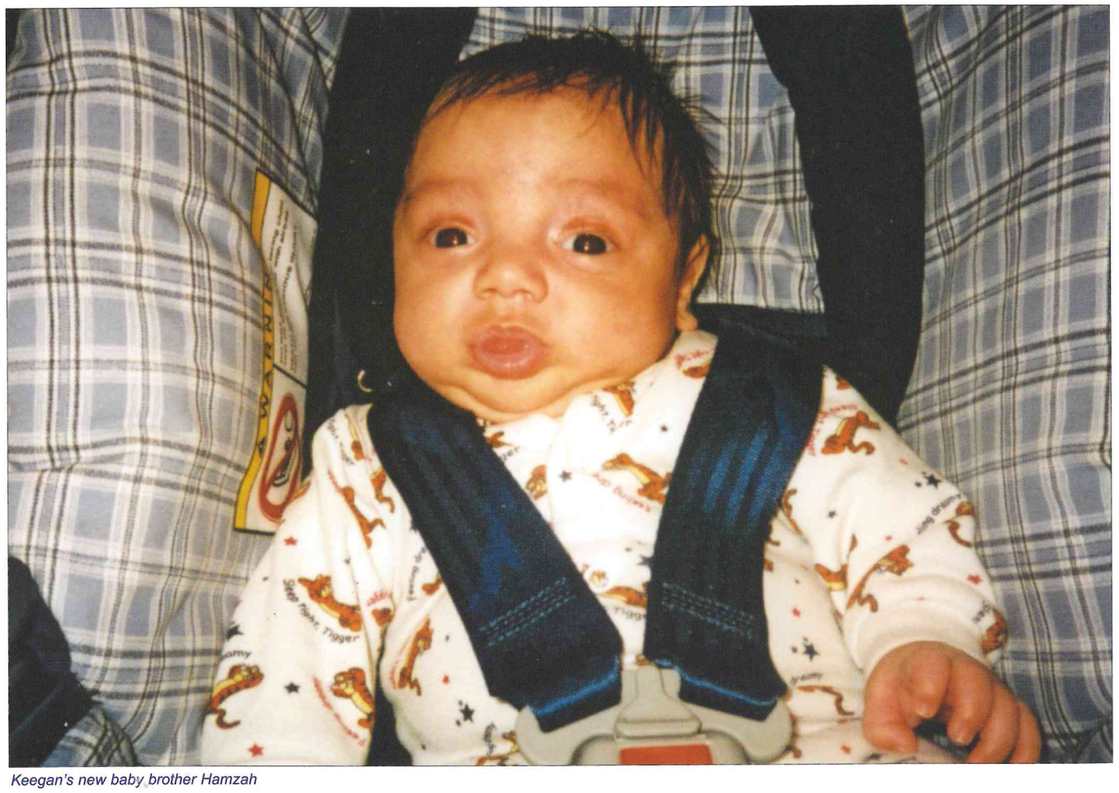
Keegan was so brave and recovered quite quickly. He was eating within a week. The next few weeks were a matter of the nurses teaching us all how to suction his tracheostomy, change the inner tube and change the tapes around his neck. It was daunting changing the whole tracheostomy.

I was so happy when I took him home from hospital. I felt like we had a second chance of being with him. Keegan adapted so well even though he could not talk anymore or walk but he never complained. He still remained that cheerful Keegan that we all knew and loved. I was so proud of him.

Keegan still loved watching his videos and loved his food. He also loved going out in the car especially to the zoo. It didn't stop us, although it was hard work lugging his wheelchair, bag and suction machine about especially whilst I was pregnant. But it was worth it to see him so happy! Keegan had his 8th birthday on 19th November. On the 20th November we moved house to be nearer the hospital, but on the 23rd I went into labour 12 weeks early and gave birth to Keegan's little brother Hamzah. He weighed 2lbs 14 oz. I was transferred to Kettering (58 miles away) because there were no special care beds in Oxford.

I took Keegan to meet him a week after and Keegan gave him a kiss. I think Hamzah was born so early because two weeks after Hamzah was born, on 8th December 2002, Keegan died. Keegan had seemed healthy enough so I was concentrating on being in hospital all the time with Hamzah. I thought I was going to lose Hamzah at that time. It never entered my head that it would be Keegan that would pass away and that Hamzah would be fine.

It just goes to show that you should treasure every second of every minute with our special gifts! We all miss Keegan so, so much and wish every day that he could still be here with us. At least I can tell his brother that Keegan kissed him.



Keegan's new baby brother Hamzah

The "Price" of Being Disabled

Marianne Stimpson

Earlier this year, my husband, Adrian and I had an idea. Our son Dominic is nine years old and has Sanfilippo Disease. We also have a two year old daughter called Jodie and it has long been our wish for them to have the "Disney experience" together. Travelling to Florida is no longer an option due to Dominic's changing condition so we decided on Disneyland Paris. This will probably be his last foreign holiday and to make it extra special we decided to invite his grandparents, aunts and uncles. Most of them were able to come making us a party of nine.

A few days later we went to the travel agent. Many of you will know that booking a holiday with a disabled child is never easy. There are many questions to be answered and forms to be filled in, G.P. and paediatrician assurance letters to be written. All irritating but understandable. Everything went quite smoothly and we were even offered a special needs room. Unfortunately, this turned out to be on the other side of the hotel to all the other rooms so our party would be split up, defeating the object of the holiday. We had already decided that one of our children would sleep in their Granny's room to avoid unnecessary disturbance and we didn't want to be too far away from them. It was a shame but we decided we could manage with a standard room if it meant us all being together. We certainly didn't expect what happened next.

We moved on to the subject of travel. We arranged to go on Eurostar from Ashford-a short journey, much easier for all concerned. When we mentioned Dominic's buggy we were first asked if it folded up. We said it did but for the safety and security of Dominic and his fellow passengers it would be better if he travelled in it. Can you imagine a nine year old with Sanfilippo dashing around a train like a human tornado? It was then the stupidity began. Apparently, Eurostar only allows wheelchair users to actually travel with them in First Class and you have to pay extra for the privilege! They say there's not enough room in the Standard carriages. The poor girl in the travel agents didn't know what to do. She was relaying all this information to us as she was getting it from the Eurostar booking office at the other end of the phone. Eventually she gave us the phone and we had a very heated debate with the man on the other end! We told him that Dominic didn't choose to be born disabled but had to pay more because he was! Then we were told that only one carer was permitted to travel with him but what if that person needed the toilet? Finally we managed to persuade him that two carers were essential. We had to agree to their terms on disabled travel but we did so "kicking and screaming". This meant that six of our party would be left behind in Standard Class, possibly with a change of train so the first class passengers would arrive approximately two hours ahead of those in Standard Class.

We are absolutely livid and disgusted with Eurostar's discriminatory rules and attitude, so much so that we've contacted "Watchdog" but as yet have heard nothing from them. The other week completely out of the blue, Eurostar contacted us to say that the rest of our party could travel First Class at a reduced rate which we've agreed to. However, this doesn't disguise the fact that this situation should never have happened. Both First Class and Standard Class coaches should have disabled facilities to give us a choice, we shouldn't be forced to travel First Class if we don't want to or can't afford it.

What do you think? Has anyone else had a similar experience with Eurostar or any other travel company we'd be interested to hear?

We're going to Disneyland Paris in August and we hope that the holiday itself goes smoother and is more enjoyable than the booking of it.

The MPS Society would be pleased to hear from any families where they have received bad service that they would like to highlight to members or a good standard of service and would like to recommend this to members please contact mps@mpssociety.co.uk

Disability Rights Commission - Potential Ryanair Class Action

The Disability Rights Commission may be starting a "class action" against Ryanair. This concerns a charge made to travellers who hire wheelchairs at airports.

If you have travelled with Ryanair since 1st October 1999 and were charged for wheelchair hire and you would like to know more, you can fill in the on-line questionnaire www.drc-gb.org or contact the DRC at ryanair.glo@drc-gb.org telephone 0161 2611840 (10am-4pm), textphone 0161 2611702.

The Disability Law Service

A free confidential legal advice and case work service for disabled people their carers and families. This service enables disabled people to access information to clarify their rights. Tel. 020 77919800 E-mail advise@dls.org.uk

'Living with Fabrys'

Margaret Cessford

My name is Margaret Cessford. I have Fabry disease and so does my son Colin. Colin is attending the Royal Manchester Children's Hospital every two weeks for his infusion of Fabrazyme. It is such a joy to see him get help. Colin is ten years old now so he is starting to suffer because of his Fabry disease. The nurses and doctors are so helpful.

I attend Hope Hospital every year seeing Dr Waldeck and Lorraine Thompson. I also have my heart checked every year at my local hospital. I have known about my Fabry disease since I was very young. I can remember Professor Desnick coming all the way from the US and examining me. Of course I didn't know much about it then.

My childhood was a very strange but happy one. My dad, Jim Ruckert, had Fabry disease but it was all very confusing for me and my mum, Elspeth Ruckert. When I think back now she must have been so confused and scared. I remember my dad being really ill and always tired. He did work but as time went on he was unable to continue working.

When he was 34 I remember my dad taking to his bed really ill. The doctor came in and prescribed tablets but they gave my dad terrible hallucinations and he thought he had seen spiders on the wall. My mum was really frightened as some of the things he saw were horrific and my dad actually ran out into the garden because he was so scared. An ambulance came and took him to a mental hospital because they thought he was going insane but I personally blame the doctor who prescribed my dad the tablets. He also suffered a stroke, but he never really got back out of the mental hospital. He was in and out and it was horrifying to see him like this. My mum did well for being so understanding.

I wish my dad was here now to see all the progress that has been made and the infusions. He died in 1987 aged 40. My mum was only 36 years old when he died and I don't know how she coped. I do suffer from sore hands now and again, sore limbs and sometimes I have muscle ache in my neck and shoulders. I have stomach cramps now and again and if I go out for a meal that can bring on the cramps.

I couldn't believe that with all the doctors and nurses working on Fabry disease that I only found out about it 3 years ago. After all, nobody really knew what was wrong with my dad. I had to tell doctors what it was. My dad must have been so frustrated knowing that the doctors didn't know how to treat him. I have read some of my dad's notes from hospital and they are horrific. They called him an "interesting case" as if he wasn't a real person at all. They treated him with drugs that actually made him worse.

I suppose its better for the children now that they have found out about ERT. I hope Colin's life is so much better than my dad's. That will be the biggest bonus for me. My daughters still haven't been tested yet. We just have to take every day as it comes and hope the future is a lot brighter.

Abby & Paige, Margaret's daughters

Top: Margaret & husband Lee Bottom: Margaret with her dad Jim (Fabry)

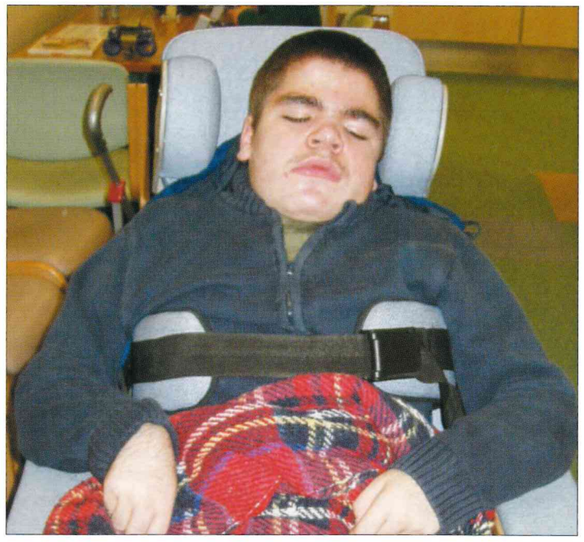
East Anglia MPS Clinic - 4th April 2003

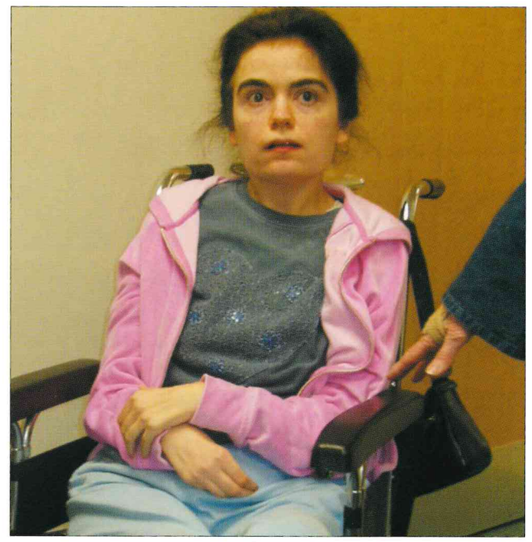
Ellie Gunary

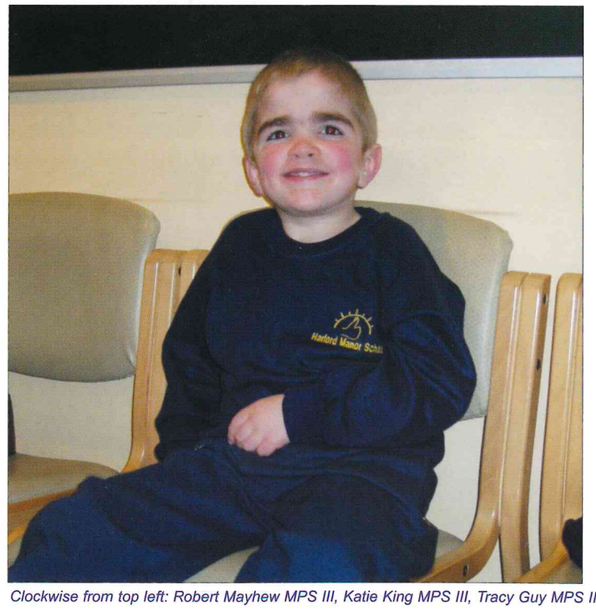
Due to the rural areas and small population in the East Anglia region there has never been the demand for this clinic that is experienced in respect of the other MPS clinics. Despite this it was extremely disappointing that only five member families attended on Friday 4th April 2003. In fact the number of patients was matched by the medical and MPS staff who had travelled from different parts of England to support this event. Needless to say it was a quiet day but from the feedback from the families attending worthwhile. It was an opportunity to access a consultation nearer home with Dr Ed Wraith who was joined by Dr Uma Ramaswami and two members of her team from Addenbrookes, Cambridge and Dr Chris Upton who is based at the Norfolk and Norwich Hospital. The nursing team made us feel very welcome and went out of their way to help.

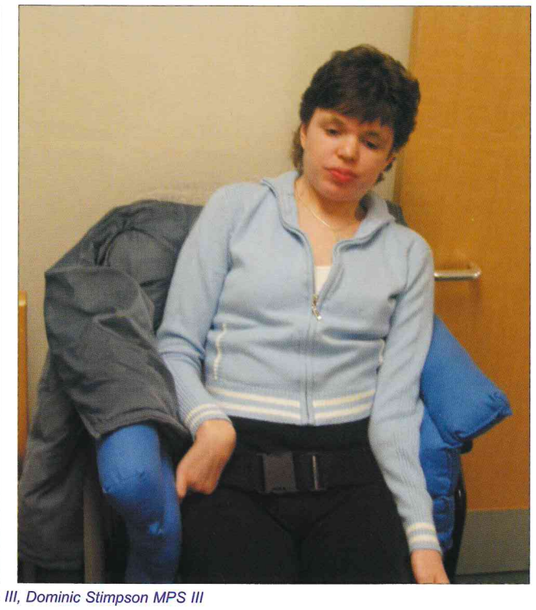
As part of the ongoing evaluation of the Society's advocacy support services all members in the East Anglia region have been consulted about the future need and location of this clinic. The Society is always mindful of the need to maximise the use of its resources and is actively considering alternative means of supporting Society members in East Anglia to access a regional consultation.

We remain indebted to Dr Chris Upton for establishing this clinic four years ago at the Norfolk and Norwich hospital and will be consulting with him and his team over the following months.









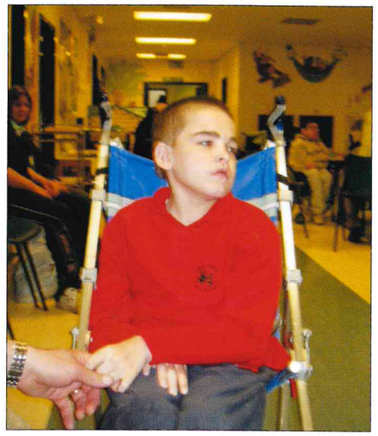
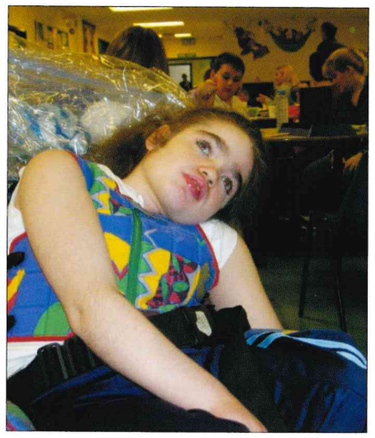
Clockwise from top left: Robert Mayhew MPS III, Katie King MPS III, Tracy Guy MPS III, Dominic Stimpson MPS III

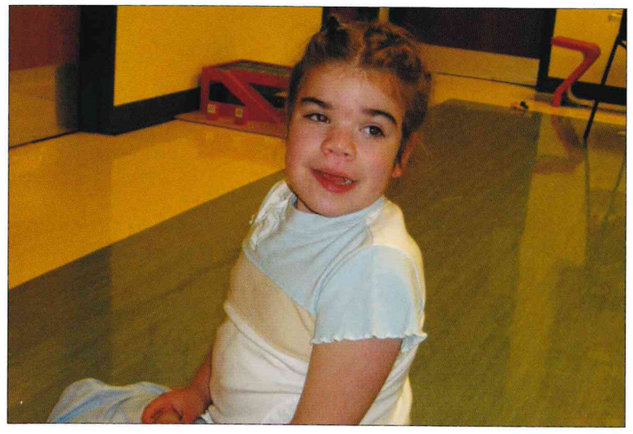
Cardiff Clinic - 30th April 2003

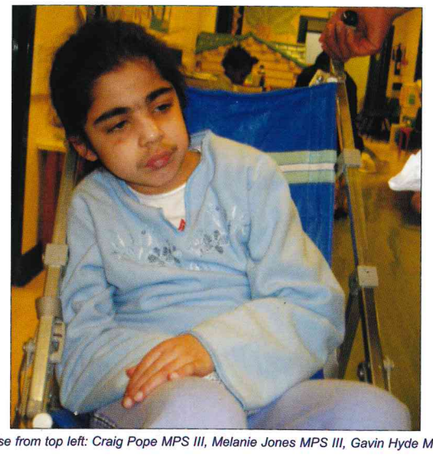
Alison West

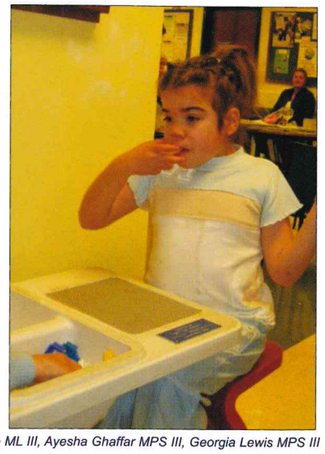
Once again we were on our way to the Cardiff clinic. How can 6 months have passed so quickly?! It was good to see so many of you again and catch up with how you all are. As usual the clinic waiting area was kept busy with a steady stream of people for their appointments and, as we were running a little late, it was hard work keeping up with everyone! However, I think we managed to speak to everyone there and were able to introduce our new Advocacy Support Team member, Jeff.

Thank you for letting us know how much you appreciate the Cardiff clinic. We value all your comments. We would like to express our thanks to Dr Wraith, Dr Shortland, Sue and all the staff at the University hospital who give their time so generously and make this clinic possible.







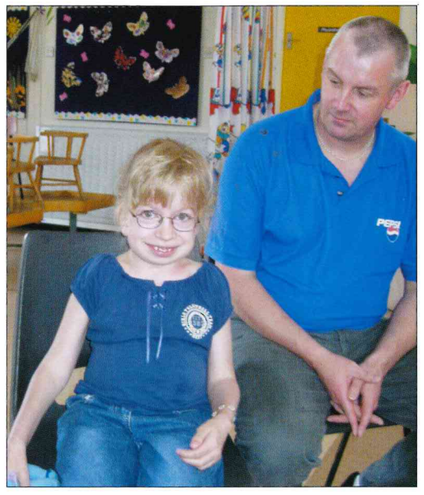
Clockwise from top left: Craig Pope MPS III, Melanie Jones MPS III, Gavin Hyde MLIII, Steven Jones MPS III, Georgia Lewis MPS III, Sarah Hyde ML III, Ayesha Ghaffar MPS III, Georgia Lewis MPS III

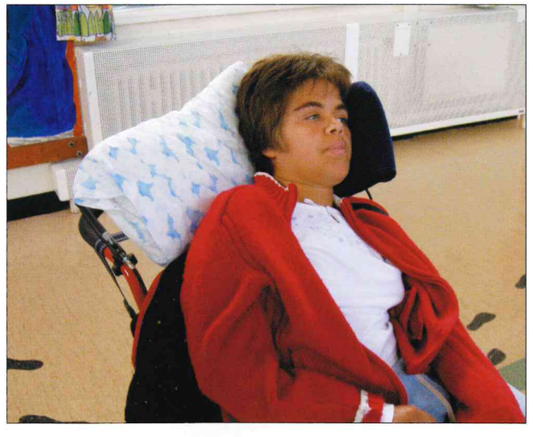
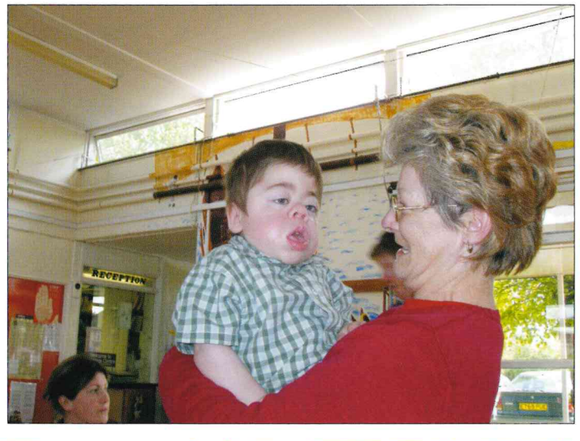
Bristol Clinic - 1st May 2003

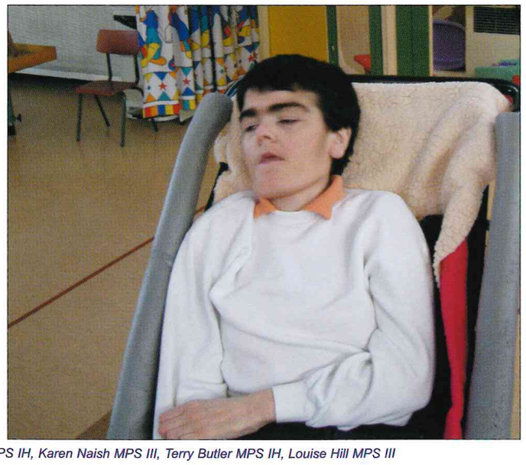
Alison West

The next day was the Bristol clinic at Frenchay, and Jeff and I were kept busy all day, introducing ourselves to new families and catching up with familiar faces. The clinic waiting area had a very sociable feel as the families met up with each other, some making new acquaintances, some re-establishing past friendships.

Once again, thank you all for your comments telling us how much you value the clinic. We really appreciate all your thoughts. Our thanks go to Dr Ed Wraith and Dr Philip Jardine who, as ever, give their time so generously, and to Deirdre and all the staff at the Children's Unit who make this clinic possible.





Clockwise from top left: Ashleigh Montgomery MPS IHS, Sophie Richards MPS IH, Ashley Tomes MPS IH, Karen Naish MPS III, Terry Butler MPS IH, Louise Hill MPS III

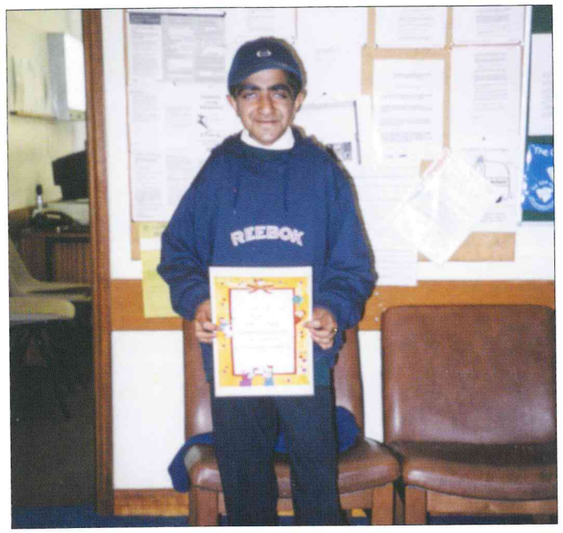
Bone Marrow Clinic - 2nd May 2003

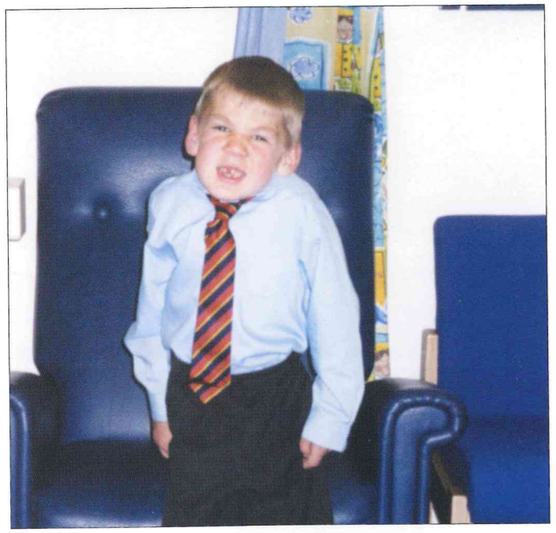
Cheryl Pitt

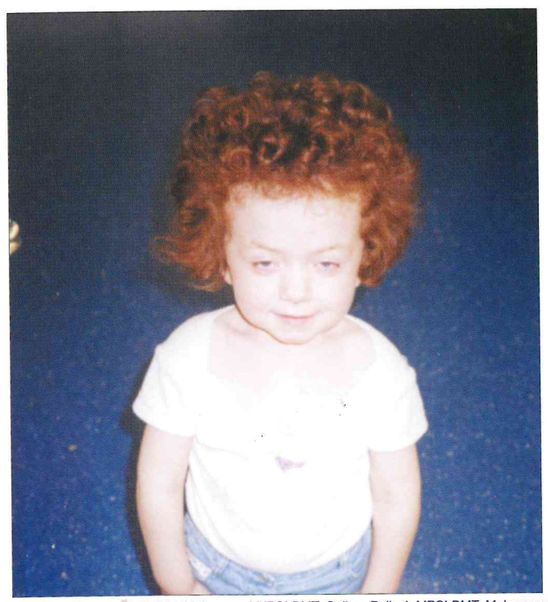
On the 2nd May, I commuted from London to Manchester by plane, which was a first for me. It was a strange experience to fly such a short distance, and it felt as though the plane had to land almost as soon as it had taken off. Since I've only ever used plane travel to visit other countries, I couldn't help but associate my journey with going abroad. This wasn't helped by the fact that I had to take my passport with me for ID purposes. So I hope you can forgive me when I tell you that I took some money out of the cash point at Heathrow, just in case I couldn't get the correct currency at my destination!

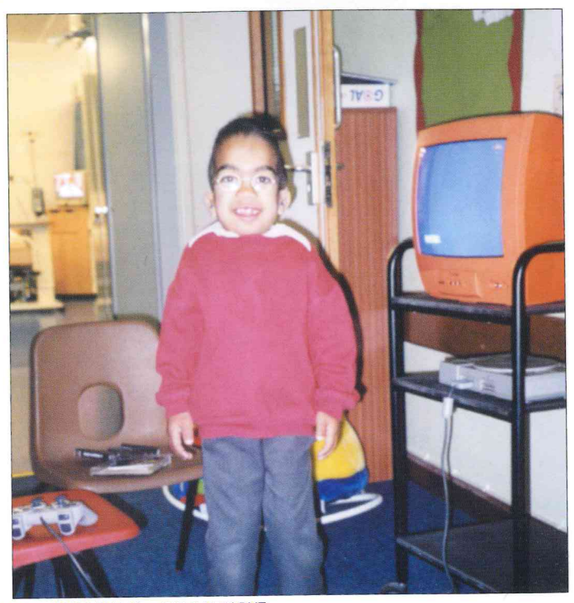
So, after a very early start I arrived at Manchester airport and made my way to the Willink to attend my first MPS clinic. At 9.30am the first patients and their families started to trickle in, and for a while I thought we were going to have a quiet day. I couldn't have been more wrong - by the end of the day I had met and chatted to thirteen families. Hopefully your experience of the clinic was a positive one, and as usual, your comments are welcome. Although I think you will agree that the new ERT suite made for an exciting new playground! It was a pleasure to meet you all, and thanks very much for your interest in the BMT research I am working on. I look forward to meeting you all again soon.

Thanks to Dr Ed Wraith and his team for making me feel very welcome and for making this clinic possible. Thanks also to Gill and Bernie for organising refreshments.









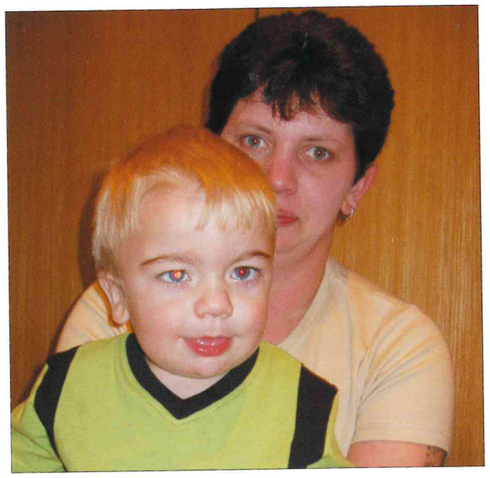
Clockwise from top left: Bilal Mohammed MPSI BMT, Callum Pollock MPSI BMT, Mohammed Razaq MPSVI BMT, Kiera O'Neil MPSI BMT

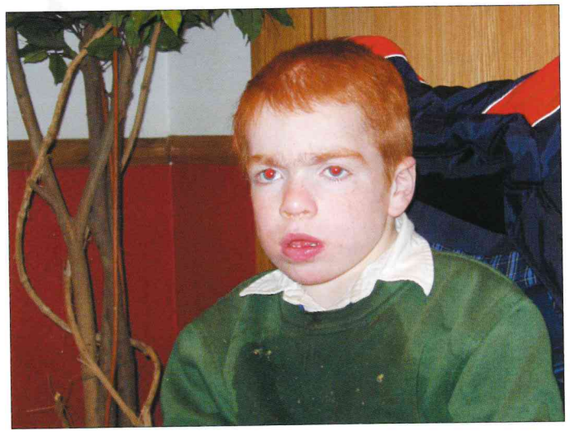
Scottish Clinic - 15th May 2003

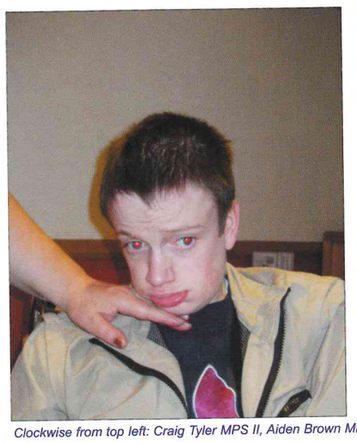
Alison West

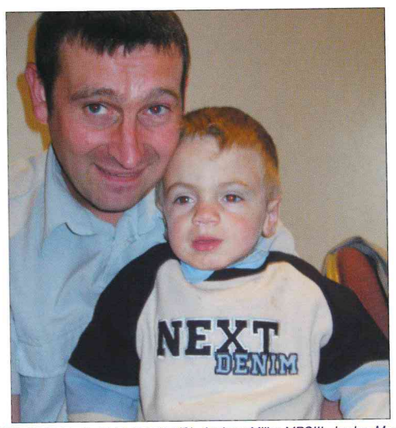
The Scottish clinic began early on the Thursday morning. Despite starting on time and families arriving early for their appointments this good fortune didn't last, and in true MPS style, by lunchtime, we were running over an hour late! However, as the families and individuals arrived and began talking to each other, the waiting area became a busy but very sociable place with all the children playing together and adults chatting as they patiently waited for their appointments.

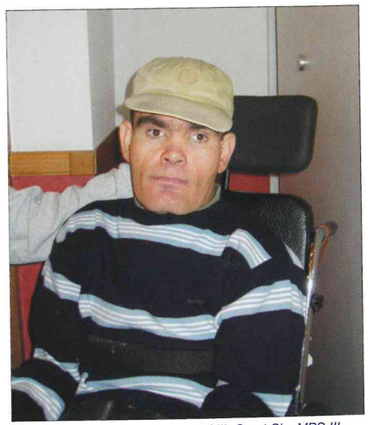
The children were all getting very excited as most were staying for our 21st Birthday Party arranged for that evening, and there was an air of anticipation as everyone discussed that evening's celebrations and the following day's conference.











Clockwise from top left: Craig Tyler MPS II, Aiden Brown MPSI BMT, Marie McCall MPS I(S), Andrew Millar MPSIII, Jordan Mount MPSI BMT, John Paul O'Neil MPSIII, Grant Sim MPS III

Northern Ireland Clinic - 21st May 2003

Jeff Bawden

The Northern Ireland MPS clinic took place on a wet and windy Wednesday in Belfast. It was a very busy clinic although not as busy as it could have been as unfortunately a number of families were unable to make their appointments. In the end twelve members and their families were seen.

This was the first clinic I attended in one of the areas I am covering, as the newest member of the advocacy support team. It was a great pleasure to meet some of the families I will be supporting and to begin to put names to faces. Ellie who attended with me was a great support, as she knew everyone so well, which made me feel very much at ease.

Many thanks to Dr Fiona Stewart and Dr Ed Wraith for making the clinic such a success. Fiona organised an official photographer to publish photographs of William revceiving Enzyme Replacement Therapy and of the team at the clinic for the hospital newsletter.

William Todd MPS I - Hurler Scheie, Dr Fiona Stewart and Dr Ed Wraith

Dr Ed Wraith, Dr Fiona Stewart, Ellie Gunary, Dr Alex McGee, Jeff Bawden and members of Belfast City Hospital Genetics Department

Looking After You Workshop

Cheryl Pitt

'Looking After You' is a workshop that is provided by the National Child Bereavement Trust, and is designed for parents or carers of individuals affected by serious illness to take some time out to look after themselves. A 'Looking After You' workshop was held at the MPS National Conference this year, and was attended by twenty parents and carers of people affected either by an MPS disease or by another lysosomal storage disorder, such as Fabry Disease. It was the perfect opportunity for people to share their experiences and support each other. It was also an opportunity for people to attend to their own feelings and emotions by taking part in therapeutic exercises. Whilst caring for a child or partner with an MPS disease has many rewarding times, it can also be physically exhausting and emotionally draining. The workshop therefore provided families with three hours in which to focus on their own emotions and needs, by relaxing, sharing, and creating. We hope those of you that attended found the time useful and we hope you managed to get something positive out of it. Although three hours of support may have felt like a drop in the ocean, perhaps the important message that can be taken from the workshop is that in order to effectively care for your loved ones, it is important to learn to be aware of your own needs as well, so that some time can be spent looking after you.

All the Fun of the Circus - Sibling Workshop

Sophie Denham

At the conference we organised a siblings workshop, which took place on Friday afternoon. All siblings aged between 5 & 11 years were invited to test their skills at the various activities, which were run by Mr Moon and his assistant Sarah.

The room was filled with an array of equipment, from brightly coloured mats and boxes. The boxes were overflowing with balls, plates (plastic I hasten to add), scarves, beanbags, batons, Diablo's, ribbons, sticks and a mystery dog! There were also unicycles, stilts and a tight rope.

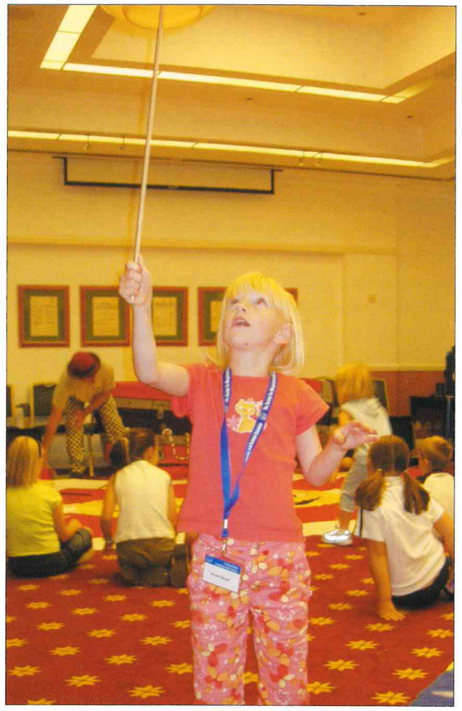
The emphasis of the workshop was to allow siblings to come together, have fun and give them the opportunity to talk with other children, whose brothers and sisters have an MPS condition.

All the children participated in the workshop; some focusing on juggling, plate spinning, stilt walking and unicycling, but the highlight of the afternoon was definitely the tightrope walking, which every child wanted to do over and over and over and over again.

After some much needed refreshments the mystery of the dog was revealed. It held balloons and a balloon pump. Mr Moon showed everyone how to make a dog out of balloons. Some gave it a try, some asked for it to be done for them, others thought it would be great fun to blow them up, have sword fights and pop them.

Some of the children found the workshop a good opportunity to speak with other siblings about their brothers and sisters, finding out which of the MPS diseases they had, how they were feeling and also to talk about their fears and hopes.

Everyone had a great time and I'm sure some new friendships were made.



MPS National Conference

Antonia Crofts

The run up to our national MPS weekend conferences is always hectic and this year was no exception. The Society has been involved in organising conferences for twenty years and over this time has gradually improved and refined both their organisation and implementation. Last year's International Symposium in Paris was rated a huge success by the professionals, families and staff who attended. Being able to top that in 2003 would be a daunting task especially as this year marks the 21st birthday year of the MPS Society.

Once again the Hilton Hotel in Northampton was our chosen venue for the annual MPS weekend conference 2003. As in previous years, continuous liaison between ourselves at the Society and staff at the Hilton was of paramount importance to ensure that the whole weekend from beginning to end would run as smoothly as possible. In reality, this is a logistical nightmare. Spread over three days from Friday lunchtime to Sunday lunchtime approximately four hundred families, professional delegates including doctors and pharmaceutical company representatives and volunteers had to be fed, watered, entertained and accommodated. An array of conferences, workshops and children's outings were co-ordinated to run concurrently with military precision.

After months of hard work, planning and preparation, anxiety and anticipation, the beginning of the conference seemed, in a way, to come as a welcome relief. The planning stages had passed, everything was ready and in place. Anything that wasn't ready was sorted and if this wasn't possible, it was probably too late to change it anyway!

MPS Society staff arrived by coach at the Hilton on Friday ready for action, ready for anything. Many of us had been through conferences before and despite the odd problem or two had emerged relatively unscathed but each conference is different and each is a new experience.

Many MPS families arrived on the Friday afternoon to take full advantage of the long weekend and also to meet and mingle with other families and find their way around the hotel and to their rooms. Registering the families and booking them into their rooms is always a busy time but it seemed to go smoothly nonetheless. Some parents had booked themselves a little rest and relaxation in the Looking After You Workshop for the Friday afternoon. As for the kids – the Siblings workshop, facilitated by Alison and Sophie and focussing on circus skills, proved a great success. Even Alison and Sophie were boasting about their newest talent - plate spinning!

Saturday was the big day for us. Three conferences ran concurrently, each tackling a different aspect of MPS. This was a chance for families and professionals to choose the conference most suited to their needs and experiences and it was a means of covering as many issues as possible in a short period of time. Unable to be in three places at the same time I chose to sit in on Conference A. In particular, I never fail to be moved by the family speakers who, with their photos and personal accounts of living with MPS, seldom leave many dry eyes in the house.

At about 5pm the children and volunteers returned from the children's outing. Drayton Manor Park is always a favourite place to visit and the kids come back with many stories to tell from their day out. Volunteers, on the other hand, usually return from the day feeling very fulfilled but completely worn out from being constantly "on the go"!

To celebrate the launch of the Enzyme Replacement Therapy for MPS I, "Aldurazyme", a reception was held in the hotel entrance. A circus act kept the children occupied whilst the adults toasted the launch and listened to speeches from Christine, Ellie, Barry Wilson the Chairman of the Society's Trustees and Julie Kelly from Genzyme. Two beautiful cakes were cut to celebrate both the launch of Aldurazyme and the Society's 21st Birthday. As a special treat "Rhapsody in Blue" roses were given to those professionals, volunteers and families who had contributed in an outstanding way to either the work of the Society or more generally to the field of MPS diseases. Ellie also received a rose, although she didn't know about it until her name was called out!

As per tradition, Saturday evening is gala dinner night with the opportunity to sit down to a three course evening meal whilst watching a slide show of the children's outing earlier that day. For many parents this is a real treat to see what their children have been up to and how much they have enjoyed their day. Dr Emil Kakkis gave the after dinner speech which was followed by an astounding sale on the raffle tickets. MPS staff didn't need to try out their relentless sales technique as so many people voluntarily emptied their pockets in the name of MPS!

Sunday morning saw an early start once again as the children's outing left early for Gulliver's Land. As the coaches get ready to depart this can be a logistical nightmare ensuring that each child is successfully paired up with the correct volunteer and also has the necessary nappies, warm clothing, etc. Once the parents had waved goodbye and the coaches departed the Sunday morning conference can begin.

The theme of this particular part of the conference was "Therapeutic approaches to MPS and Related Diseases" and featured eminent speakers from the medical world talking on Enzyme Replacement Therapy. Once again, it was one of the family speakers, James Garthwaite, who gave a very personal experience of waiting for treatment. His moving and sometimes humorous talk provided a direct contrast to the more scientific, clinical content of the other Sunday morning conference talks.

The conference weekend ended at lunchtime on Sunday. Certainly from our perspective at the MPS Society, this conference has been the most successful ever. We have received some really positive feedback from many families and professionals. The doctors and pharmaceutical company representatives in particular really do seem to welcome the opportunity this conference affords to mix with and learn from the families who live with MPS on an everyday basis. The chance to meet new families, re-establish old contacts and share experiences remains an invaluable part of the weekend conference. We hope that all those who attended the weekend found it as inspiring, informative and fun as we did!

Clockwise from top left: Sinead McAlister MPS IV with parents, conference Hotel Foyer, Tara Murphy and Adam Hackett MPSIH BMT with their mums, Ben Davies MPS IV with his brother & dad

21st Birthday Reception

Christine Lavery - Director

Over the last 20 years, literally thousands of people have played a role in steering the Society on its extraordinary journey to being one of the UK's leading voluntary patient support organisations for specific diseases. Most importantly the Society has through good governance and working in partnership been able to achieve a high level of public awareness, a truly unique advocacy support service and has funded nearly £2.5 million of scientific and clinical research.

As part of our 21st birthday celebrations the Society took the opportunity to acknowledge and thank some people who have contributed in the most exceptional way to helping those suffering from MPS, their families and the Society.

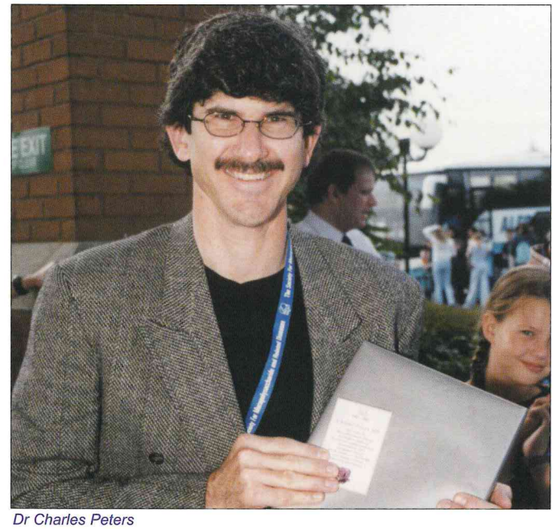
Liz Young

When an MPS or a related disease is suspected, samples of urine and blood are taken to look for the missing enzymes. Three people in particular have led the field in the biochemical diagnosis of MPS. Elizabeth Young who has delivered this service at the Institute of Child Health, London for over 30 years until her retirement earlier this year and from Manchester Alan Cooper and Margaret Thornley. Between them they have many, many years of providing the service and a majority of the Society's members have personal experience of their skills in the area of pre-natal diagnosis.

There are no miracle answers to the management of MPS and related diseases. From a clinical perspective it can be no easy task giving the news to individuals or their families that they or their child, even children, have a degenerative disease and that there is no cure and for many no treatment. Dr Ed Wraith, consultant paediatrician at the Royal Manchester Children's Hospital, Dr's Maureen Cleary and Ashok Vellodi from the Hospital for Sick Children, Great Ormond Street and Dr Fiona Stewart from the City Hospital, Belfast, between them face doing this more than 50 times a year as well as provide ongoing clinical care to a majority of MPS families in Great Britain and Northern Ireland.

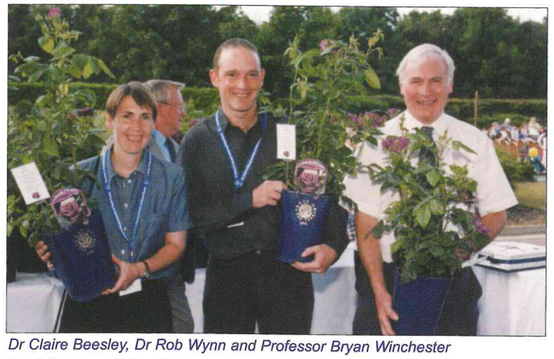
Dr Maureen Cleary, Dr Fiona Stewart & Dr Ed Wraith

In 1980 UK doctors Professor Jack Hobbs and Dr Kenneth Hugh Jones pioneered bone marrow transplantation for MPS. Over the years BMT for MPS I Hurler has become a routine treatment option and work continues to improve survival and clinical outcome for these patients and we take took this opportunity to recognise the work of Dr Charles Peters at the University of Minnesota, Minneapolis.



Dr Charles Peters

In fulfilling the Society's strapline 'Care Today, Hope Tomorrow', hope for tomorrow has come through three very dedicated researchers who have been and continue to be at the cutting edge of research into Mucopolysaccharide and other lysosomal storage diseases. Professor Bryan Winchester and Dr Claire Beesly from the Institute of Child Health, London and Dr Rob Wynn at the Royal Manchester Children's Hospital who with the help of 'Jeans for Genes' funding carry the hopes and aspirations of many members on their shoulders.



Dr Claire Beesley, Dr Rob Wynn and Professor Bryan Winchester

In 21 years the Society has never lost hope that much needed treatment for these diseases will become a reality. In August 2001 Enzyme Replacement Therapy for Fabry disease was licensed in Europe and through specialist treatment centres in London, Cambridge and Manchester patients receive ERT. Dr Atul Mehta from the Royal Free, London, Dr Steve Waldeck from Hope Hospital, Manchester, and Dr Uma Ramaswani from Addenbrookes Hospital, Cambridge, as well as Dr Wraith, have worked closely with the Society in enabling patients to access their treatment.

Dr Uma Ramaswami & Dr Steve Waldeck

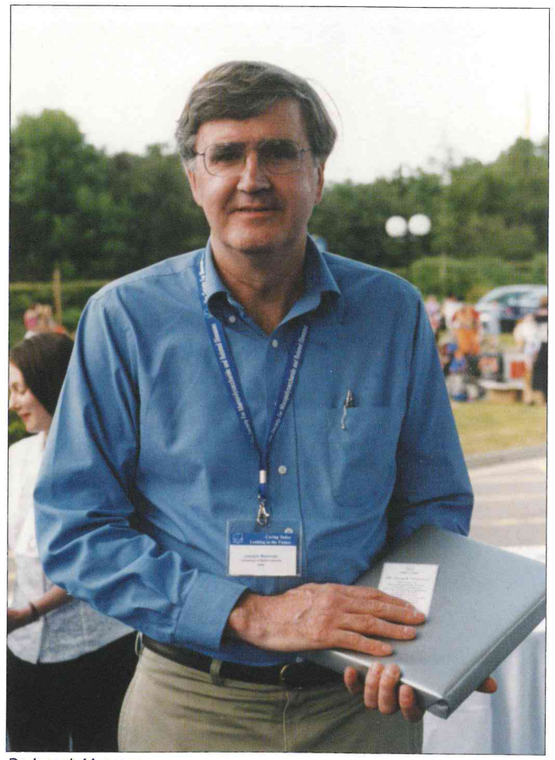
In a week when we have seen Enzyme Replacement Therapy licensed in Europe for MPS I it couldn't have been more appropriate to recognise the sheer determination in the face of adversity of Dr Emil Kakkis, formerly of Harbor UCLA Medical School and now Biomarin Pharmaceuticals, in developing this treatment. If it wasn't for Emil's unique commitment I believe that ERT for MPS I and some of the other diseases would still be only a hope on the horizon.

Dr Emil Kakkis

In order to get to the position we are in today, 45 MPS I children and adults in Europe and North America participated in a worldwide clinical trial. When the trial started in Manchester, Jean Mercer was the only ERT nurse and MPS staff regularly visited Manchester taking their turns in entertaining the trial patients.

Nurses play a vital role in the care of the MPS patients. One particular nurse, Lorraine Burnett, who left the Royal Manchester Children's Hospital a couple of years ago, gave considerable commitment to MPS patients over many years as well as becoming a childcare volunteer for the Society and very recently undertaking some individual advocacy support as a consultant to the Society.

Enzyme Replacement Therapy for MPSII, Hunter disease and MPSVI, Maroteaux Lamy disease are now being trialled clinically in the United States and we took this opportunity to thank Dr Joseph Muenzer from Chapel Hill, North Carolina for his services to patients with Hunter Disease and Dr Paul Harmatz from the Children's Hospital, Oakland, California for his services to the patients with Maroteaux Lamy disease.

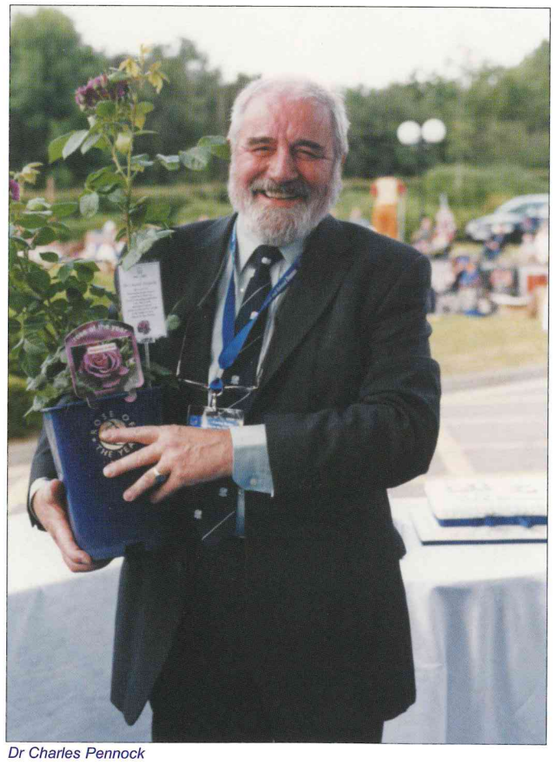


Dr Joseph Muenzer

Bringing an Enzyme Replacement Therapy to the market place and making it accessible to patients requires both major financial investment and the ability to produce large quantities of enzyme for distribution worldwide. Biomarin Pharmaceuticals, through Emil Kakkis, Genzyme Corporation, TKT inc and TKT 5S Europe took up the challenge. In recognition of the work of Genzyme Corporation, TKT Inc and TKT 5S Europe we showed our appreciation through the UK representatives Julie Kelly for Genzyme and Sheila Bone for TKT 5S.

Sheila Bone & Julie Kelly

We also recognised the support of Dr Charles Pennock, formerly from Bristol Children's and Maternity Hospital, who has chaired the MPS conference since the mid eighties.



Dr Charles Pennock

During the Conference weekend those present could not have failed to have witnessed the sixty strong team of volunteers who are providing invaluable childcare to over 100 MPS children and their brothers and sisters. Most childcare volunteers come back again and again and whether they were here for the first time or becoming a regular, on behalf of all the Society's members we conveyed our deepest appreciation. However over the last 20 years there are a few volunteers who have come back year after year for more than 10 years. At the reception we recognised this commitment to the Society by Jenny Bates, Brin and Chris Edwards, Helen Patterson, Val Evans and Robin Lavery. We also acknowledge the grandparents of Simon Lavery, Ken and Pam Ballard, who have shown exceptional commitment as childcare volunteers for sixteen years, fundraisers for all the used stamps and foreign currency donated to the Society and the volunteer packers of virtually every MPS newsletter posted to members since it was published in 1982.

The Society under charity law is governed by a Board of Trustees, a voluntary undertaking that carries heavy legal and financial responsibilities. We are grateful to all Trustees who have served the Society in the last 20 years. However there are two Trustees who have given exceptional service. Wilma Robins has served the Society as a Trustee without a break since January 1987 missing only a handful of meetings in 16 years. Lynne Grandidge served the Society as Trustee and Treasurer between 1993 and 1999, a period when the number of staff doubled and the Society's income increased threefold. When Lynne retired it was necessary for her onerous duties to be taken over by a paid finance officer.

We also acknowledged all the members, their families and friends who have supported the Society over 21 years. The efforts of everybody, however big or small, is greatly appreciated, however in these birthday celebrations there were five members we chose to single out for special recognition. There are some things totally irreplaceable and that is the life experiences of our members. The Society's publications are sort after by families and professionals worldwide and we are grateful to the many, many families who have shared their children's lives with us for the purpose of these publications.

To Dawn and Graham Cawthorne, parents of Lauren, and Peter and Sue Stuart, parents of Jessica who died last July we are particularly indebted, as we are to Claire Garthwaite, mother of Tom and Louis who drafted and worked with the Society in bringing to print the 'I've Got Morquio's' and 'I've Got Hunter's booklets.

Finally looking to the future I offered a personal thank you to the whole MPS staff team and Society's Board of Trustees who serve the Society's members unfalteringly, not least Barry Wilson who as Chairman gives more commitment and time than most to the Society. Life can be tough at the top but it can be equally tough being second in command. Ellie Gunary came to the Society a little over four years ago and in that time has steered the Society's advocacy service to conform to the highest standards we see today. A service that is equitable and accessible to its members and adheres to best practice.To both of them on behalf of the Society I say a huge thank you.

Robin Lavery, Brin Edwards, Jenny Bates, Pam & Ken Ballard

Peter & Sue Stuart

Val Evans

Julie Kelly, Emil Kakkis & Samantha Walker (MPSI)

Preparations for the Childcare Programme

Ellie Gunary

Thomas Birch MPS II with his mother Sharon and Winnie the Pooh which Thomas won at the conference

Preparation for the childcare programme for the MPS National Conference 2003 began many months ago. Outing venues were researched, coaches booked and volunteers recruited. For each childcare volunteer, a completed application form has to be received, two references sought and clearance from the criminal records bureau achieved.

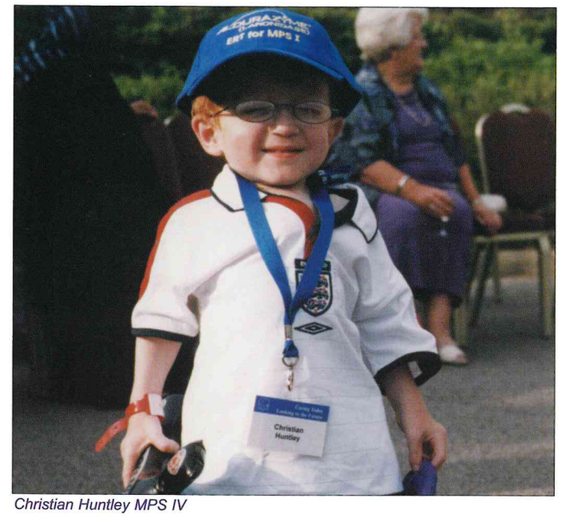
Information is then sought in the form of a completed questionnaire from the families attending with children about the care each child partaking in the children's activity programme requires. This is translated into an individual management plan and an appropriately skilled and experienced volunteer allocated.

The week leading up to the childcare volunteers training which took place on Saturday 7th June 2003 in Chalfont St Peter, Buckinghamshire was spent finalising the allocation of volunteers to children and compiling the volunteers outing packs. These packs comprise of the individual management plan or registration form for each child the volunteer is caring for during the weekend, the relevant specific disease booklets, the volunteer carers conduct policy, the Society's child protection policy, protocols to follow when changing nappies and in the event of a medical emergency, gloves and bed pads. The MPS office looks like a medical supplies outlet for a short time as these items fill desk space.

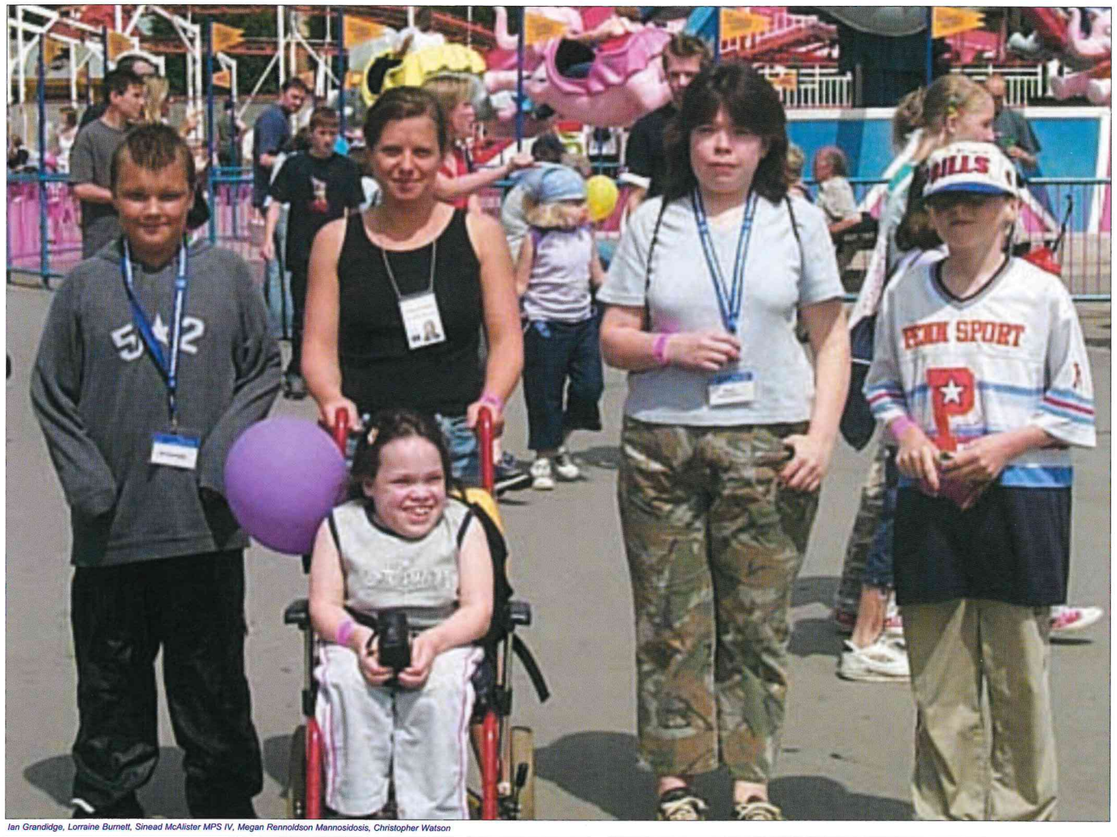
Saturday 7th June and 57 volunteers travel from throughout England to a church hall in Chalfont St Peter for a training day. A qualified moving and handling trainer teaches safe moving and handling techniques and those volunteers who do not know each other soon get to know each other as they hold hands and stand close together practising how to support a person to stand up from a chair, walk safely, roll onto their side and move up a bed. The importance of respect and encouraging independence is promoted, as is management of hyperactive behaviour. A full briefing follows on the whole childcare programme including the loading of drinks onto the coaches, the role of coach leaders and ensuring the labelling of buggies. Imagine the chaos if buggies weren't labelled when unloading the coaches at Drayton Manor Park and Gulliver's Land! Finally the volunteers are trained in following the policies and procedures, which generates considerable discussion and debate as clarification of these is sought.

Saturday over the fortnight running up to the conference always brings unexpected changes, a child is ill and the family can no longer attend, a volunteer has a family emergency and many more, changes which can only be expected when managing a childcare programme on this scale. 106 children were supported on the Conference childcare programme, a record for the Society. Not one spare place was to be had.

Friday 20th June 2003, the start of the conference and the work begins by ensuring each volunteer meets with the families they are supporting. On Saturday and Sunday morning members of the Society's Advocacy Support Team act as runners for the coach leaders, running round the Hilton hotel rounding up the children not yet on the coaches before they depart. Despite the odd expression of surprise that this was not in the job description and running is not a strong point members of the advocacy support team broach this enthusiastically and in true team spirit rise to the occasion. The coaches depart and on time!!!!! The final task of the conference after the volunteers have returned from the final outing is to collect in the individual management plans from the volunteers and gain feedback as to how the childcare programme went. The Society is indebted to it's dedicated team of childcare volunteers to whom, on behalf of the families and staff team we say a big thank you.



Christian Huntley MPS IV



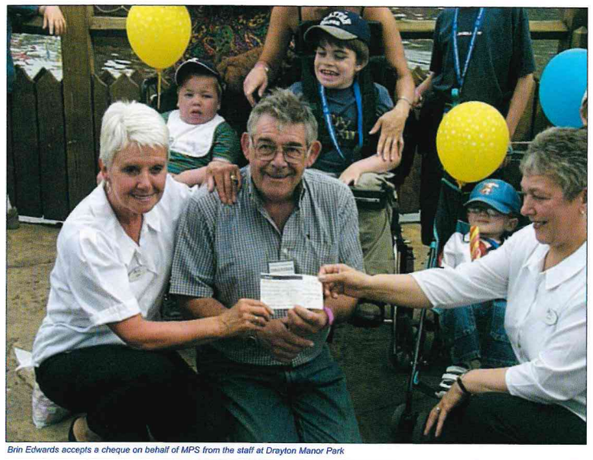
Ian Grandidge, Lorraine Burnett, Sinead McAlister MPS IV, Megan Rennoldson Mannosidosis, Christopher Watson

Sinead McAlister MPS IV

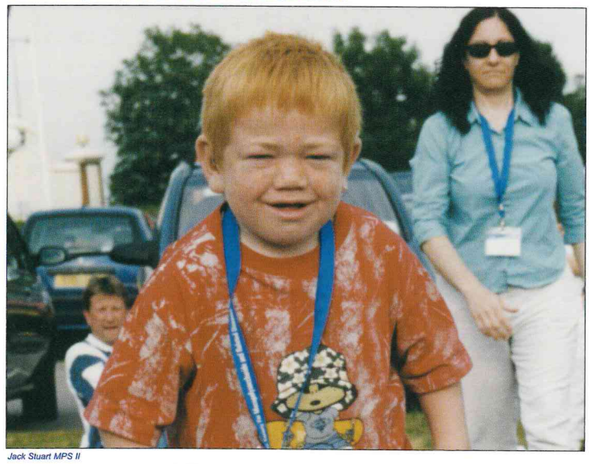
Annie Stuart

Tara MPS I BMT & Kate Murphy with volunteer Lucy

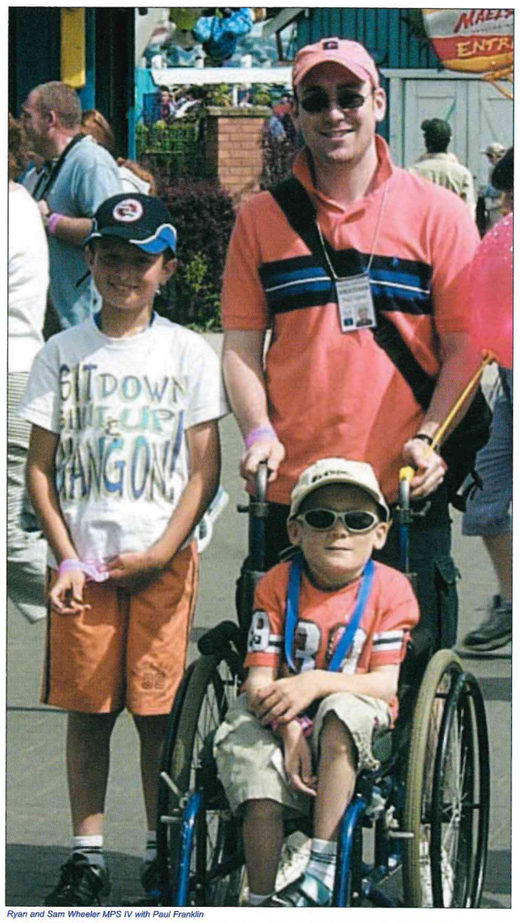
Jacqueline Talbot with Harrison King MPS III, Andrew Talbot with Harvey King, Ciara and Adam Hackett MPS I BMT



Brin Edwards accepts a cheque on behalf of MPS from the staff at Drayton Manor Park



Jack Stuart MPS II



Ryan and Sam Wheeler MPS IV with Paul Franklin

Daniel Muers with friend

Celebrating 21 Years in Scotland - MPS Conference - 16th May 2003

Alison West

In line with our other celebrations planned for this, our 21st birthday year, The MPS Society had planned a series of events to start off the celebrations in Scotland. Over 2 days we held a clinic followed by a birthday party, and a conference for all families, individuals and professionals who would like more information on the various MPS disorders.
We arranged the Hilton Hotel in Edinburgh as the setting for these events, and we would like to thank all the staff who did everything they could to accommodate us and help the events run as smoothly as they did.

21st Birthday Party
Everyone assembled outside the room nice and early for the party on the Thursday evening. The children were getting more and more excited and couldn't wait to get into the room to play - so they didn't! The hotel staff were excellent as they manoeuvred their way skillfully through a horde of hyperactive children while carrying trays of glasses and other items for the party.

Before long we were allowed into the room which looked lovely. The food was excellent and everyone there enjoyed a good feast. The 21st birthday cake was due to be cut after the meal, but unfortunately Barry Wilson, who had been invited to make a speech at the cutting of the cake, still hadn't arrived. He was stuck on a delayed flight from Birmingham!

This was not a problem however, as the Children's Entertainer was ready to begin his act. All the children (and even some of the adults!) thoroughly enjoyed his mix of magic, party games, and a puppet show which passed the time beautifully until Barry arrived.

The birthday cake was presented and Barry spoke about the last 21 years of the MPS Society and how far it had come in that time. He then invited the children to blow out the candles and everyone toasted the 21st birthday of the Society with a glass of wine and a piece of birthday cake.

Scottish Conference
After a hectic start organising the children on their outing to Edinburgh Zoo, we could turn our attention to greeting the delegates who were arriving for the day's conference. As people arrived I was aware that I hadn't met a lot of these families or individuals before, although I had spoken to many of them on the phone, and it was great to be able to finally put a face to a name.

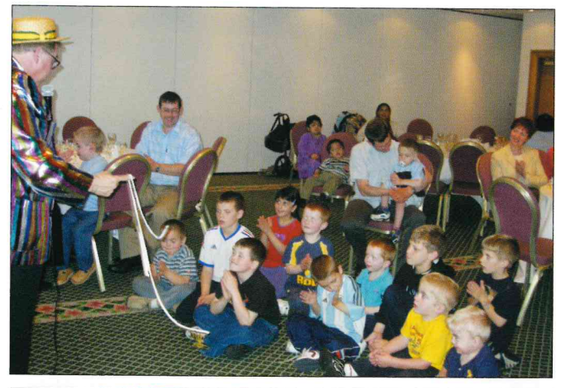
Dr Peter Robinson introduced all the speakers, who covered a wide range of topics from the new treatment trials for MPS I to the work of the hospices in Scotland. The Society would like to thank all the speakers for provoking some stimulating discussions. Dr Robinson, our chairman, did an excellent job, monitoring proceedings carefully, keeping speakers on time and initiating some fascinating discussions. The families and individuals who attended all said how much they had learned and benefited from meeting other people in the same or similar situations.

As the conference came to an end and the families collected the children as they returned from the outing, everyone began to slowly make their way back home. Some had further to travel than others, but all seemed to agree that it had been a very worthwhile two days and everyone would take something away with them.

We would like to extend our thanks to Dr Cleary and Dr Robinson for giving their time so generously over these two days and providing the support, advice and information these families and individuals need.

The MPS Society would also like to take this opportunity to acknowledge the tremendous contribution from the Community Fund whose grant made this conference possible.

Lastly, we would like to thank all the families, individuals and professionals who attended these events. Without you this would not have been possible, so many thanks to all of you for coming and making the MPS Society's first official 21st Birthday Party celebration so successful.



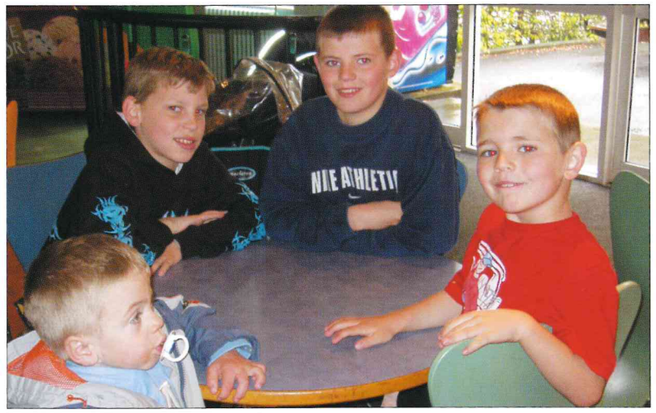
Edinburgh Zoo

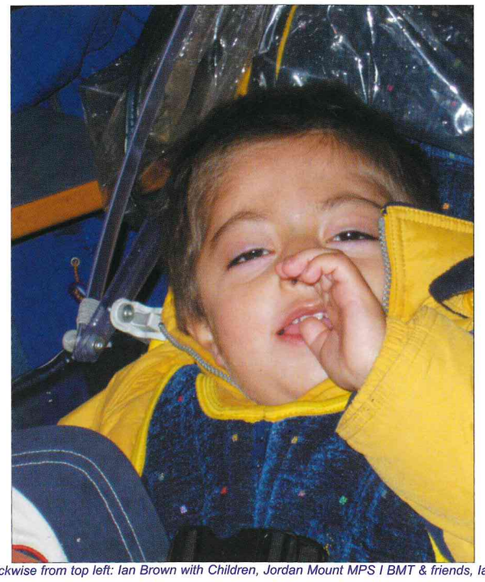
Jeff Bawden

On the day of the Scottish MPS conference which was wet and windy, the children went to Edinburgh Zoo and much fun was had by all. The train to the top of the hill was a very big hit and everyone went on it at least three times. But no one could get off at the top because they would have been carried away by the gale force winds up there. Everyone also loved the monkey house, I think they saw a lot of themselves in the monkeys.

Unfortunately we had to return early because the cold weather just got too much for some, but the hotel were very good at coping with an unexpected horde of children.

We would like to thank all the volunteers who gave their time to support the children around the slopes of Edinburgh Zoo in far from ideal weather conditions.





Clockwise from top left: Ian Brown with Children, Jordan Mount MPS I BMT & friends, Ian Brown with Connor Tyler MPS II, volunteers & children, Aman Mirza - MPS II, Jeff with children at the zoo

Robin House - Scotland's Second Children's Hospice

Gilbert Watterson

Robyn Watterson MPS IH

Hi everyone in MPS land, I have been promising to write a small article for our newsletter for over 5 years now so I`m glad that Ellie & Co haven`t held their breath!
This is an abbreviated version of the story so far.

Scotland at the moment only has one children's hospice, Rachel House. We initially started using Rachel House shortly after Robyn`s diagnosis of Hurler disease, back in April 1998, the referral coming from the MPS society. At first we were extremely nervous and probably like most families feeling rather vulnerable as we were still trying to cope with the "mini bereavment" known as diagnosis. Anyway as anyone who has been using the children's hospices throughout the UK will know these services are totally essential in helping families maintain a good quality of life and giving parents a well earned rest.

Robyn was building up her trust with the staff at the hospice and the respite given was great but after the first year we were starting to notice that the amount of new referrals coming through the doors was very much on the up. On one hand the fact that more families were getting help was brilliant, on the other hand it didn`t take long to realise that the hospice was quickly running out of availability and although they would never turn anyone away from the door, we all knew that something had to be done to ensure that the high standard of care and respite could continue.

A few families were asked to come along to a meeting at the Childrens Hospice Association Scotland and to see if we would like to help in a new campaign to raise £10 million to build a second hospice as it was so badly needed. Sylvia, Robyn and myself along with some other parents decided to help.

On the 13th of September, just two days before Robyn`s third birthday and a Sunday night drive back from the annual MPS conference in Northampton, we received a phone call saying that the family who were going to spearhead the campaign had pulled out due to their daughters` ill health and could we fill in for them. Things started happening really fast after that and we became the Media family and Robyn was now the face of CHAS`s new hospice appeal.

The first article came out a week later and was front page news on Scotland`s top selling Daily Record. Sylvia and I were stunned and quickly realised that everyone in Scotland and especially our neighbourhood would now know that wee Robyn was not only disabled but "terminal". This was quite upsetting to say the least but it had to be done in order to educate people and obviously to raise the required money to build our new hospice. I have to say that knowing what is up with your daughter is bad enough but reading it in print is even harder.

Anyway it was full steam ahead and lots of press coverage and tv etc. The money started pouring in and the whole of Scotland started doing loads of fundraising. We knew this was not going to be an overnight success but with the bit between our teeth and "dug" in for the long haul we carried on. To be honest it has been the most worthwhile venture that I

Robyn with mum Sylvia and dad Gilbert

Robyn & Grandma

have ever been involved with and after three and a half years and with Robyn now approaching her 7th birthday we decided to give our wee star a bit of a rest and to make sure that Robyn wasn`t overexposed plus the little honey is slowing down a wee bit and doesn`t always feel like posing for the camera.

I remember the excitement when we reached our first milestone, the first million pounds. It was on the 11th of March 2000. The fundraising was going really well and we had the pleasure of meeting several of Scotlands top celebs, it is no surprise to me that so many successful Scots were up for helping us, after all we are one of the warmest nations in the world! A wee touch of Braveheart there?

We must now have at least £6 million in the bank and looking back a few things do come to mind, here is just a couple:
Live television the best laxative that I have ever tried. I remember going on a show called "summer at the top", a sort of poor man`s Richard and Judy. I was caked in makeup and was sat on a sofa with Andrea the head of care at Rachel house. My heart was racing and I was trying to sound as polite as possible but it all came out a bit "Graham Norton". One of my friends even asked me who the gay guy was that did my voiceover! Since then I just talk total Glaswegian, and it is miles better.

We took Robyn along to the Grand Lodge of Scotland to meet Grand Master Mason Archibald Orr Ewing and picked up a cheque for a cool quarter of a million pounds, raised by Freemasons throughout the UK. Also meeting business tycoon Vera Wisefeld who had invited us to her house as she saw wee Robyn on the telly and was keen to help. This woman is truly remarkable and had a cheque for £100,000 towards our new hospice appeal. Other celebs have included Sharleen and the rest of her band Texas. They gave another £100,000 from their millennium gig in Edinburgh. We also meet Carol Smillie, just about every one from itv`s Taggart, Fred Macaulay and a host of others but my favourite has to be Euan Macgregor who has done so much to raise awareness and is never far from the kids at Rachel House when he comes home. I`m sure that everyone who uses the hospice would like to thank all who have helped in any way, shape and form in the last ten years with the Scottish Hospice Association.

The two most important things that have come out of this appeal have been obviously the money to build our new hospice and to try and encourage families who are out there who aren`t aware of the help available to them to come forward. The most amazing thing which has been the biggest bonus is that a wee girl with such a devastating condition as Hurler could be so well known in Scotland. Robyn and children like her have been hiding in the shadows way too long and the people who may have been `nasty` due to ignorance are now well in the process of being "educated" that people who are severely disabled and may look different to "the norm" can still have a great quality of life and can also serve a most worthwhile cause in the community!

As I said at the start we now have enough money to build Robin House, named after the wee tenacious bird, and although we haven`t got to our target, I`m sure the fundraisers will get there in the end to ensure the hospice`s day to day running costs will be sorted. The building is due to start soon and hopefully the doors will be open for respite in the summer of 2004. I would also like to take the chance to thank all the staff at Rachel House for the excellent work they do and also to say hello to everyone at Derrian House and all childrens hospices in the UK. Anyone who is reading this and has a child with a terminal illness and who is currently not using these services should certainly summon up the courage to have a closer look as the help offered is second to none. Please think about it as it could really help you to get through some of your most trying times. On a finishing note I would also like to say that the help through bereavment given to parents and siblings is invaluable and this has been passed on to me from several families whose children it has been an honour for me to meet. I hope this has been worth a read to you all and to all MPS related families please try to grasp the little positives out there. They may be few and far between but priceless and it never really helps to dwell on the negative.


Robyn and dad Gilbert

Jennifer's Story

Jennifer Johnston

The Enzyme Replacement trial started in January 2001. This trial was for Hurler Scheie/Scheie sufferers and was going to provide them with the missing alpha-L-iduronidase enzyme.

I'm Jennifer Johnston, aged twenty. I live in the small village of Balerno on the outskirts of Edinburgh. I grew up with a happy and content childhood. I attended the local Primary school. I had a selection of friends and the mps was never a major part of my life. It only affected me slightly when I had difficulty sitting cross-legged in the classroom and at assembly. I had the odd problem at physical education, but that wasn't important. I managed.

I have Hurler Scheie and at the beginning of the trial I was 17 and my condition was deteriorating. I had been told I had a back condition known as spondylolithesis. Spondylolithesis causes the lower vertebrae to slip on one another and on me one of my vertebrae had completely slipped out of place. This caused me to be in excruciating pain most of the time. My mobility was greatly reduced and I had to resort to using a wheelchair. Even when sitting in the wheelchair I would still get these pains, mainly down my legs. I would use shopping trolleys to lean on while walking. It soon was too uncomfortable for me to leave the house. I found it unbearable to travel in the car. I tried all types of painkillers but nothing worked. The main way in which I eased this pain was not to sit or stand as this is what caused it. I leaned over things. While watching T.V I could not sit on a chair or sofa so used a round pooffee to drape my body over. For me meal times were not spent sitting eating at the table but leaning over it. Leaning caused my elbows and knees to develop fatty deposits.

Once my back started to affect me daily life took a dive for the worst. I found, after starting fifth year at secondary school it was getting harder and harder for me to cope with the average tolls of student life. I was suffering much pain walking from class to class carrying books and folders, then sitting still in a class which, for me, seemed like hours. In class you had to concentrate hard and take down many notes. I could not concentrate in class. I was thinking about, not the teacher but the pain in my legs, while seated. I would often squirm about holding on to my legs while sitting in the class. My back was starting to bend over a lot and I could only walk while holding on to my knees.

Stiff joints, caused by the MPS, made everyday tasks awkward, hard and sometimes impossible. I am very short for my age, 4 foot 7. Standard Grade Art and Design was effected, as equipment and material were out of range for my short stature and low reach. Writing became increasingly difficult, gradually slowing me down at school. I had to use a scribe for exams and wasn't a regular scribe user. I managed to write slowly, in class. In exams, I was surprised in having to spell simple vocabulary for the scribe - time spent unnecessarily.

Earlier, learning at primary school was difficult because, being physically disabled also classed me as having learning disabilities. This prohibited my learning path and placed me in-groups and classes for children with these special needs. I was excused from doing physical education and field trips were disallowed.

In later years of school life I had few friends, as my condition deteriorated and I was often laughed at, name called e.g. "dwarf" and picked on. This was often done everyday. I was alienated at school and always dreaded the next day. I often spent my lunch times in the library, alone, to avoid the unpleasant abuse, for being different. I only carried on at school in 5th year, for a few days, until it was just getting too difficult for my mobility and me. At home, I struggled on and passed Higher Biology with the help of the Outreach Teaching service. This was very hard for me as I found it unbearingly uncomfortable and awkward to sit and write, or type, because of my sore joints, easily strained neck and the constant pain I was enduring.

It was about a year after leaving High school, that the trial came along and I was still studying Higher Biology. Altogether, there were twelve patients. Before the trial began we were all told it was a double blind trial. This meant that six were going to be on the real enzyme and six would be on a placebo. A placebo is not the enzyme solution but a saline solution i.e. water.

At the beginning of the trial I had to undergo a sleep study at Great Ormond Street Children's Hospital, a walk test and a lung function test, a MRI scan, an ultra sound, an eye examination and then a physical examination.

For a sleep study small metal electrodes were positioned at different points around my head. They were stuck down with glue (which was very smelly). One electrode was also glued on the bottom of my leg, ECG pads were stuck to my chest and a tube positioned at the bottom of my nose. This measured carbon dioxide breathed out. Finally, a belt was tightened around my waist to measure how many breaths I took. Around thirteen wires led from my head to a machine which measured my brain activity while, I slept. This was the worst part - trying to sleep. I was lying alone in a very hot hospital, lots of noises around and lights shining in from corridors and rooms, being tied to the bed with wires protruding from my head. When I turned over they tugged (ouch).

The lung function test and walk test were both completed together. My lips were placed over a plastic tube, attached to the lung machine, my nose closed using a nose clip. I was asked to blow as hard as I could down the tube, with out stopping. As you blow

down into the lung function machine you gradually run out of breath, feeling there is nothing left. The lung function nurse then said "keep blowing" but you felt you were blowing nothing out. At the end I felt energyless and thought of the walk test I had still had to do. For the walk test I had to walk up and down a corridor continuously for five minutes. The first ever walk test was exceptionally hard for me. I often needed to take a rest. I could not manage to walk without stopping.

The MRI scan was scary the first time. I had to lie flat for a short time, which seemed like ages. There was a horrendous hammering noise pounding in my ears and I had to be deadly still. I felt trapped as I was enclosed in a tight tube, which I felt I could not get out of. The ultra-sound was easy. They squeezed cold jelly on my chest and moved the ultra-sound device about, to inspect my heart.

My eyes were examined with the doctor looking through a lens. He squirted coloured dye in my eyes. Any clouding glowed, as a luminous light was shone into them. He then squirted air into them, for just a second, measuring the pressure of my eye balls.

At the physical examination, my liver was measured at around nine centimetres. Physiotherapists measured the flexibility and movement of my joints. All of these different procedures were carried out constantly throughout different periods of the trial.

On the first day I was quite nervous. I did not know what was going to happen and would I receive the real enzyme? If I did, I wondered what it would feel like and would a reaction occur? Before being attached to the infusion everyone was given pre-medication (tablet form). This was to prevent any allergic reactions occurring. I was not good at taking this. I had great difficulty swallowing them. I tried them soluble but the taste was just too foul and so opted for them crushed. I swallowed them crushed for many months. I was always very anxious before and while taking the pre-medication and often retched them out afterwards.

A canula was used in the arm for the infusion. They had no trouble with this, with me and I was never worried by it. Altogether, the infusion lasted four hours and during the first hour different measurements were taken e.g. temperature, blood pressure. The double-blind trial lasted for six months and I had no complications. Mum and I travelled down from Edinburgh to Manchester and back again, the following night, every week, for two years. Once the double-blind trial was finished, we were all given the opportunity to receive the real enzyme, whether or not we had been given it before. I couldn't wait. This was what I had been waiting for. In the first six months there had been no significant change or benefits. I had been on the placebo. The disappointment was outweighed by the excitement of finally receiving the real enzyme. Before the trial started I told myself to look forward only to the six-month deadline, when I knew for sure I would receive the enzyme.

My liver gradually decreased from nine centimetres to six centimetres in approximately six weeks. I was told frequently that I was standing straighter and was not walking so much on my toes. I began to feel more energetic. The first walk test after receiving enzyme showed signs of improvements, as I could walk a little further. My tongue may have shrunk. I did not notice that it was ever large. But as the weeks went on, I began to swallow the tablets, without them being crushed. It was easier and more straightforward. This was a huge step for me and I've done this ever since. Even though I felt I had more energy, I felt restricted in using it because of my back condition. My joints started to become more flexible. It happened rather gradually. I noticed it in my fingers while writing and in my jaw, when cleaning my teeth.

Whilst the weeks went on the texture of my hair changed from being slightly kinky and rough to straighter and smoother. Under a magnifying glass you could see the difference between new growth and old, and where the changes had occurred. All the changes are continually ongoing and happening slowly. I realize this. The MPS gradually worsened over years as I grew up. Will it need just as long to improve? I would say it does. People who haven't seen me regularly, comment on how different I look from the last time they have seen me.

An enormous improvement, that I really appreciate, is that I am now able to sit a great deal longer and more comfortably than before. I have less need to lean and kneel over things. Pain caused by the spondylolithesis is less, also, while walking. Standing and showering I can now do for a short time. The bath would have always and only been used. While walking I often fell over and tripped up. One day when going to the Willink, on route to the taxi, the driver tripped me up with his long legs. I was expecting to hit the ground but, to my amazement, I ran with the trip and managed to keep myself up. Before the enzyme I would have certainly fallen. This shows that my balance has definitely improved. I now eat apples without them being cut up, something I would have never attempted before. I bite with that satisfying crunch - a crunch I've always wanted to make! The Dentist now has an easier job at examining my mouth as it opens a lot wider.

Jennifer with Dr Fitzimmons

My liver has been frequently measured. It has been getting smaller and smaller, until now, when it's virtually non-existent! There is still a lot of loose flesh, where it originally was. I'm hoping to lose this using a toning machine, so my stomach is completely flat- something I have always wanted. My lung capacity from the beginning of the trial in 2001 has increased an overall 25 to 30%.

I want to get back into studying so I am continuing with my education and have now applied for two courses at college, in law and computing.

I thank everyone at the Willink, but especially Dr. Wraith, and the team of professional and very hard working trial nurses, Jean, Jane and Caroline, for making my infusion days happy, relaxed and non-traumatic.

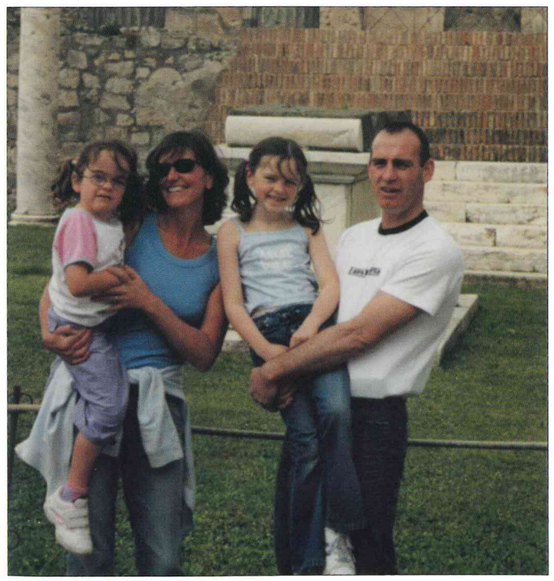
Since leaving the Willink I would like to show my appreciation to the staff at Ward 7B particularly nurse Leslie Stirton, Dr. Fitzimmons and Dr. Soutar. They have thankfully taken on the satellite centre in Scotland at the Gartnaval General Hospital in Glasgow and have been friendly and made me feel very welcome.

Naples

Christine Lavery

Ellie Gunary, Cheryl Pitt, the Wilson and Murphy families and I were guests of Genzyme at a two day meeting on the clinical management of MPS I. Key Note Presentations by Roscoe Brady and Emil Kakkis addressed the historical perspective and Enzyme Replacement Therapy as a treatment for MPS I. A highlight of the meeting was the three lay presentations. Barry Wilson spoke on having a daughter, Joanna with Scheie disease. Maria Murphy spoke on life with Tara who is five years old and has had a Bone Marrow Transplant for the classic form of MPS I. Finally Anna Serafini a representative of the Italian MPS Society spoke about life with the attenuated form of MPS I. All credit goes to Anna who spoke in her second language English.

After the meeting there was a little time to explore Naples and it is environs. The news that Enzyme Replacement Therapy, Aldurazyme™, for MPS would be approved for use in Europe in June left everyone feeling very upbeat. The picture below of Tara and Joanna at the top of Mount Versuvius is a poignant reminder of how high our dedicated researchers and Clinicians have to reach to make strides in the common goal of treatment for all those suffering from Mucopolysaccharide and related diseases.


The Murphy family at Pompeii


Tara Murphy and Joanna Wilson on the rim of Mout Versuvius

MPS Society Announces ERT for MPS I

Christine Lavery
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PRESS RELEASE

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For Immediate Release
12 June 2003

Enzyme Replacement Therapy (ERT) for Patients with Mucopolysaccharide Type I (MPS I)

During 2001 and 2002 twelve patients suffering from Mucopolysaccharidosis Type I from England, Northern Ireland and Scotland participated in a pivotal worldwide Phase III double blind placebo controlled study of the Enzyme Replacement Therapy, Aldurazyme, carried out at the Royal Manchester Children's Hospital.

Working closely with the patients on the ERT study in Manchester, the Society's advocacy team have experienced first hand the positive impact "Aldurazyme" has had on the lives of these patients. The Society for Mucopolysaccharide Diseases therefore positively welcomes 'Aldurazyme', the first drug approved for MPS I in the European Union.

MPS I is a life threatening genetic disease caused by a deficiency of the enzyme a-L-iduronidase. MPS I comprises a wide spectrum of severity and clinical involvement. Children with classic, severe form of MPS I, sometimes caller Hurler disease, have progressive developmental delay, and physical disability due to the early advancement of the disease and usually die before the age of ten years. Although it is known the enzyme cannot cross the blood brain barrier. Parents of these patients are likely to request ERT as a means of improving quality of life.

Some children and adults with the attenuated form of MPS I do not have progressive developmental delay and their physical problems advance more slowly. There are others whose disease pattern falls between the two ends of the spectrum. The attenuated form is sometimes referred to as Hurler Scheie and Scheie disease.

Jennifer's Story
Jennifer is 19 and has Hurler-Scheie. She has had worsening problems with mobility, especially with bending, stretching and walking due to the spondylolithesis caused by the disease. Walking and moving was further hampered by joint stiffness and pain. Everyday activities which people often take for granted had been severely affected as her bad back and joint pain prevented her from moving easily or lifting anything. Jennifer felt worried all the time as she felt she was getting worse and worse and she constantly felt tired. In January 2001 the Enzyme Replacement Therapy (ERT) trial started at the Royal Manchester Children's Hospital. One group of sufferers were placed on a placebo while another group were given enzyme treatment for the first 6 months. Jennifer received the placebo during this initial period of the trial and felt no change in her condition. At the end of this period, she was told that she had been on the placebo and that she would now be receiving the enzyme. Jennifer was looking forward to receiving the same benefits as the other group had had for the past 6 months. Jennifer has now been on the enzyme for 2 years. The impact on Jennifer's life has been enormous. She says she feels like a new person and the ERT has changed her life. Her mobility has improved dramatically, and she can now walk long distances. Jennifer is now looking at learning to drive and is keen to develop her independence. She is able to help around the house without the joint stiffness and pain she used to feel, and she doesn't get tired as she used to. There are additional benefits to ERT as well, as Jennifer's skin feels better and her hair is softer. Jennifer has also expressed how good she feels inside as she can feel that she is getting better.

Jessica's Story
Jessica has Hurler Disease. For nine years the family have lived with the knowledge that their ten-year-old daughter is dying.

Over the years Jessica has slowly lost her sight and her hearing and can only utter the word "Daddy". On some days she is in so much pain Jessica has to be given morphine and will sleep on a bean bag in the living room. Her head is very heavy and Jessica's family believe she has lots of headaches. Sadly, there is no way Jessica can show where she is hurting.

This is a very different Jessica from the bubbly enigmatic toddler who charmed everybody singing nursery rhymes and dancing to Robbie Williams. Sadly, as the mucopolysaccharides have deposited themselves throughout Jessica's body her life has ebbed away.

Data from European Fabry Outcome Survey Confirms Beneficial Effects of Replagal

Findings Presented at 2003 European Society of Human Genetic Meeting 5 May 2003 Birmingham

Yahoo Finance

TKT Europe - 5S AB today announced at the European Society of Human Genetics 2003 conference that participating European physicians of FOS, the Fabry Outcome Survey, reported data confirming the clinical benefits of Replagal(TM) (agalsidase alfa) in patients with Fabry disease. FOS is a European multi-center, clinical outcomes survey, which is designed to monitor the long-term effects of Replagal. Untreated patients are also followed in FOS, in order to increase knowledge of the natural history of Fabry disease. Of the 336 patients enrolled in FOS at the end of last year, 217 patients, or 65%, are receiving Replagal, with 58% treated for over one year.

"FOS is a remarkable tool for clinicians to gain access to important data relating to Replagal and in treating Fabry disease," stated Professor Michael Beck, who is a member of the FOS executive committee. "The findings generated from FOS are very encouraging as they confirm the benefits of Replagal in clinical practice. Considering the multi-organ involvement in Fabry patients and what we know from the natural history of the disease, it is astounding to see the effects of Replagal on the kidney, pain and quality of life, particularly over this relatively short period of time."

Long-term Follow-up in FOS Confirms Beneficial Effects of Replagal.
At the Human Genetics meeting, Dr. Michael Beck of the University of Mainz in Germany presented a data analysis of FOS, on behalf of the FOS European Board, showing treatment with Replagal has positive effects on renal and neurological manifestations of the disease. The renal findings were based on an analysis of 285 patients enrolled in FOS, of which 70% have received Replagal for at least 12 months. Upon entry into FOS, 65% of these patients had impaired renal function. After 12 months of treatment, renal function, as assessed by creatinine clearance or glomerular filtration rate, stabilized or improved in patients with mild to moderate renal impairment. Data on pain was also presented. At baseline, 85% of men and 63% of women were suffering from chronic pain and impaired pain related quality of life measured by the Brief Pain Inventory (BPI). Treatment with Replagal was accompanied by significant improvement in 8 out of 11 dimensions of the BPI (n=90).

About FOS
FOS is an outcomes database developed to provide further understanding about Fabry disease as well as to improve the clinical management of patients with the disorder. Eligible for inclusion in FOS are patients with Fabry disease who are receiving Replagal or are candidates for enzyme replacement therapy. Patients not receiving treatment, but who participate in the database, provide a potential source of matched control data. FOS was developed in 2001 by TKT Europe - 5S in close collaboration with leading European experts on lysosomal storage diseases. The Press Release Source: TKT Europe - 5S ABFOS European Board is responsible for the scientific analysis and publications from the database.

FDA Approves First Treatment for Fabry Disease

Food & Drug Administration USA

The Food and Drug Administration in the United States today approved the first treatment for patients with Fabry Disease, a serious metabolic genetic disorder affecting approximately one in 40,000 males. While it is believed that fewer females suffer the most serious consequences of the disease, they can be similarly and seriously affected as well. Because of a deficiency in an enzyme, alpha-galactosidase A, Fabry Disease causes certain fats to accumulate in the blood vessels over many years, leading to the involvement of various tissues and organs of the body, including the kidneys and the heart, which can then cause organ failure. As a result, patients with Fabry Disease often must cope with significant pain and disability and typically have a shortened life span.

The new product, called Fabrazyme (agalsidase beta), is a version of the human form of the natural enzyme produced by recombinant DNA technology. It is given intravenously. This replacement of the missing enzyme reduces a particular type of lipid (fat) accumulation in many types of cells, including blood vessels in the kidney and other organs. It is believed likely that this reduction of fat deposition will prevent the development of life-threatening organ damage and have a positive health effect on patients.

"This priority approval of an orphan drug illustrates FDA's commitment to approving innovative new therapies for patients with serious and life-threatening diseases quickly, based on response to treatment of biological markers likely to predict long-term clinical benefit." said FDA Commissioner Mark B. McClellan, M.D., Ph.D. "The orphan drugs program provides crucial incentives for innovators to develop new treatments for rare diseases. By approving this new biotechnology therapy under the 'accelerated approval' process, we are making this product available more quickly to patients who need it."

Fabrazyme was approved under an accelerated or early approval mechanism. This policy allows for expediting the approval of therapies that treat serious or life-threatening illnesses when studies of the products indicate early favorable outcomes that are likely to predict clinical benefit. The approval is based on I!surrogate endpointsI! -laboratory measurements or physical signs -for evidence of effectiveness. The surrogate endpoints are believed to be likely to predict benefit for the patient. In this case, the manufacturer of Fabrazyme (Genzyme Corporation, Cambridge, Mass.) has performed biopsies looking at the cells lining the blood vessels within the kidney and other organs in patients with Fabry Disease. Many (but not all) of the cells examined have shown significant clearance of lipid deposits in patients treated with Fabrazyme.

"A key part of our accelerated approval process involves further study of the new treatment after approval, to confirm clinical benefit," said Dr. Jesse Goodman, Director of FDA's CBER. "In this case, FDA has worked closely with the product developer to make sure that, despite the relatively small number of patients with this disease, all reasonable steps will be pursued to make sure that we learn more about the product's clinical benefits and long-term safety once it is on the market."

One of the requirements of an accelerated approval is that the sponsor completes a postmarket study verifying that patients will benefit from the product. Genzyme has committed to continue conducting their ongoing randomized placebo-controlled trial to verify Fabrazyme's benefit to patients and by assessing the drug's effects on the progression of kidney and heart disease and the incidence of strokes.

In addition, Genzyme is taking further steps to assure the availability of information to determine long- term effects of treatment with Fabrazyme. Genzyme has set up a patient registry to follow the long-term progress of patients who have been treated to better understand Fabry disease and to evaluate the long- term effects of treatment. Enrollment in this registry is voluntary.

FDA and Genzyme are also discussing a variety of novel statistical approaches to analyze data and better assess the effectiveness of the treatment, to augment the data collected through the clinical trial. The potential approaches being evaluated include measures such as within-patient analyses of trends in creatinine levels (a measure of kidney function) on placebo and on Fabrazyme, and modeling utilizing historical information from matched patients.

"Effective studies after approval are essential for collecting valuable evidence on the clinical benefits and longer-term effects of products approved on an accelerated basis," said FDA Commissioner McClellan. "FDA intends to evaluate whether the continuation of the Fabrazyme clinical trial, potentially coupled with additional analyses of data from patients who many not complete the trial and of data from patients receiving long-term treatment, represents the most effective approach. to acquiring important confirmatory evidence after approval."

In clinical studies of Fabrazyme, the main safety concern in patients receiving Fabrazyme was infusion reactions, some of which were severe. These include fever, chest tightness, blood pressure changes, abdominal pain and headache. Most patients also develop antibodies to the product and some patients who experience allergic reactions may need to be further evaluated. Because of the potential for these severe reactions, appropriate medical observation and support should be available when Fabrazyme is administered.

Orphan products are developed to treat rare diseases, or conditions that affect fewer than 200,000 people in the U.S such as Fabry Disease. Under the Orphan Drug Act, FDA provides modest grants to organizations to develop products to treat "orphan" diseases. The act provides a seven-year period of exclusive marketing to the first sponsor who obtains marketing approval for a designated orphan drug.

Jeans for Genes Research Update

Dr Claire Beesley

In the year 2000, the MPS Society through Jeans for Genes awarded a three year programme grant to the Institute of Child Health, London and the Willink Biochemical Genetics Unit, Manchester. In this report we detail the results obtained during the second year.

Institute of Child Health, London

At the Institute of Child Health, we continued to screen newly diagnosed MPS I, IIIA and IIIB patients for the disease-causing mutations. A total of 8 patients were analysed and all mutations were identified, 6 of which were novel. Liz Young and her colleagues in the Enzyme Laboratory at Great Ormond Street Hospital diagnosed a rare case of Sanfilippo type D (MPS IIID). MPS IIID is a deficiency of the enzyme N-acetylglucosamine-6-sulphatase (G6S) which is involved in the same biochemical pathway as the enzymes deficient in MPS IIIA and IIIB, hence similarities in the clinical phenotype. Only 12 patients with MPS IIID have been reported worldwide and no mutational analysis in humans had been carried out. Using information from the human genome mapping project we were able to screen the G6S gene from our patient and to discover the first mutations in a MPS IIID patient (recently published in the Journal of Medical Genetics, 2003;40:192-4).

Over several years, we have identified many novel mutations in the MPS I, IIIA and IIIB. We can predict that some of the mutations will be disease-causing but a large proportion result in the substitution of one amino acid for another in the protein chain. It is not possible usually to predict the consequence of such a change and it is necessary to investigate the precise effect on enzyme function to prove that these changes are indeed disease-causing. We have developed a new experimental system to investigate MPS I, both Hurler and Scheie, MPS IIIA and MPS IIIB mutations. It enables us to tell whether the mutation completely destroys the activity of the enzyme or produces some residual enzyme activity or an unstable enzyme. Some residual activity from the Scheie mutations has been detected but further work is required. Seven novel MPS IIIA missense mutations have been investigated and all have been shown to be disease-causing. Further work to investigate how the mutation affects biosynthesis and functioning is required. For this we need antibodies to the MPS I, IIIA and IIIB proteins. For MPS IIIA, we successfully purified a portion of the protein and injected it into rabbits. The rabbits invoked an immune response and produced antibodies to our protein. We have collected these antibodies and are currently assessing their specificity and preliminary experiments are encouraging. The next stage is to use the antibodies to look at the effect of the mutations on the enzyme. These experiments will be repeated to produce antibodies to human MPS I and IIIB proteins and mouse MPS IIIA and IIIB proteins.With the prospect of therapy for some MPS disorders, it is becoming increasingly important to determine whether patients selected for treatment have some residual protein present because of the potential immunogenicity of exogenous enzyme protein.

The major symptoms of Hurler and Sanfilippo syndrome are due to the accumulation of storage products inside the brain and CNS. Gene therapy using herpes simplex virus offers a means by which a therapeutic gene can be carried into the brain and produce functional enzyme. We have mouse models of MPS I, IIIA and IIIB and are using them to develop new virus vectors that can express the gene once injected into the brain. Initial experiments with a vector containing an element that improves expression of the gene in cells has shown expression of a marker gene 2 weeks after injection that has moved to other areas of the brain. The marker gene will be replaced by the MPS genes and experiments will be carried out to investigate correction of the disease phenotype.

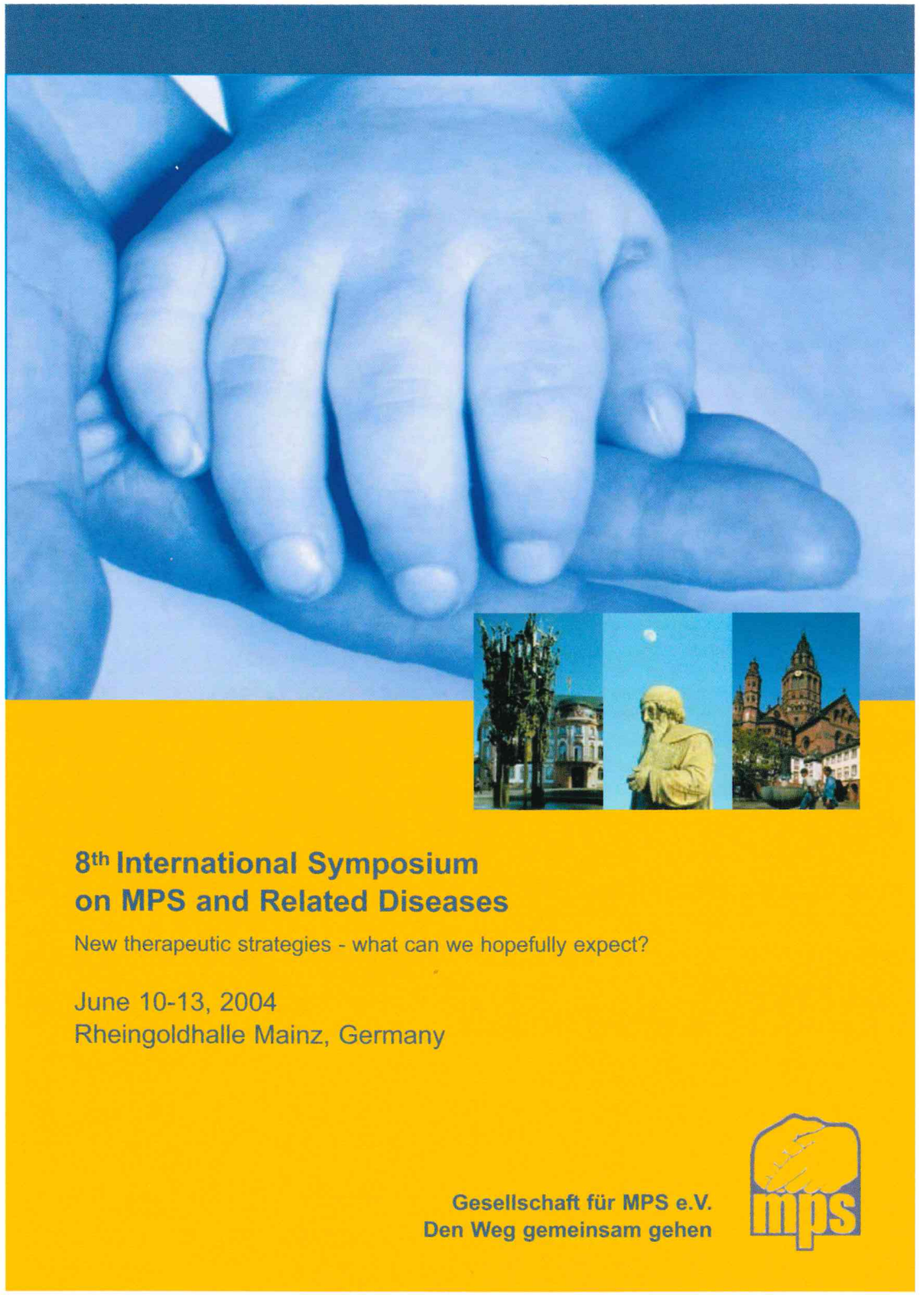
Willink Biochemical Genetics Unit, Manchester

Manchester has continued to develop its activities and its research program includes basic laboratory research aimed at future therapies for MPS and development of therapies that are more immediately relevant.

In current therapies Ed Wraith's involvement with enzyme replacement (ERT) is well documented elsewhere. In the field of transplantation in MPS I we are collaborating with other transplant teams in the USA and UK to include ERT with transplant, and the use of newer and less toxic drugs in the conditioning process.

In the laboratories a team lead by Ilaria Bellantuono has continued to investigate the potential of Mesenchymal stem cells in the management of MPS. We have already shown that these cells are easily isolated from the bone marrow of all people, and are particularly suitable to genetic manipulation – including in MPS. In the last year we have made progress investigating the basic properties of these cells and their growth characteristics in MPS and non-MPS patients. We have been particularly interested in the ability of these cells to become nerve cells, as has been claimed by other groups. This has obvious relevance to MPS where there is a functional problem with these cells. We have also established research into the interaction between these cells and blood stem cells as this may influence current transplant protocols, and as part of this work we are looking at the factors that influence where these various stem cells migrate to when injected. Clearly in order for them to have a clinical benefit they have to be able to get to where they are needed.

Although most of our work has focussed on MPS I we now have disease models for both MPS I and III. In this last year we have sought to replicate the work that we have already done on MPS I in the laboratory with MPS II cells. As we look forward we are aware of the problems that bony disease presents for both current therapy and future therapy, and as we look to see what these cells can do in this respect we will examine the applicability of the cells to include MPS IV and VI.



Dear Friends,

hereby, we would like to inform you, that the website of the

8th International Symposium on MPS and Related Diseases
10-13 June 2004
Rheingoldhalle, Mainz/Germany
is accessible from now on.
www.mps-kongress2004.de

Thank you very much.
With kind regards,

Bettina Wildi
German MPS Society

The MPS Society is currently exploring the possibility of group travel to the 8th International symposium on MPS related diseases and financial help to individuals and their families wishing to attend. There will be more information about this in the next newsletter.

Morquio Disease in South India

Elizabeth Herridge

I came to South India - to Chennai (Madras) as the wife of the Deputy British High Commissioner for a four year posting. I was soon 'persuaded' to go to teach English for an hour once a week at a very poor, orthopaedic hostel for children. These children are unable to attend normal schools as they will be shunned because of their orthopaedic problems. Their parents put them into the hostel and they receive education to Tenth Standard and operations if they need them.

There is no screening done on entry to the hostel. I had been there a short while when I noticed a little boy called Manikandan. I noticed his smile! He was new to the hostel, sat in the corner and everyone fell over him or trod on him. I spoke to him a few times (we dont speak one word of a common language!) and we became 'friends'. I was in the process of bringing in wheelchairs from England and I could see immediately that he would benefit from one. When it arrived, he was a new person! He wheeled himself everywhere and was able to live life at the same 'level' as the other children in the hostel. I then decided to take him to a doctor and he was 'diagnosed' as having rickets. This seemed sort of reasonable as he had sat for the first nine of years of his life inside a palm hut on the local beach which was his home! But... then I saw the movie about a young man with Morquio's Disease ... by absolute chance ... and I knew that Manikandan had the same disease. I searched out a geneticist who was happy to see us and did all the tests for free and it was confirmed. Unfortunately it was too late for any treatment but at least we knew what he had! Later I moved him to the Cheshire Home in Chennai and he is now attending a proper, good school for children with disabilities. He is extremely happy and is a star in this town! Everyone knows him! Manikandan's mother has been abandoned by her husband and she brings up three children in the hut on the beach and works on the roads for a daily rate of less than £1 a day. Manikandan now gets around much faster on a tricycle.

Imagine my surprise when at the start of another year of teaching English I found a brother and sister had started at the hostel. They presented identical symptoms to Manikandan so I took them to the geneticist who agreed, and we did the tests and they were positive. Their names are Marimuthu and Radhika. Their parents live about two hundred miles from Chennai in a small village and they are also daily labourers. They have one other daughter. I have now put them in Cheshire Home where all three can stay for the rest of their lives and they are very bright, especially Radhika, and they too have started at the same school as Manikandan. Marimuthu can no longer walk and has a tricyle supplied by the Inside Out Trust, a UK charity.

The three children are all between eleven and twelve. What distinguishes them from the other children are their lovely smiles. Radhika is an organiser and she looks after the two boys and is 'in charge' of their clothing trunks and generally keeps them in good order! Their parents come and see them at Cheshire Home. Cheshire Home also provides lots of activities and so they are really busy! It is about to be given a swimming pool which will help with physiotherapy for the three children.

We have had them at home a lot and we have come to think of them as 'our' children. I will be looking after Manikandan for the rest of his life and I have got UK sponsorship for Marimuthu and Radhika for life for their education, transportation and costs at Cheshire Home. I have asked the sponsors to write to them regularly telling them about life in England. If anyone from MPS Society would like to write to them with odd bits of news and photos (though they cant write back in English).. the address is Radhika, Marimuthu and Manikandan, c/o Maureen Murari, 97 Landons Road, Chennai 600-010, Tamil Nadu, South India.

Morquio is quite prevalent here compared with UK and now many more people in this town have been educated about MPS diseases thanks to these three lovely children.

Marimuthu and Radhika phoning their mother who lives in a village two hundred miles away from Chennai

Benefits Change - Incapacity Benefit

People with mental health problems and people with disabilities could lose their right to incapacity benefit and be forced into work under government plans announced in March 2003.

The changes centre on the personal capacity assessment (PCA) which shows whether a person is incapable of work for the purpose of benefits.

The government wants to remove a clause in the law which states that a person would be seen as unable to work if there was "a substantial risk" to their mental or physical health if they did so.

This raises concerns that some people could fail a PCA but still be unable to work because a secondary factor, such as stress, triggers the condition.

This is the second time the government has attempted to do away with the clause. It was initially removed from legislation in 1997, a move ruled unlawful last year after a person who had previously been entitled to incapacity benefit took the government to court.

European Disability Rights Law

A draft of Europe's first comprehensive disability rights law was launched in March 2003 by Richard Howitt, president of the European parliament's disability rights group of MEP's. The new law would extend the UK's Disability Discrimination Act to make discrimination illegal in social security and to ensure manufactured goods are designed to be used by disabled people.

Do Housing Grants Discriminate against Disabled Children?

Homes Fit for Children (HFC) is a campaign to end what it describes as the “gross unfairness of the present grant system”. Campaigners claim that although the Disabled Facilities Grant exists to provide funding to adapt homes, only the very poorest families qualify for the means-tested grant. They want the law changed so that children can apply for the DFG themselves, bringing an end to what they describe as “age discrimination”. A government spokeswoman responded that children could not be allowed to apply as all of them would qualify for a 100 per cent grant, even if their parents were millionaires! HFC consider it unlikely that millionaire parents would want to go to the bother and bureaucracy of applying for the DFG! Research recently carried out by the Joseph Rowntree Foundation shows that 90 per cent of families with a disabled child have had a housing related problem and that they are more likely to live in poorer housing conditions than families on a similar income with a non-disabled child.

For a copy of the research Housing Matters: national evidence relating to disabled children and their housing, priced £13.95, call 01235 465500 or email direct.orders@marston.co.uk If you have experienced problems with the Disabled Facilities Grant, HoDis are keen to hear from you. Contact them at HoDis, 17 Priory Street, York, YO1 6ET, 01904 653888, www.hodis.org.uk

Direct Payments Push Needed

Half the local authorities in England have less than 30 users of direct payments each, according to government figures. From this month, all local authorities have a duty to offer disabled people the option of using direct payments, a system which gives them cash to pay for their own care services. The Social Services Inspectorate figures show an increase in people using the scheme from 5,423 in 2001 to 7,882 in 2002. But in 73 of the 147 areas there were less than 30 people - including older people, carers and disabled children -using direct payments. Nick Danagher, co-chair of the National Centre for Independent Living, said service providers were still failing to publicise direct payments to disabled people and often presented them in a “negative light”. “

Disability Rights Commission

FORMAL INVESTIGATION INTO WEBSITE ACCESSIBILITY FOR DISABLED PEOPLE. COMMENCING 28 MARCH 2003

NOTICE OF INVESTIGATION AND TERMS OF REFERENCE

Notice is hereby given, pursuant to sections 3(1), (4) and (5) of, and paragraphs 1 and 2(1), (2) and (4) of Schedule 3 to, the Disability Rights Commission Act 1999 (DRCA), of the holding of a formal investigation into website accessibility for disabled persons, and the terms of reference for the investigation.

The investigation is in connection with the duties of the Disability Rights Commission (DRC) under section 2(1) of the DRCA.

Michael John Burton and Christopher Holmes, Commissioners, have been nominated by the DRC to conduct the investigation on its behalf and, for that purpose, are authorised to exercise any of the functions given to the DRC by section 3(5) of, and Part Ior Part IV of Schedule 3 to, the DRCA.

The terms of reference for the investigation are as follows:

to conduct, by examining a representative sample of sites, a systematic evaluation of the extent to which the current design of websites accessed via the Internet facilitates or hinders use by disabled people in England, Scotland and Wales;

to produce an analysis of the reasons for any recurrent barriers identified by the evaluation, including a provisional assessment of any technical and commercial considerations which are presently discouraging inclusive design;

to make recommendations for further work which will contribute towards enabling disabled people to enjoy full access to, and use of, the Web.

For the avoidance of doubt, and subject to any revision of the terms in accordance with the DRCA, the terms of reference do not confine the investigation to the activities of one or more named persons, and so shall not extend to an investigation of whether any person has committed or is committing an unlawful act for the purposes of paragraph 3 of Schedule 3.

Michael John Burton and Christopher Holmes,
Commissioners

For and on behalf of the Disability Rights Commission

28 March 2003

Change by advice, conciliation and legal enforcement .

Leisure Time Stories Wanted

A project funded by Joseph Rowntree Foundation would like to hear about the experiences of young disabled people (aged 12 -25) in their leisure time. This project is the continuation of a report published in 2002 -*Hello! Are You Listening?* Disabled Teenagers' *Experience of Access to Inclusive Leisure*. The popular parts of this report were the sections written by young disabled people. We want more people (especially those who plan and deliver services) to know about the reality (good and bad) of the lives of young disabled people. The service planners have told us that listening directly to young disabled people helps them to understand their lives and plan services for them more appropriately. A young disabled women involved in the project said, '*People can engage with and remember stories'.*

The stories will contribute to a book to be published in the Autumn. Contributions can be made in any way writing (prose and/or poetry); photographs; artwork; and/or talking into a tape recorder. Contributions can be of any length and can cover any aspect of your life you would like to tell other people about.

Contact Pippa Murray, 5 Moor View Drive, Sheffield S8 OHL Tel: 0114 235 9276 e-mail: pippa@pippamurray.fsnet.co.uk

Dreams Come True!

The Dreams Come True Charity exists to bring some laughter and happiness into the lives of terminally or seriously ill children (aged between 2 -21) in the UK. Quite simply, it makes their most treasured dreams come true, whether that is going to Disney World, or meeting a favourite pop star or even visiting Santa in the Arctic Circle.

If you would like to apply to the Dreams Come True Charity, send a letter telling them a little about the child and if possible an idea of their 'dream'.

As soon as they receive this, they will send an application form. Return the completed form together with a letter from a Doctor or Consultant confirming the child's condition. Please send your letter to:

Dreams Come True Charity,
York House,
Knock hundred Row,
Midhurst, West Sussex,
GU29 9DQ

Publications

Disability Alliance has published Charges for Community Care Services. This guide is aimed at disabled people and carers in England and Wales, and explains what the law says about how councils should calculate charges and what people should do if they think they are being over-charged or can't afford to pay. It also contains a full list of benefits available to disabled people and carers and information on useful organisations to approach for further help and information. Tel 020 72478776

Skill, the national bureau for students with disabilities, has published Into Higher Education 2003. This guide features advice on completing applications, getting support, grants and benefits as well as student experiences. The guide also includes listings written by institutions offering higher education courses and contact details for their own disability co-ordinator. Priced £12.50 for professionals. £2.50 for disabled students. Tel 0207 500620 or email: skill@skill.org.uk

RADAR has published a range of books aimed at people who care for disabled children. Children First - educational services for disabled children (£5), provides useful information and advice for teachers, families and young people, about educational resources throughout the country. Children First a guide to services for disabled children (£5), aims to provide valuable information to improve the quality of life for carers of disabled children. Topics covered include social services, housing, transport, education and leisure. Children First a guide to the needs of children in school (£10.50), is a practical guide to the educational implications of many forms of disability. It addresses many issues which teachers may need to consider when preparing to take a disabled child into their class. To order copies of the Children First series contact RADAR, 12 City Forum, 250 City Road, London ECIV 8AF, or tel 020 7250 222, or email: radar@radar.org.uk

Jeans for Genes Campaign 2003

Antonia Crofts

Time has flown by and once again the Society is heavily involved in the Jeans for Genes campaign 2003.

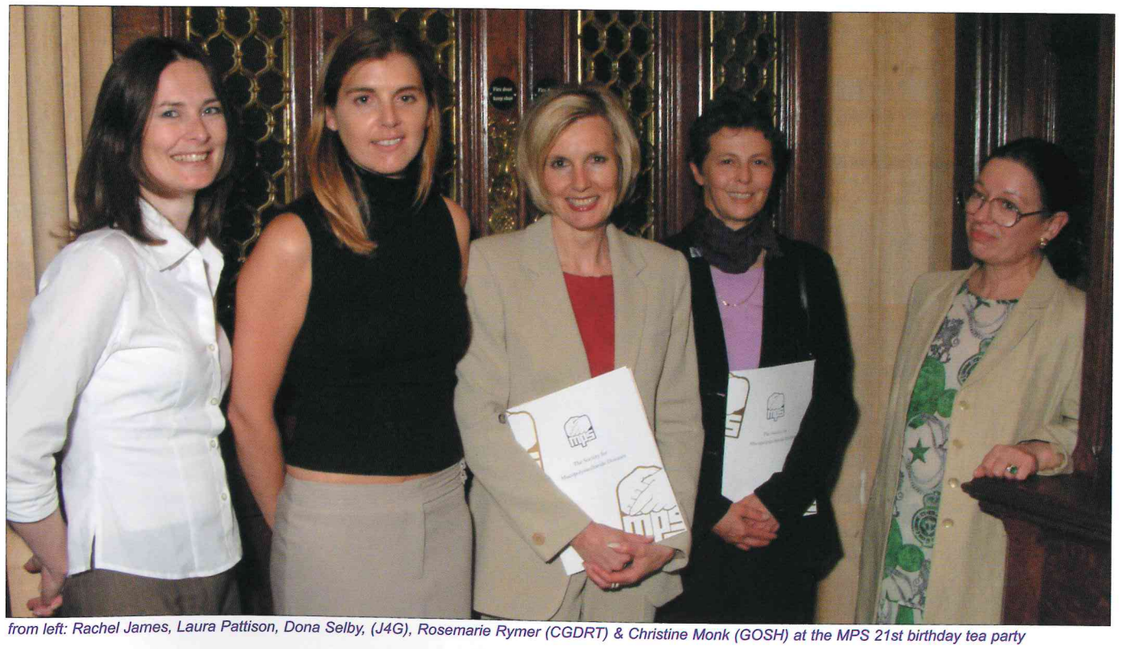
For those of you unfamiliar with the concept, Jeans for Genes is a national campaign operating from headquarters at Great Ormond Street Children's Charity, London. Jeans for Genes consists of several partner charities active in the field of genetic diseases who each receive money raised from the campaign which is in turn put towards funding for research and advocacy support. 60% of the Jeans for Genes money received by the MPS Society goes towards research and 40% towards advocacy support. In 2002 the 40% was the equivalent of funding the entire advocacy support service for one year.

Held on a yearly basis Jeans for Genes day this year will be Friday 3rd October 2003. In preparation for the big day each of the charities and staff at the campaign headquarters have to work to a strict timetable throughout the year to ensure that the advertising, marketing, merchandising and mailouts are prepared well in advance of the actual day. This gives everybody time to pro-actively promote Jeans for Genes.

As one of the partner charities of Jeans for Genes receiving a proportion of the money raised the MPS Society has a duty to raise awareness of Jeans for Genes. Staff at the MPS Society are in frequent contact with both member families and Jeans for Genes staff liasing over possible leads for stories from members who are willing to support the campaign by giving media interviews, either in the local or national press, on the radio or the TV. Some people really enjoy this opportunity to "tell their story" whilst also raising awareness of both MPS diseases and Jeans for Genes. Other members are happy to give talks at local schools. We often get families who are willing to talk but need a little help in preparing what to say. School assemblies can often be quite fun especially when you get the audience involved too!

Other ways of supporting both Jeans for Genes and the MPS Society are by offering a small amount of time to distribute leaflets, posters or sell badges. Schools are often a very good place to drum up some support as children really enjoy the opportunity to get out of their school uniforms and into jeans for the day! However, the work place would also be a good place to try wearing your denim instead of a suit. After all, it is for a good cause! If you are interested in selling badges for £1 each, please contact the MPS Society as we can order these for you on-line.

Further information on any of these ways to support the campaign can be found in the Jeans for Genes fundraising packs available from the MPS Society or directly from Jeans for Genes on freephone 0800 980 4800 or by visiting www.jeansforgenes.com. You can also call the MPS Society if you have any queries relating to Jeans for Genes or would like to offer your support as a volunteer.

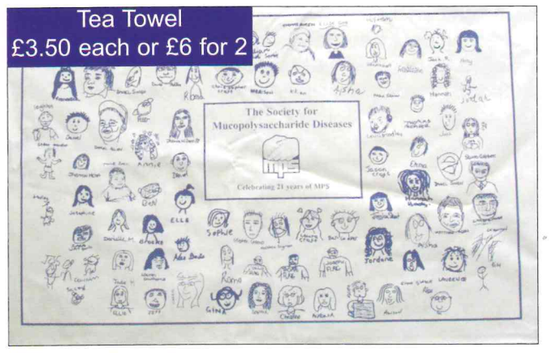


from left: Rachel James, Laura Pattison, Dona Selby, (J4G), Rosemarie Rymer (CGDRT) & Christine Monk (GOSH) at the MPS 21st birthday tea party

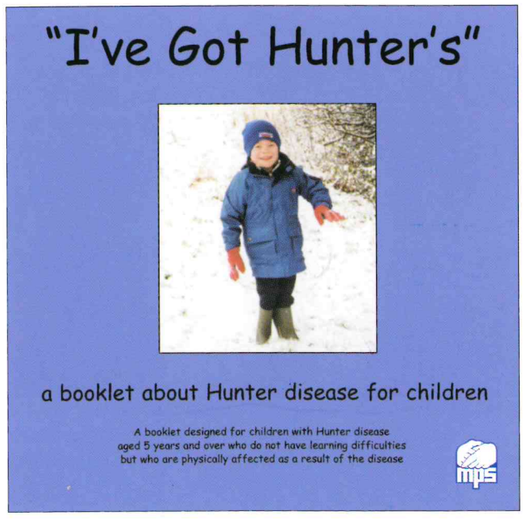
To order please see enclosed Promotional Goods Order Form

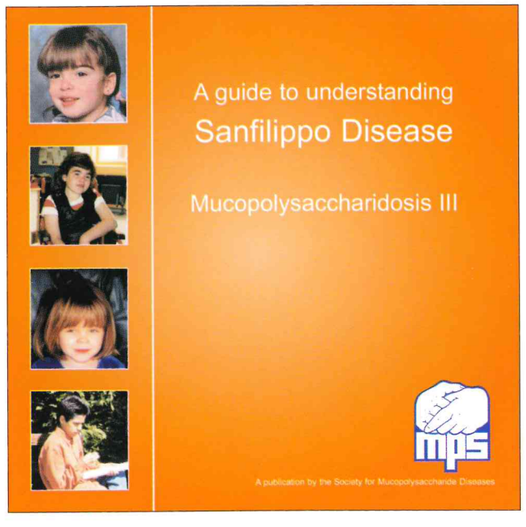
Gift Wrap

21st Birthday Celebration Items

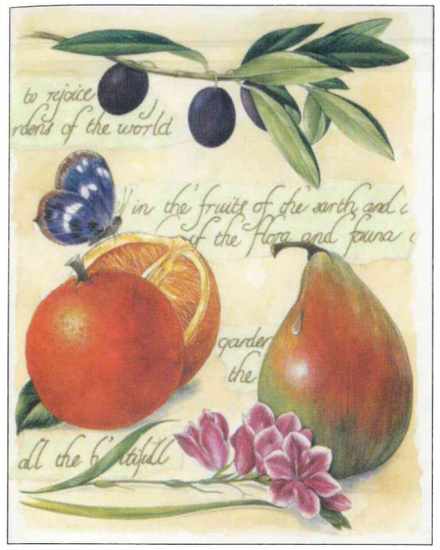


Booklets - see publication list enclosed

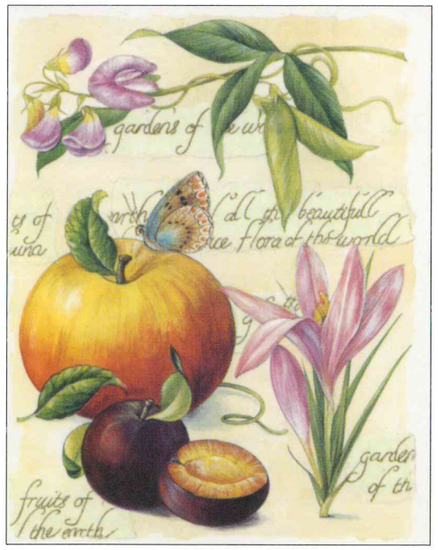


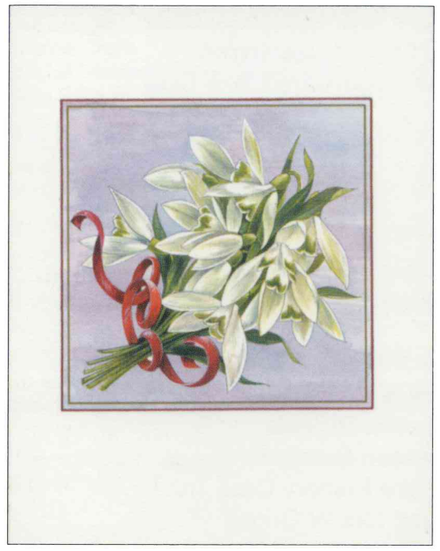


Notecards

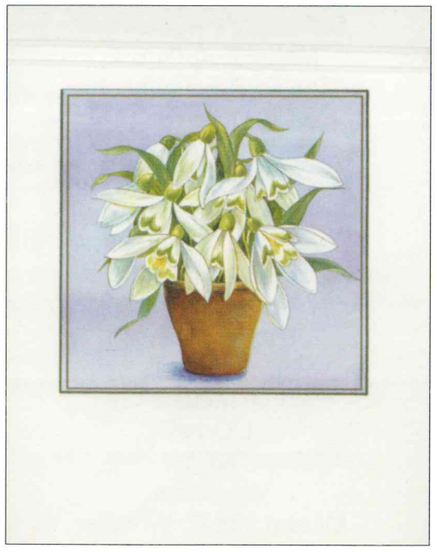


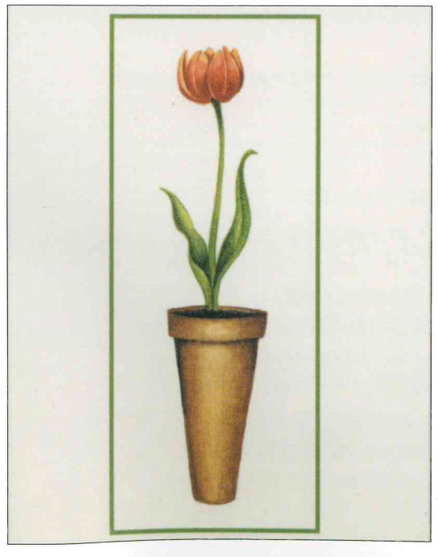
Fruit & Flora
10 pack of 2 designs
£2.50





Snow Drop
10 pack of 2 designs
£2.50





Tulip
10 pack of 2 designs
£2.50

Donations

Abbott Laboratories Ltd
The Sir Jules Thorn Charitable Trust
Actelion Pharmaceuticals
Buckinghamshire Building Society
Cane Adam Decorating Materials - Farnham
Haroon Aktar
Allied British Bank (GB)
David Gosling
Colin and Kath Jones
Elsie and Frank Brown
Ross Stuart
Philip Barker Charity
Bostik Findley Ltd
V Lazenby
Marks & Spencer - Milton Keynes
Sainsbury's - Chesham
Mattel Europa B.V
The PPP Foundation
Norsk Hydro (UK) Ltd
Transkaryotic Therapies inc.
Joseph Strong Frazer Trust
Tesco - Amersham
Genzyme Therapeutics
M Seeber - Harrogate
Susan Lowry - Hertfordshire
Marjorie Taylor
Debenhams
Martin McCloskey
L'Oreal
TKT 5s
Val Evans
Mr and Mrs R Wheeler
KWI Grinding Ltd
Doug & Alison Gunary
Staff at Stanshawes Service Station
The Baily Thomas Charitable Fund
Mrs Cock
Clydesdale Bank PLC
Mrs J Palmer
Promar International

Collection Boxes

Wilma and Peter Robins
G & W Tiles St Clears
St Clears Post Office

Fundraising

Overleigh St Mary's Primary School
Nags Head - Farndon, Cheshire
Valerie Casey - Catalogue Sales
Margaret Mount
Mrs Anne Rennie
Northgate
Jean Mercer - 10K Road Race
Polmont Loyal Rangers Supporters Club
Sandra Singh - Half Marathon
Wintershall Tennis Tournament - Surrey
Pitteuchar West Primary School
Charity Flowers Direct
Drayton Manor Park Staff
Karen & Andrew Weedall - Webb Ivory Sales
Kim Coney, Jane Coney, Erica Coney - Sponsored Walk
Compaq Computer Ltd Social Club
Rotary Club of Rutherglen
Dave Foster - Sponsored Bike Ride
Greenfield Methodist Church - Coffee morning

Stamps & Foreign Coins

Norman J Wigley & Partners
Chris & Dawn Jones
Letitia Ricketts
Alison Sawyer
West Wiltshire Primary Care Trust
Mr and Mrs W Griggs
Karen & Andrew Weedall
Mrs C Garthwaite

In Memory

Sarah Lowry
Gordon Wilson
Jeremy Stephen Papworth
Mr S J Blanch
Miss Jacqueline Elmer
James Edwards

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YOUR HELP NEEDED

Do let us have your stories, experiences 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 on the left.

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.

Photos

If you would like copies of any of the photographs inside this edition please contact the office.

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email: mps@mpssociety.co.uk
Web site: www.mpssociety.co.uk
Fabry: www.fabry.org.uk

NEWSLETTER DEADLINES

Autumn
30 September 2003

Winter
17 December 2003

Spring
31 March 2004

SUMMER
30 June 2004

Bounce in Denim...

On Jeans for Genes Day, everyone across the UK is being invited to jump into their jeans and donate £1 to the appeal.

In the UK, one baby in every 33 is born with a genetic disorder or birth defect. Please help **Jeans for Genes** to raise £2.5million to fund pioneering research and to provide valuable support services for affected children and their families.

For your FREE fundraising pack,
call freephone 0800 980 4800
visit www.jeansforgenes.com
or return the coupon below

Whatever you do on Friday 3 October

Do it in Denim

Jeans for Genes Day 2003

Eight national charities **working together** to help sick children

Great Ormond Street Hospital Children's Charity
Reg. Charity No. 235825
©1989 GOSHCC

The Primary Immunodeficiency Association
Reg. Charity No. 803217

The Chronic Granulomatous Disorder Research Trust
Reg. Charity No. 1003425

The Society for Mucopolysaccharide Diseases
Reg. Charity No. 287034

The Jennifer Trust for Spinal Muscular Atrophy
Reg. Charity No. 327669

Rett Syndrome Association UK
Reg. Charity No. 327309

Batten Disease Family Association
Reg. Charity No. 1084908

The Haemophilia Society
Reg. Charity No. 288260

The net proceeds from the 2003 Jeans for Genes Campaign will be distributed among the eight charities. Jeans for Genes Campaign™ is a reg. charity no. 1062206.

Registration Coupon

If you registered with the appeal in previous years you will automatically receive your pack in early September

Name

Position

Organisation

Address

Postcode

Telephone

Email