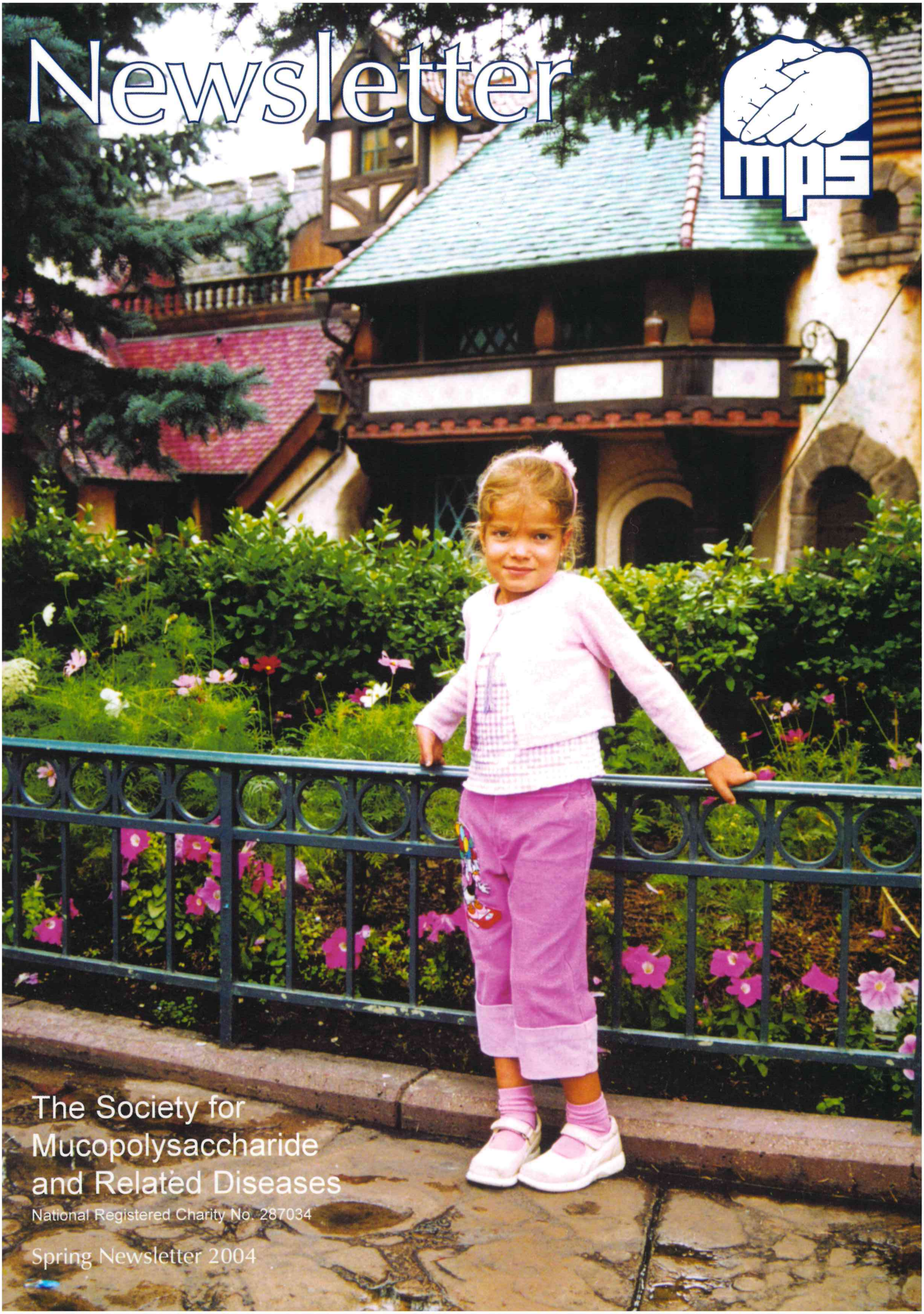
Newsletter



The Society for Mucopolysaccharide and Related Diseases

National Registered Charity No. 287034

Spring Newsletter 2004

Care Today, Hope Tomorrow

What is the Society for Mucopolysaccharide Diseases?

The Society for Mucopolysaccharide Diseases (the MPS Society) is a voluntary support group founded in 1982, which represents from throughout the UK over 1000 children and adults suffering from Mucopolysaccharide and Related Lysosomal Storage Diseases, their families, carers and professionals. It is a registered charity entirely supported by voluntary donations and fundraising and is managed by the members themselves.

The Society has the following aims:

- To act as a support network for those affected by MPS & Related Diseases
- To bring about more public awareness of MPS & Related Diseases
- To promote and support research into MPS & Related Diseases

How does the Society meet these Aims?

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

Telephone Helpline
Includes out of hours listening service

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

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

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

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

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

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

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

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

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

Front cover photograph: Sofia (MPS III)

Newsletter

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Registered Charity No. 287034

Newsletter Deadlines

Summer	1 Jun 2004
Autumn	1 Sep 2004
Winter	1 Dec 2004
Spring	1 Mar 2005

Subscriptions
Subscriptions may be taken out from the UK or Overseas by contacting the MPS Society's Office.

Spring 2004

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Note from the Editor

Dear All

Welcome to the Spring 2004 edition of the MPS newsletter. Many thanks to everyone who gave us feedback on the last newsletter and for your contributions to this edition. The newsletter is one of the main ways in which you can get your opinions, news and stories on issues related to MPS and Related Diseases read by others, throughout the UK and even across the world!

On the following pages you will find all manner of articles, updates and stories which we have collated over the last few months. We are always on the look out for anything newsworthy. Just as important are the photos that accompany each feature, not to mention the all-important front cover. Do you have a photo that you would like to see on the front cover of the next newsletter? If so, get in touch with me at a.crofts@mpssociety.co.uk.

Fundraising is a key aspect of our work at the Society to ensure our services continue to go from strength to strength. As a new venture we are pleased to offer advertising space in the MPS newsletter for companies and organisations wishing to promote their products and services. Give the Society a call for further information.

I hope you enjoy this edition!

Antonia Crofts, Editor

The Annual General Meeting of the Society for Mucopolysaccharide Diseases will be held at 6.30pm on Saturday 1 May 2004 at Splash Landings Hotel, Alton Towers, Staffordshire.

Chief Executive's Report

To say the last three months have been a challenge would almost be an understatement. As you will read, clinics in Newcastle and Birmingham have taken place and registrations to Alton Towers have taken the Society by storm. Although the deadline has passed we still have a small number of rooms left but only for occupancy of up to four people. I am sorry but all the six-bedded rooms have been taken.

In January the Society was invited by the Rt. Hon. Gordon Brown MP to host a reception and children's party at number 11 Downing Street on 6 April 2004. Naturally we are delighted to have such an opportunity but only sorry we couldn't invite all our members. To be absolutely fair to everyone we put all our member families in a hat whose children fitted the age range 5 to 11, are able to negotiate steps and stairs, and who didn't have the opportunity to attend the MPS Downing Street Christmas party back in 1998. We also had a reserves list.

Where are all the siblings? Over the years many families have indicated that their siblings would benefit from an event of their own. We have in the past organised sibling workshops as part of the conference programme but the Sibling Activity Weekend is something really different. Due to the small number of bookings we have cancelled the weekend at the beginning of September but the Sibling Weekend 16-18 July 2004 is going ahead as we have reached ten children. We still have room for 40 more. So find the booking form that came with the last newsletter or ring Alison or Sam for a new one.

One of the major challenges continues to be the difficulty facing the families of children and young adults with Fabry Disease and MPS I to achieve NHS funded Enzyme Replacement Therapy. There have been some successes, particularly for the members with Fabry Disease, but when it comes to MPS I you have to wonder why the UK ever took a lead role in developing European Orphan Drug legislation and the British Government signed up to it. On 3 March 2004 Dr Ed Wraith, Dr Atul Mehta and I took our concerns to a meeting with the Minister of Health, John Hutton MP. The meeting was constructive and the Minister clearly indicated his willingness to look at a way forward for funding ERT for rare diseases. In case things don't improve, if you are a sufferer of MPS II or MPS VI or parents of children with these diseases either on a clinical trial or expecting to receive ERT when the drug is approved by the EMEA, it may be wise to start briefing your MP and copying the MPS Society into any correspondence.

We are delighted that Cheryl Pitt, the Society's Research Co-Ordinator, has been invited to give presentations at two International Symposiums on MPS disorders this year. The WORLD Lysosomal Disease Research Network Symposium takes place in Minneapolis, USA, in May, and the 8th International Symposium on Mucopolysaccharide and Related Diseases takes place in Mainz, Germany, in June. Cheryl's work over the last year has been to conduct a pilot study on the psychosocial outcomes of bone marrow transplant for children and young people affected by MPS I Hurler Disease. The findings from this study will be presented at these two events. With the pilot project now complete, Cheryl is now busy preparing a main study which will explore the psychological and social development of MPS I patients post-BMT on a wider scale and in a more systematic way, inviting all families affected by this disorder in the UK to participate. This is a very important piece of research, which we hope will lead to a greater understanding of the long-term outcomes of bone marrow transplant for MPS I, and will help to improve quality of life for these individuals. This research is also being greatly supported by the University of Minnesota, who have been conducting research into the neurological and development trajectories of MPS disorders with and without BMT for some years.

As you will read in the article 'Relocation Relocation', Alison West and I have been seeing rather a lot of Cambridge recently. In fact until then the number of visits annually could be counted on one hand. However, apart from the privilege of helping our guest members from Italy, France and South Africa relocating for a year for the MPS II clinical trial, it has allowed us to set aside time on most visits to see Sue Stuart. You may recall having read in the Newsletter that Sue, mum of Jessica who died from Hurler disease in July 2002, was involved in a road traffic accident. Sue has now moved out of neuro critical intensive care onto the rehab ward.Whilst progress is slow, Alison and I have seen some over the past two months and it is lovely that Sue now recognises us when we visit and greets us with a huge smile. We send our best wishes to Sue, her husband Peter and daughters Hollie and Annie.

Christine Lavery
Chief Executive

Meet One of Our Office Volunteers!

Gyorgyi Barnard

Hello everyone! My name is Gyorgyi. I have been volunteering at the Society since March last year, and willingly spending most of my Fridays in the MPS office.

I came over to England from Hungary four years ago with high ambitions; I didn't speak much English, except the odd greeting and some basic phrases. Since then I have been very busy studying. I completed an advanced Cambridge English exam, and I just recently finished my HNC Business Studies.

Working for the Society is a chance for me to get some work experience in an office environment, and find out what office life is like, before I enter the corporate business world. So far I have learnt a lot, and I find the work very useful and interesting, since I have never worked in an office before. I also enjoy learning about MPS and Related Diseases.

Editor's Note: Our thanks goes to Gyorgyi for all her hard work over the past year. We wish her the very best for the future as she now leaves us to pursue other interests and embark upon a new career.

Gyorgyi Barnard

Cheryl Pitt

Spotlight on... Cheryl Pitt

A Poem by Cheryl Pitt aged 36¾

A researcher's life is a thoughtful one, especially if you are me,
There are miles and miles of road ahead, and I mean that metaphorically.
There is no beginning and there is never an end, you just start again where you finish,
When one question is answered another appears and hopes of a conclusion diminish.

You get to meet people and travel the world, but mostly I whizz around Britain,
From Glasgow to Penzance, armed with a clean pair of pants, I search endlessly for homes that seem hidden.
But with great cheer and a smile you welcome me in and sometimes I even get refreshments,
We talk about life and your wonderful kids, and I ask you endless amounts of questions.

But most of the time I sit in my office, tapping away on the computer,
You see there are many other jobs in a researcher's life besides being a commuter.
It's been almost a year since I started work here and there is so much I have learnt,
I've met some great minds and made some great strides, and even stood up at the lectern.

So for now I'll bid you farewell, and wish you all well, until the next time we meet,
Thanks to you all who have given up time, and to those of you I'm still yet to greet.

News from the Management Committee

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed at the Management Committee Meeting in February.

Personnel

The Trustees congratulated Sophie Denham on passing the first year of her two year training to achieve a Diploma in Social Work. The Trustees supported the confirmation of Sophie's second year (85 day) placement with a statutory authority.

Recognising significant increase in the advocacy work, and to ensure that the Society continues to meet the needs of all its members, particularly those who rely on the core areas of support such as disability benefits, home adaptations, palliative care and special educational needs, the Management Committee with grants from TKT5s and Genzyme, have agreed to a new post of Senior Advocacy Support Worker to address the areas of work resulting from new therapies.

Accounts

The Chief Executive spoke to the Society's draft accounts. These were considered by Trustees and were agreed. It was noted that the Society's administration costs were just under 10% and that there had been a considerable increase in restricted income. It was agreed the Society continues to need the help of its members in fundraising.

Policies

All trustees hold their own file containing all the Society's agreed policies. Policy statements on the use of funds in animal research, stress management in the workplace, and internet and e-mail usage by employees were considered and agreed. A gifts and hospitality policy as well as a whistleblowers policy were also agreed.

Introducing the Society's Trustees

Continuing on from the Winter 2003 newsletter, we would like to introduce two more members of the Society's Board of Trustees.

Wilma Robins

Trustee

I am Wilma Robins, a name still familiar perhaps to families who have been members of the Society for many years.

Wilma Robins

I am the longest serving Trustee and after undertaking many a key role on the Management Committee, I am very content now to use my experience to provide an overview of matters and allow newer Trustees to have the specific role responsibilities.

Some of you remember the red MPS poster of our son, Gethin. Gethin was born in 1979 and diagnosed with MPS I Hurler Syndrome, at the age of ten months, in Great Ormond Street Hospital.

Gethin's diagnosis was two years before the founding of the Society and we felt terribly alone during this time, thinking there was no other family like us with something called mucopolysaccharide disease. When we heard of the support group and met Christine, and attended the first family conference in Birmingham in 1983, our lives were transformed.

Sadly, Gethin died in 1984 at the age of 5½, but my husband Peter and I continued to be involved with the Society, attending the family conferences and looking after Matthew who had Hunter Syndrome and Natalie who had Sanfilippo, on the children's outings.

I volunteered to help in other ways and began to assist families with their disability benefit claims, my experience as a civil servant being invaluable to deal with the overly complex forms, and appeals process.

Eventually I became a Trustee and I am very proud to have served on the Management Committee when the Trustees were faced with the development of the organisation to comply with the challenging intricacies of the law in respect of charitable organisations, and other legislation on employment and data protection.

Today the MPS Society is respected throughout the world as the leading authority on the management of mucopolysaccharide and associated diseases, as well as being a key participant in the most innovative of developing research practices.

Never forgetting, however, that the care for our families will always be integral to our very existence as a family organisation. ■

Adam Turner

Trustee

I became a Trustee in September 2001 after attending the Northampton BMT Conference and chipping in after a presentation on education.

My son, Isaac, was born in November 1998, diagnosed with Hurler disease, MPS I, in July 1999, and underwent Bone Marrow Transplant in the summer of 2000. In June 2003 my wife Lou gave birth to a baby girl, Eliza, who is unaffected by MPS disorders.

I was born in May 1970 and have spent most of my life living in Manchester. In September 1994 I qualified as a secondary school teacher, and taught in a secondary school in Bacup, Lancashire for seven years before becoming Head of History at a secondary school in Darwen, Lancashire. Secondary school teaching is very demanding and offers new challenges every day, but can be very rewarding.

The news of Isaac's transplant, treatment and ongoing problems has brought a fantastic response from pupils at both schools I have taught at, and hundreds of pounds have been raised for charities including the MPS Society. This is a side of teaching which cannot be measured by government targets and league tables.

Speaking of league tables I have been a life-long Manchester United supporter. In fact, they are my local team and before the births of my children, I used to watch them play.

In the rare moments of spare time I can snatch I try to indulge my love of shopping for records, playing the guitar and writing songs, reading and continuing my ongoing investigation into the public houses of south and central Manchester.

It is at times a sobering thought to think that five years ago I had never heard of the disease that has so dramatically affected our lives.

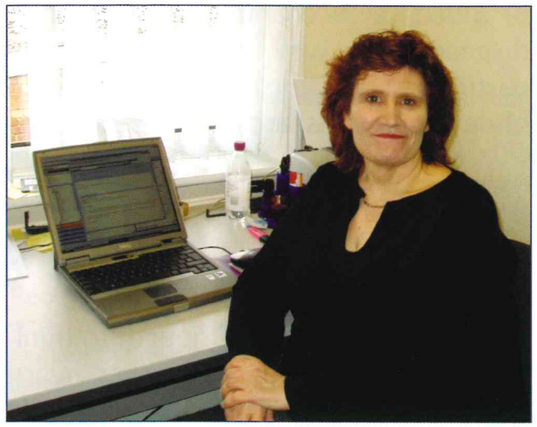
Isaac's MPS diagnosis has also led me to becoming a Trustee of the MPS Society; a job which is difficult to fit into the timetable of a young family but which I felt honoured to be invited to.

I hope that my experience and background add something to the Board of Trustees. If nothing else I think it is important that somebody with youth on their side is on the Board of Trustees (that should upset the rest of them).

Adam Turner and Isaac (MPS IH)

MPS Society Supports GOLD

Global Organisation for Lysosomal Diseases



Dr Ann Hale

The offices of the MPS Society in Amersham now have a new tenant! Dr Ann Hale is the Chief Executive of GOLD, the Global Organisation for Lysosomal Diseases.

GOLD is a new international collaborative organisation. Due to the rarity of MPS and other Lysosomal Storage Disorders numbering over 40 individual diseases, worldwide efforts are critical to providing better understanding of the global incidence, prevalence, and natural history of the diseases.

Ann says 'I am delighted that the MPS Society is providing so much practical help and support to GOLD. Their advocacy work and their publications provide model programmes for patient education and support which is of great help to developing patient organisations for a wider range of LSDs in other countries'.

The idea for GOLD began in 2002, when a group of highly respected basic and clinical researchers, patient advocates and pharmaceutical company representatives, debated how collaboration could best improve diagnosis, treatment and care for all those affected by LSDs.

Christine Lavery, Chief Executive of the MPS Society is one of the founding members and played a critical role in developing GOLD as an organisation, along with John Hopwood (Australia), William Sly, Edwin Kolodny and Abbey Meyers (USA) and Michael Beck (Germany).

Christine said 'GOLD will be an umbrella organisation, acting worldwide for collaboration between patient organisation representatives, clinicians and researchers, and pharmaceutical companies'.

Worldwide efforts are critical to providing better understanding of the global incidence, prevalence, and natural history of the diseases.

26 patient organisations have already joined GOLD, amongst them several European and worldwide MPS societies, in addition to organisations providing support to patients with Battens Disease, Tay Sachs, Niemann Pick, Gauchers and others.

A website is being developed at www.goldinfo.org

GOLD has just achieved registration with the Charities Commission of England & Wales, (Charity Registration No 1102478).

GOLD has the following aims:

- Understand the global incidence, prevalence & natural history of the diseases
- Promote basic research on each LSD
- Encourage the development of new therapies
- Provide accurate patient and physician education
- Increase funding for research, diagnosis, education and advocacy
- Speak with one voice to regulatory agencies and the public

Tribute to Dr Rosemary Stephens

Christine Lavery

Dr Rosemary Stephens came into our lives in September 1976 delivering to us most devastating news that our first-born son, Simon, had Hunter Disease. However through the fog that is diagnosis and bereavement rolled into one, Dr Stephens was to become our ray of sunshine. Over the years and long after Simon had died at the age of 7 years, in fact to this day, our whole family warmly regards Dr Stephens.

No-one, not even Dr Stephens, could take away Simon's disease however she gave us the confidence to continue with our plans for Robin to take up his post in the British Embassy in Tokyo some two months after the diagnosis, and for Simon and I to move with Robin to take up our lives in Japan. Whilst more conservative foreign office doctors deliberated long and hard, Dr Stephens was telling us to go and that the next four years could be Simon's best years of his life. How right she was.

Each year we used to bring Simon home to see Dr Stephens at Great Ormond Street Children's Hospital. She used to line up all the appointments and accompanied us to each over a period of just one or two days. Without Dr Stephens we may never have gone on to have more children, let alone healthy ones. Between 1978 and 1986 she accompanied us to three pre-natal diagnoses, taking away the precious vials of amniotic fluid for safe delivery to Liz Young at the Institute of Child Health. Thankfully, none of our pregnancies were affected and we have two sons and a daughter.

In tribute to Dr Stephens, Lucy, our 17 year old daughter, has Rosemary as a middle name. I know so many families in the MPS Society who have such affection for a truly remarkable doctor who was ahead of her time in the way she communicated, valued the lives of those affected, and embraced the whole family of the child in her care.

Dr Rosemary Stephens

Tay Sachs research fellow and honorary consultant paediatrician Great Ormond Street 1969 – 89 (b Harrow on the Hill) 1924; q University College Hospital, London, 1948), suffered a second stroke and died on 29 November 2003.

After schooling at Wycombe Abbey, Rosemary Stephens graduated from University College Hospital, London. From a Welsh family with medical connections, she was committed to a career in paediatrics. She held junior appointments at Paddington Green Children's Hospital, the Hospital for Sick Children, Great Ormond Street, and Charing Cross Hospital, whence she was appointed consultant paediatrician to Northampton and Kettering District Hospitals in 1967 and consultant physician in infectious diseases at Harborough Road Hospital, Northampton.

In 1969 she was appointed Tay Sachs research fellow and honorary consultant paediatrician at the Hospital for Sick Children, Great Ormond Street, where she remained until retirement. There, Rosemary devoted herself to clinical research into heredo-metabolic neurological diseases of infancy and early childhood, notably Tay Sachs disease, ceroid lipofuscinosis, metachromatic leucodystrophy, and Sanfilippo Disease. Although primarily a research paediatrician, her major contribution was her unfailing support of afflicted families, whose many letters of appreciation speak of her encouragement and compassion, priceless elements of medical care.

This was especially important in the days when the short term prospect for any therapeutic intervention in these conditions was scant, prenatal diagnosis was experimental, and, for most conditions, not usually available. As honorary consultant geneticist to Queen Charlotte's hospital she was closely involved in the early studies of prenatal diagnosis of lysosomal diseases. In this she was a valued and integral member of a team comprising biochemists, histopathologists, geneticists, and neurologists.

Rosemary was also strongly supportive of the specialised respite care unit for children with progressive neurological diseases at Tadworth Court, the former country branch of the Hospital for Sick Children, with residential facilities for parents. This was a model forerunner of children's hospices.

Notwithstanding her Welsh patriotism, she also introduced the convivial culture and cuisine of Burns Night to the neurology department at Great Ormond Street, where she was a loyal and valued member of the team! After she retired she continued a lifelong interest in horticulture, growing her own vegetable produce until 1998 when the death of her sister from cancer affected her physical health adversely with a severe bout of shingles.

Until she had a first stroke in September 2002, she lived in the house her father brought during early childhood, and nursed her parents there until their deaths.

This obituary appeared in the British Medical Journal, March 2004; 328:587-e. Permission for this article to be reproduced here has been given by the BMJ Publishing Group who are the copyright owners. The authors are David Kind and John Wilson.

A Happy Move

Jennifer Sangster

How Do We Know Lisa is Happy?

Lisa is 33 years old and has Sanfilippo Disease. Lisa is now in residential care. Lisa's parents, Rosemary and Harry, wanted Lisa to be close so that they could be near at hand for any emergency and also visit her regularly. So, with those objectives in mind, they set out last year to relocate Lisa from her residential care home that was closing.

Lisa has very special care needs and everyone needed to be assured that the new home could fulfil these and also make Lisa's life as happy as possible. Rosemary and Harry were very happy when a home was found that was only 15 minutes away in Sittingbourne, Kent, called Newington Court and so a transfer package was set in place.

It was arranged that over a period of six weeks, on one day a week, a member of staff from the old home that Lisa new well would accompany her for an overnight visit, both in order to settle her down in her new surroundings and to train the staff in the new home about all of Lisa's needs. Rosemary and Harry were on hand to spend the day with Lisa in her new surroundings and the care worker stayed overnight to make sure that the new staff were confident in looking after Lisa overnight.

After six weeks when Rosemary and Harry felt confident that Lisa was happy in her new surroundings , she was finally moved into her new home at Newington Court. The first question that the staff asked of Rosemary was "How can we tell if Lisa is happy?" This they soon learnt was very easy as Lisa has lots of smiley faces and can let you know easily if she is not happy about something.

The home provides all fresh food cooked by a qualified chef, and this is a good sign that everything is ok with Lisa. If she is enjoying her food then everything is alright with the world!! The cleaning lady who cleans Lisa's room also said that Lisa has very characteristic facial expressions, and the other day Lisa gave her an 'old-fashioned look' when she was talking as if to say, "Thanks, but go away". So the care staff now know Lisa's likes and dislikes!

Lisa goes home to Rosemary and Harry every Friday afternoon for the weekend and goes back to the home on Sunday evening. Lisa loves going out, so now that Rosemary and Harry are getting some respite time to charge their batteries they really enjoy taking Lisa out for a trip to the coast, come rain or shine!! Rosemary and Harry visit Lisa every Wednesday afternoon in the home and can check for themselves that she is happy in her new surroundings. The home allows visits at any time, which also gives Rosemary and Harry confidence .

Lisa is housed in a small block, consisting of ten bedrooms, where the residents share the lounge. She seems to be quite a star with the other residents. Annie is very protective of Lisa and always gets help if Lisa needs it, and Peggy chats away to her. Now there is a new resident. Sarah, who is 92, thinks Lisa is wonderful and misses her when she is not well enough to get up and sit in the lounge.

Lisa also has her favourite 'soaps' to watch and loves Eastenders, Coronation Street and Emmerdale. As soon as the intro music is played she sits up and smiles and obviously enjoys watching the programmes. When Lisa cannot leave her room, Rosemary has taped some episodes so that Lisa can enjoy them on her video. Lisa's room is very pretty with en suite facilities, her own TV, video and radio and she has some of her own pictures on the wall.

Lisa has a dedicated carer called Carol from Tuesday to Friday for her personal needs and so is now confident with that person. Rosemary is still trying to get her into a Day Care Centre so that Lisa can take part in some activities and be stimulated, but she is on a waiting list and Lisa will have to wait for a vacancy. In the meantime Rosemary is negotiating with Lambeth Council to provide some one to one care for Lisa on a daily basis so that she can be stimulated and enjoy a full life. Lisa enjoys a massage once a week, and the visiting masseuse now takes on staff and relatives if she has time! Rosemary was able to speak first hand of how good the massage was as she took Lisa's appointment last week when Lisa wasn't feeling well!

The home has told Rosemary that they are intending to make the block that houses Lisa into one dedicated for younger people and to this end they have a new toy boy on the block only 44 years young! There is also the possibility in the future of the company building a dedicated 20-bedded home for younger disabled people, so there is lots to look forward to. But, as Lisa is happy where she is, it may not be easy to get her to move again.

Since the dreadful fire in a nursing home in Scotland last year, rules are such that all the residents' doors are shut at night. A few weeks ago Rosemary gave the carers a baby alarm so that they could carry it about at night and hear any changes in Lisa as she is unable to press the buzzer for assistance. Just a few days later Lisa had a seizure and the carers were able to go to her straight away, thanks to the baby alarm .

Finally, Rosemary and Harry feel so confident with Lisa's new home that they have booked a holiday in Spain to recharge their batteries. The home have assured them that if Lisa has to go into hospital, then her dedicated carer, Carol, will go with her and if necessary stay. Carol has even offered to be called out if she is off duty. You can have no better assurance than that. ■

Lisa (MPS III)

Members' Announcements

New Members

Tracey and Peter Conlin's son, Joshua Benjamin, has recently been diagnosed with Hurler Scheie Disease. The family live in the North East and they also have a daughter, Sarah, who is seven.

Margaret Smith and her son, Louis, have been diagnosed with Fabry Disease. Margaret is 53 and Louis is 16 . They live in the South West.

Jake and Geraldine's son, Archie, has recently been diagnosed with Hunter Disease. Archie is three years old. The family live in the Midlands area.

Births

Harry Brockie was born on 3 January 2004. He is a brother to Samantha (MPS IHS).

In Memory of Enzo

He was a little man
who had done no harm, to anyone.

He didn't deserve what happened to him,
but he was deprived of his freedom.

So he has decided to forget his illness
and stop fighting with life.

He has flown away up to heaven
to play with his friends the angels.

And he is happy today,
he is smiling all the time,
this little man who had done no harm,
to anyone

For Enzo...
... from a dear friend

In memory of Enzo Dorso (MPS IH) who died aged 5 years on 21 January 2004.

Introducing ... Colin Arrowsmith

Colin (MPS II)

Hi! My name is Colin Arrowsmith. I am a 24 year old man with Hunter's Disease. I was diagnosed at the age of 4. From the age of 18 months until about the age of 5 I had a number of hernia and grommet operations. When I was 8 I had an operation for carpel tunnel syndrome on my left hand and my left leg straightened, then 6 months later I had my right hand and leg done.

I went to the local primary school, then at the age of 11, I went to a school for children with physical disabilities. I kept well and active. We played five-a-side football and went to Keilder Water about twice a year for sailing trips.

When I was 14 I had an MRI scan which showed I had compression of the spinal cord. I had to go to Royal Manchester Children's Hospital for an operation on my spine but two weeks later I started having really bad headaches. It was being caused by debris left from my spinal operation getting into my spinal fluid and causing hydrocephalus. I had to go back to the hospital for a shunt to be inserted in my head and within a few days I was feeling really well for the first time in six weeks. It took about five months to get my strength back that summer but I was able to play football again and ride my bike.

When I was 16 I passed my driving test at the first attempt. I also took my GCSEs that year. I stayed at school until I was 18, then got a job at Northern Electric in the offices.

At that time I felt I had a lot of control over my life and independence.

From the age of 19 my health started to deteriate and I was unable to walk any more because of the pain in my hips. I am now in a wheelchair all the time but I still drive my car locally. I then had to leave work at the age of 21 because of ill health.

I started the ERT trial on 14 January this year at Royal Manchester Children's Hospital and we travel down from Newcastle once a week for an infusion. This is a double blind placebo trial so I will not know until next year if I am on drug or placebo.

I am a fanatical Newcastle United supporter and have had a season ticket for eleven years. My father and I go to as many away games as possible. We travelled to Milan last year and hope to go abroad again this year to see them play!

I hope to keep you updated on how I am doing on the ERT trial.

We hope that Colin will be able to contribute regularly to the Society's Newsletter. For more information on the MPS II ERT Trials check out pages 26 & 27.

Stop Press ...

Two families have shared their experiences of Enzyme Replacement Therapy and are having their story published in the June edition of Family Circle Magazine. Knowing the press, please don't hold us to this!

You may wish to glance at the May and July editions on the newsagent's shelf!

Viewpoint - The Need for Improved Public Facilities

Sally Summerton

My name is Sally Summerton. I am married to Tim and we have two children, William aged 8 and Sophie aged 6. Sophie was diagnosed three years ago with Sanfilippo and how our lives have changed.

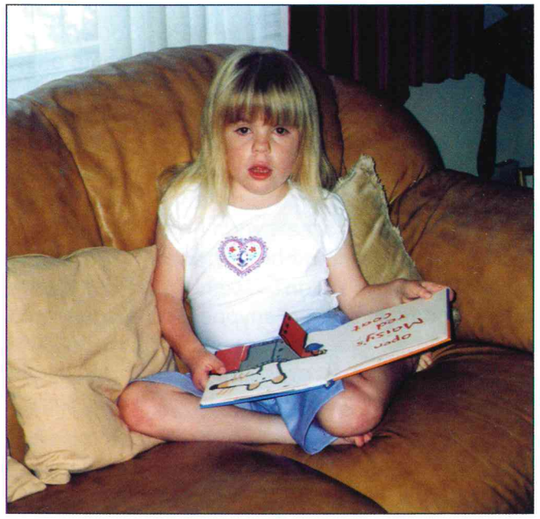
We always felt Sophie was 'different' as we had William to compare her development with, but what a shock the diagnosis gave us and all our family and friends. She is a lovely little girl, very affectionate and happy most of the time.

Since the diagnosis we have moved house to enable us to build a room for Sophie, argued with the local Education Department, struggled to get nappies, learned about respite, wheelchairs, hoists, medication and endless other things you never imagine you would have to know about.

We have good and bad days. Sometimes, when we take the children out, people are so kind and helpful and other times everyone seems to be intolerant, preoccupied or quite shocked by Sophie. She does make a lot of noise as she often sings and screams when we are out! She brings pleasure to some but embarrassment to others.

When you feel strong you cope well and ignore the problems as best you can, but other times you feel you would rather just stay in where it is safe.

I feel very strongly about the lack of public toilet facilities for children like Sophie. I am tired of dirty toilet floors and the effort of finding suitable places to change Sophie's nappy. I have been in touch with various disability groups and the CAB to see what might be done to improve the situation.



Sophie (MPS III)

The result of these enquiries has been the strong message that you need to get support to get anything considered. I realise that as a minority group you tend to be overlooked but I am sure facilities could be improved. I would be grateful to know if you feel the same way and would be prepared to offer your name to support a campaign to improve facilities.

Realistically, one toilet in every shopping centre would be desirable and must be possible. All that is needed is a fold-up bench about a foot off the floor which can hold the weight of larger children. Admittedly you would still be kneeling on the floor but at least the child would be slightly elevated. Also hopefully you would have a little more space to manoeuvre.

Please contact the Society if you would like to support Sally and put forward your views.

Can You Help?

Do you have a story or experience that you could share to help others deal with their own circumstances? Or, let us know if you have any questions that our readers may be able to answer.

To submit information for the newsletter please send text by e-mail and post original photos which we will return.

Members' Perspectives

Relocation Relocation

The Saga of the MPS II ERT Trials at Addenbrookes

Move over Channel 4, the MPS Society has a new line in relocating. And it has not come easy. Who would have thought that helping a few families relocate to the UK to take part in the Cambridge MPS II ERT trials would be so difficult?

1 The Snow Saga

Does anyone remember the snow? You would be forgiven if you didn't, seeing as it only really lasted a day. However, anyone who actually had to get anywhere on that particular day will probably have it forever etched on their minds as the day it took them 2 hours to travel half a mile.

Christine and I are unlikely to forget this day as we were supporting all the families attending the first day of the ERT trials at Addenbrookes in Cambridge including all those who need to relocate from another country. This suddenly became much harder when we discovered that Cambridge had in fact moved - to the Arctic!

While others were struggling to travel a few miles home from work, we were trying to get back to Bucks in a blizzard and, quite frankly, Arctic conditions. As we left the hospital to return to the car we were cold, and quickly turning into snowmen, but hopeful.

Two hours later. We hadn't got out of the hospital car park and we were less hopeful.

Three hours later and we still hadn't left Cambridge. At this point I have to say hope was fading rapidly and the prospect of a further 70 odd miles to go was not a pleasant one.

Five hours later having experienced the M11, A1(M) and all minor roads with traffic at a standstill, I have to be honest here and say that hope had now been replaced with a reluctant acceptance of our situation.

To add to our misery we had to endure countless reports on the radio explaining how well all the major roads have been gritted. Gritted my ****! The debris of abandoned cars on the side of the roads as well as first hand experiences of a terrifying lack of grip and driving speeds reduced to two miles an hour at times were testament to the fact that someone was lying. Far from looking like a motorway it seemed much more likely that we would see arctic animals wandering by on the hard shoulder. Instead of being overtaken by cars I occasionally mistook a large white van for a polar bear ambling past: he would have no need to rush - a geriatric polar bear with cramp and a side stitch could have easily passed us!

A short seven and a half hours later we made it home. This works out as an average speed of 10 miles an hour. On the plus side we fared better than some. A friend of mine had to travel 13 miles to get home after work. It took her 5 and a half hours - an average speed of 2 miles an hour. Gin and tonics all round I think!

2 Letting Agencies

Does anyone want a really good letting agency in Cambridge? Well, don't ask us - we don't know any!

They say that moving is one of the most stressful things a person can do. I can now vouch for the fact that it is also extremely stressful for everyone involved (however indirectly!).

Everyone in the MPS Society had their role. There were those of us on the front line, assisting the families and working face to face with the letting agencies; the calm but efficient General rallying the troops; and in our Control Centre was Sam who had to not only deal with the mess that came flying in at all angles but also had to mop it up afterwards.

We obviously cannot 'Name and Shame' the letting agencies involved but we can '*Names have been changed and Shame':

The Really Excellent Leasing Agency and The Amazingly Splendid Letting Agency have been helping us with the move of 3 families. Actually 'help' may be rather a strong word. I would say that they were just present and leave it at that, but that really doesn't do them justice. This would suggest that at least they didn't get in the way. No, these people were actually less than helpful.

Oh, it started off well - they would promise us the world - but what they actually delivered was slightly less than that: bedrooms with no beds, carpets with no hoovers, kitchens with no kitchen equipment and, possibly worst of all, no toaster!

One family were shown a lovely property in the middle of town. Well I say lovely... it was OK. I say OK... it was actually rather a dump, but the family loved it. Well I say loved it... you get the picture.

After haggling to get the place cleaned (one might have expected this to be included in the rental, but one would be wrong) the moving date then had to be changed several times so that the credit references and contracts could be completed.

OK, so it wasn't all their fault. The situation in all these cases was not aided by the fact that the contracts and tenancy agreements had to go to America to be signed before being returned to the UK leasing agents. This has made for a rather confusing situation. Not to mention long-winded.

Not one move went smoothly with at least one thing absent in each case. Some being less important than others, such as a TV, while some were slightly more fundamental, such as a bed! At one point Sam was calling the letting agencies so often she knew their telephone and fax numbers off by heart. A useful party trick I think you'll agree!

3 How to Furnish your Home for £789!

Another of our duties as pseudo-letting agency was to purchase equipment not already provided by the landlord. Strangely this was not as easy as you may think. In the absence of an appropriate inventory we embarked on a kind of guessing game as to what might actually be provided in the rented accommodation. Sometimes we guessed right. Sometimes we didn't. This was all part of the merry-go-round which was our relocation experience!

We suddenly became experts in bargain hunting and after regular trips to local bedding supply shops, numerous online shopping exercises, and desperate shopping trips for essential kitchen equipment which we had been promised would be supplied but at the last minute discovered that it actually wasn't, we now know everything there is to know about furnishing your home on a budget.

And then we became removal men. My favourite part of the job! Cars filled with bedding and cutlery and crockery making regular trips to Cambridge to assist the families on moving day. (I was also supposed to take a TV but couldn't fit it in my car. Well I'm sorry but when I signed up for this job I hadn't realised that I would need to be a removal man on the side so I got a thoroughly lovely but thoroughly impractical vehicle. D'oh!).

Thank goodness we had cars at all though. Could you imagine trying to do this using public transport? Having seen that it was too much for one car, we had a (rather amusing) mental picture of staff weighed down with boxes on the train: TV under one arm, bedding under the other, with possibly a pillow between the legs...

4 Some good things

Dresden Villa: No not a small holiday resort in this beautiful German city, but the guest house where some of our families stayed before we moved them. We must say a big thank you to the hosts Raf and Vincenzia who could not have done more to help. They gave above and beyond the call. Literally. On the odd occasions when Sam would call and ask them to run down the road and deliver a message to a family for whom we had no contact number - they actually did!

Sorrento Hotel: Quite apart from it being clean, smart, friendly, and close to the hospital, where else can you see palm trees in the snow?

Team building: This whole experience was an excellent opportunity for us to develop our team building and communication skills, as well as trying our hand at new things such as knowledge of international finances, and becoming fully conversant in all matters pertaining to lettings and removals. All of which will be incredibly useful in our jobs at the Society...

So why did we do it? Because Christine can't help herself and believed that the centres providing the treatment should be allowed to concentrate on the nursing and medical efforts and because she didn't want England and the MPS Society to seem inhospitable.

So would we do it again? Of course. The MPS Society wouldn't be the MPS Society without a bit of drama!

And finally... A tribute: To TKT Inc without whom we wouldn't have an MPS II clinical trial in the UK and the MPS Society wouldn't have been afforded the experience of Relocation Relocation. ■

Alison

Relocation Relocation

Sleep Disorders in Sanfilippo Syndrome: A Polygraphic Study

Editor's Note: Permission for this article to be re-produced here has been granted by the Italian MPS Society.

P. Mariotti, G. Della Marca, L. Iuvone, S. Vernacotola, R. Ricco, G.F. Mennuni & S. Mazza

Abstract

Objective: A high prevalence of sleep disorders is reported in patients affected by MPS III (Sanfilippo syndrome). These disorders have never been investigated by prolonged, objective, and instrumental evaluations. The present work is based on sleep duration and structure in Sanfilippo patients.

Study Design: The features of sleep/wake cycle in 6 Sanfilippo patients and 6 healthy controls were evaluated by means of sleep diaries and 48 hour ambulatory EEG and polygraphic recordings. Statistical analysis was performed by means of the U-test (Mann-Whitney).

Results: Four out of six Sanfilippo patients, the oldest patients in our sample, showed an extremely irregular sleep pattern, with several sleep episodes of inconstant duration, irregularly distributed along 24 hours. The two younger patients showed sleep maintenance insomnia with several nocturnal awakenings.

Conclusions: These results suggest that sleep disruption in Sanfilippo syndrome consists of an irregular sleep/wake pattern, which at its onset might appear as a disorder of initiating or maintaining sleep. This could explain why some patients do not respond to conventional hypnotics. The present observation might suggest attempting therapies aimed at resynchronization, such as behavioural treatment, light therapy or melatonin.

Introduction

Mentally retarded patients can show a particularly high prevalence of sleep disorders, ranging to about 33%.[1] Sleep disruption in these patients seems to be related to daytime behavioural and cognitive dysfunction.[1] Mucopolysaccharidoses are inherited lysosomal storage disorders, characterized by a deficiency of the specific enzymes involved in the degradation of mucopolysaccharides. These diseases are associated with mental retardation and frequently with sleep problems. The prevalence of sleep problems in mucopolysaccharidoses, according to Bax and Colvile,[2] as high as 71%, reaches values of 86-87% in mucopolysaccharidoses III (Sanfilippo syndrome).[2 3] Sleep problems can influence the clinical and therapeutic assistance of these patients. In particular, the patients' management and care appear to be extremely troublesome for their families.

Clinical experience suggests that sleep disturbances in Sanfilippo patients seem to be unresponsive to conventional pharmacological treatments such as sedative-hypnotic drugs. Techniques of behavioural intervention have been tested, with good results in some cases. Sleep disorders in Sanfilippo patients have seldom been investigated, mainly because the syndrome is rare (1 in 280,000 for Sanfilippo syndrome in an Irish study), and because of the difficulties in performing prolonged sleep recordings in mentally retarded patients. The most systematic study appears to be the one by Bax and Colville.[2] The authors report 62 patients affected by Sanfilippo syndrome, with a high prevalence of sleep disorders (87%), such as waking in the night, settling difficulties, reduced duration of night sleep, sometimes with all night wakefulness, and bizarre behaviours like laughing and singing and walking at night.

The studies available in literature have all been executed by means of parental questionnaires, which constitute a subjective and indirect method for analyzing sleep patterns. Moreover, most of these studies mainly focus on night time behaviour and sleep habits. Evidence from clinical observation suggests that Sanfilippo patients show an altered sleep/wake behaviour, which makes it necessary to evaluate a 24-hour sleep profile. For these reasons, we thought that prolonged polygraphic recordings made with EEG ambulatory devices could be useful for assessing and defining the sleep difficulties in Sanfilippo patients. In fact, an accurate clinical definition of sleep problems could also be useful in suggesting a specific treatment.

The aim of our work was to objectively measure the duration and the circadian distribution of sleep episodes in Sanfilippo patients, by prolonged (48 hours) sleep EEG and polygraphic recordings, in order: (1) to classify these patients' disturbances either as "dyssomnia" or as "irregular sleep/wake pattern," in accordance with the International Classification of Sleep Disorders; (2) to exclude other possible causes of sleep disruption, such as concurrent sleep-related respiratory disorders, by standard laboratory polysomnography; and (3) to investigate the sleep/wake cycle architecture, and the sleep phases organization in Sanfilippo patients, wherever possible (i.e., if the sleep EEG is not completely deteriorated by a neurological impairment).

Material & Methods

In our Institution 38 patients with mucopolysaccharidoses were followed. Among the 10 were affected by Sanfilippo syndrome type A all had relevant sleep problems, mainly consisting of frequent nocturnal awakenings and sleep schedule disruption. The families of three patients refused to participate in the study. A fourth subject was excluded because his previous polysomnography evidenced an obstructive sleep apnoea syndrome. Our samples consists of 6 inpatients (Table 1), 2 males and 4 females, aged from 7 to 20 years (mean age 14.1), all affected by Sanfilippo syndrome type A. The diagnosis of Sanfilippo syndrome was made on the basis of urinary tests and enzymatic assaying in cultured skin fibroblasts. None of our cases had visual impairment.

At the time of our observation the parents reported severe sleep disturbances unresponsive to conventional hypnotic drugs in all patients (benzodiazepines and neuroleptics had been administered in all cases). Informed consent was obtained from the subjects' parents. For each patient, a healthy control subject of the same age and sex was recruited from the Sleep Laboratory of the Catholic University. This kind of control group was chosen in order to compare the sleep habits of our patients with control subjects recorded under the same conditions and with the same methodology. All control subjects underwent an ambulatory EEG recording for two reasons: to rule out epilepsy in three cases, and to rule out parasomnias in the other three. The EEG recording excluded the presence of epilepsy and sleep disorders; subsequently, the control subjects underwent the same study protocol as the patients.

A summary of the main clinical data concerning our patient population is given in Table 1.

Table 1

Patients included in the study

Patient	Sex	Age	Diagnosis	Onset Age
PE	F	7	MPS III A	3 years
CA	M	9	MPS III A	8 years
PN	F	12	MPS III A	6 years
MS	F	17	MPS III A	4 years
MN	M	20	MPS III A	10 years
RR	F	20	MPS III A	6 years

Case 1

P.E., female, 7-years-old, the second child of non-consanguineous parents. At the age of 2, she presented with coarse facies, recurrent upper tract respiratory infections, speech delay and hyperactivity. Presently, she shows a marked neurological impairment: she doesn't walk and she has neurosensory deafness. Sleep disturbances started at the age of 3. They consisted of frequent night time awakenings (about 2 or 3 per night every night, lasting more than 30 minutes, often associated with whispering and tears). The parents also reported frequent early morning awakenings. Sleep latency was reported to be variable, but usually longer than 20 minutes.

Case 2

C.A.., male, 9-years-old, child of non-consanguineous parents. No remarkable events during neonatal period. At the age of 3, he developed coarse facies, hyperactivity, speech delay and frequent upper respiratory tract infections. At the time of our observation, he did not show hyperactivity, but presented mild neurosensory deafness, mild hepatomegaly, and no dystostosis. The patient could pronounce some phonemes and could walk if helped.

Sleep disturbances started at the age of 8. They consisted of frequent night time awakenings (about 3 per night every night according to the sleep diary, lasting 15 to 30 minutes). Sometimes awakenings were associated with abnormal behaviour, in particular laughing. His parents also reported frequent early morning awakenings, nightmares and nocturnal terror, and daytime sleepiness. Sleep latency was reported to be quite variable.

Case 3

P.N., female, 12-years-old, first child of non-consanguineous parents. At the age of 3, the quantitative analysis confirmed a diagnosis of Sanfilippo A. Since then her general condition worsened, particularly for neurological impairment. At 10 years she could not walk even if helped. Presently, her neurological conditions are severe. Sleep disturbances started at the age of 6, consisting of frequent night awakenings, enuresis, bruxism, early morning awakenings and daytime sleepiness. At the time of our observation she showed a serious insomnia characterized by difficulty in falling asleep (she took more than 30 minutes to start sleeping), frequent nightly noisy awakenings, early morning awakenings with daytime fatigue and sleepiness.

Case 4

M.S., female, 17-years-old. Neonatal period was unremarkable. She suffered from frequent upper respiratory tract infections. She presented with hyperactivity at the age of 3-4. The patient did not have coarse facies and skeletal abnormalities. She could walk if helped, and pronounced a few

Special Interest

phonemes. The patient started presenting sleep anomalies at the age of 4, and her parents reported completely disorganized sleep since age 8. During our observation she showed difficulty starting sleep (taking more than 1 hour), and 1 or 2 long nightly awakenings (lasting more than 30 minutes). Sleep duration was reported to be on the average 5 hours per night, associated with several daytime sleep episodes.

Case 5

N.M., male, 20-years-old. Neonatal development was normal, but it was characterized by frequent respiratory infections and otitides. He soon developed coarse facies, aggressive behaviour and hyperactivity. At age 9, he underwent a percutaneous gastrostomy as a consequence of failure to swallow and malnutrition. His neurological conditions have worsened since age 7, with severe hearing loss, seizures, and inability to walk. Presently he is forced to be bed ridden because of worsening neurological conditions. Severe insomnia started at the age of 10, with difficulty in falling asleep and frequent long awakenings and enuresis. During our observation the main problems appeared to be nocturnal awakenings (more than 3 per night every night), and daytime sleepiness. The parents' main complaint was the short night sleep duration.

Case 6

R.R., female, 20-years-old, first child of non-consanguineous parents. Her neonatal period was unremarkable. She did not show any dysmorphic features and began to exhibit great restlessness at age 3. She walked alone until the age of 13, and since then she has presented with frequent respiratory infections. The severe neurological impairment necessitated the application of nasogastric enteral nutrition at the age of 20. At age 6, she started to exhibit sleep disturbances consisting of insomnia with frequent night time awakenings. Her parents also reported a severe disorganization of sleep schedule since age12. At the time of our observation she showed difficulty starting sleep (she took more than 1 hour), 3 or 4 long nocturnal awakenings (lasting more than 30 minutes), and several short sleep episodes during the day.

Technique of Sleep Analysis

Each patient underwent a full-night polysomnographic recording attended by a technician, which started at 10:00 pm and lasted till 7:00 am, with EEG recording leads Fp1, Fp2, C3, C4, T3, T4, O1, O2, two EOGs channels, with leads positioned at the eye canthus and connected to A1, intercostal and submental surface EMG, airflow measured by thermocouple, thoracic and abdominal effort recorded by means of two strain gauges, peripheral hemoglobinic saturation monitored with finger probe, a pulsoximeter, and EKG (D2 modified derivation). Polysomnography was recorded in order to detect or to exclude obstructive sleep apneas, other sleep-related respiratory disturbances, or other known causes of sleep disturbances.

Subsequently, the patients underwent a 48-hour continuous polygraphic monitoring, by means of an ambulatory recorder (Oxford Medilog 9200). The montage was 6 EEG traces (Fp2-C4, C4-T4, T4-O2, Fp1-C3, C3-T3, T3-01), EOG and submental EMG. All recordings were performed in hospital, starting a 12:00 am and ending at 12:00 am 2 days later. During the recording days, all patients were hosted in singled bedrooms located within the unit of Pediatrics at the Catholic University. All subjects were allowed to sleep and to be awake according to their habitual schedule, and to keep all their normal habits. None of the subjects had sleep-inducing drugs or any medication that could interfere with sleep. The first 24 hours were considered an adaptation period, and therefore excluded from the analysis. The patient's parents were requested to fill in a sleep diary, starting 14 days prior to the recording.

A physician trained in sleep analysis scored the tracings in the group of patients and controls. The severity of the neurological impairment allowed the identification of the sleep stages only in 4 patients. In the remaining 2 patients, only the presence of waking or sleeping states could be distinguished, but no stages were identifiable. Data obtained from EEG tracings and sleep scoring were compared to those obtained from a control group composed of 6 healthy subjects of the same age and sex (Tables 2,3). Statistical analysis was performed by means of the U-test (Mann-Whitney).

Results

Useful EEG recordings were available in all the cases studied as well as in the control group. The sleep diaries showed that the sleep habits, during the recording day, appeared to be similar to those of the previous weeks. Polysomnography allowed the exclusion of disordered breathing in all of the patients in the sample.

In four patients, EEG tracings allowed identification of awake and sleep stages according to the criteria of Rechtschaffern and Kales, because of the persistence of phasic sleep events (vertex waves, spindles, K-complexes, REM saw-toothed waves). In these cases, the sleep architecture was characterized by a significant decrease of slow wave sleep (stage III and IV NREM, $p<0.01$) and REM sleep percentages ($p<0.05$). One patient (P.N.) showed a shortened REM latency (15 minutes).

In the other two patients, there was a severe impairment of the sleep EEG features. In these cases, the EEG could however, provide a clear definition of sleep and waking states. In fact, sleep states were defined by a diffuse slowing of EEG rhythms, associated with a decrease in muscle tone and disappearance of artifacts caused by body and eye movements.

The patients' group showed a global reduction of night time sleep duration ($p<0.05$) and a significant increase in daytime sleep duration ($p<0.05$). In one case (RR), only a few minutes of sleep state were detected in the 24 hours of analysis, thus representing a condition of almost total insomnia.

In four out of six cases (the oldest patients) sleep appeared to be extremely fragmented, and characterized by several sleep episodes, of inconstant duration, irregularly distributed during daytime and night time. No circadian rhythm of the sleep/wake cycle was observed. In the two youngest patients, sleep episodes occurred mainly during night time, but were characterized by several awakenings.

Discussion

Polygraphic recording as well a clinical observation suggest that all the patients in our sample presented sleep disorders. In all the cases the sleep problem was referred at its onset, essentially as a disorder in initiating and maintaining sleep, characterized by difficulty in falling asleep and by a high number of nocturnal awakenings often associated with abnormal behaviours. These findings are in accordance with those reported in the literature.

In the oldest patients in our sample (cases 3,4,5 and 6), the disorder appeared to be characterized by a progressive reduction of night sleep time and an increase in day sleep time with the appearance of several daytime sleep episodes. In one case (case 6) the polygraphic recording showed an almost total insomnia.

In four cases polygraphic recordings showed a preserved microstructural sleep pattern, with persistence of physiological sleep patterns (vertex waves, spindles and K-complexes in NREM sleep and saw-tooth waves in REM). Sleep architecture was characterized by reduced amounts of slow wave sleep and REM sleep. Two patients (cases 4 and 5) showed a monomorphic sleep pattern, without any physiological paroxysms. This finding had been previously reported in a Sanfilippo patient by Kriel et al, and might be related to the progression of the neurological impairment. In these latest patients, we could distinguish wake from sleep, but we were not able to classify any sleep stages. Polygraphic data confirmed the clinical observations.

In our sample of Sanfilippo patients, or at least in the oldest patients of our group, the sleep disorder might be better classified as an irregular sleep/wake pattern, rather than insomnia.[1] The reduction of total sleep and irregular sleep/wake pattern have been previously reported in other cases of severe mental retardation. Our hypothesis will necessitate confirmationby means of clinical-electrophysiological observation of larger samples of patients of different ages, as well as follow-up studies.

Classifying this sleep disorder as irregular sleep/wake pattern, at least in some older cases, explains why the hypno-inducing drugs are of scarce benefit of patients with Sanfilippo, as well as for other patients with irregular sleep/wake pattern. Our data can also suggest that therapies aimed at resynchronization should be taken into account in the treatment of sleep disorders in these patients. Resynchronization treatment for irregular sleep/wake pattern has been suggested by several authors, who proposed the behavioural therapy alone (Colville et al) or behavioural and light therapy combined, or melatonin treatment. ■

Table 2

Sleep duration in MPS III patients and sleep parameters in patients with still recognisable sleep stages

Patient	P.E.	C.A.	P.N.	M.S.	M.N.	R.R.	Mean	St.Dev.
Age	7	9	12	17	20	20	14.17	5.64
Total Sleep Time	430	501	347	443	210	45	329.33	172
Day Sleep Duration	58	46	116	198	103	12	88.83	65.64
Night Sleep Duration	372	455	231	245	107	33	240.5	157.67
Sleep Latency (min)	24	113	6	/	/	49	48	46.78
REM Latency (min)	107	161	15	/	/	/	94.33	73.82
Awakenings	5	17	12	/	/	3	9.25	6.45
Wake after sleep onset (%)	3.3	53.7	59.8	/	/	60	44.2	7.42
Stage I (%)	9.1	3.8	2.9	/	/	8.3	6.03	3.13
Stage II (%)	52.3	32.2	17.2	/	/	24.6	31.58	15.11
Stage III+ IV (%)	15.1	2.9	15.6	/	/	7.1	10.18	6.22
REM (%)	20.2	7.4	4.5	/	/	0	8.03	8.67

Sleep duration is expressed in minutes. Day from 7am to 9pm. Night from 9pm to 7am.

Table 3

Sleep parameters in controls

Controls	M.E.	G.M.	C.L.	E.D.F.	S.S.	S.L.	Mean	St.Dev.
Age	8	10	12	16	21	23	15.00	6.07
Total Sleep Time	490	569	451	502	420	467	483.16	51.09
Day Sleep Duration	87	0	0	62	0	0	24.83	39.28
Night Sleep Duration	403	569	451	440	420	467	458.33	58.73
Sleep Latency (min)	15	17	19	17	28	18	19.00	4.60
REM Latency (min)	90	85	88	52	95	100	85.00	17.02
Awakenings	2	4	8	5	8	11	6.33	3.27
Wake after sleep onset (%)	4.6	4.3	3.1	4.0	2.8	6.1	3.53	0.05
Stage I (%)	5.0	4.7	3.2	6.9	4.8	7.9	6.25	1.78
Stage II (%)	48.2	41.1	49.1	44.1	40.2	42.9	44.27	3.67
Stage III (%)	23.9	27.3	22.0	21.2	30.3	23.1	24.63	3.49
REM (%)	18.3	22.6	22.6	23.8	21.9	20.0	21.53	2.02

Advocacy Support Bulletin Board

OUT OF HOURS PHONE LINE
07712 653 258
7-9am Mon-Fri
5-10pm Mon-Fri
7am-10pm Saturday + Sunday

Regional Clinics and Events 2004

April

Wed 28	Cardiff Clinic
Thu 29	Bristol Clinic

May

Sat 1 - Sun 2	Weekend Get Together, Alton Towers
Thu 13	Scottish Clinic
tbc	Northern Ireland Clinic

June

Thu 10 - Sun 13	International Symposium on MPS and Related Diseases Mainz, Germany

July

Sun 11	Childhood Wood, Remembrance Day
Fri 16	Birmingham Clinic
Fri 16 - Sun 18	Sibling Activity Weekend

September

Fri 3 - Sun 5	Siblings Activity Weekend
Wed 29	Cardiff Clinic
Thu 30	Bristol Clinic

October

Fri 1	Jeans for Genes Day
Fri 22	Childhood Wood Tree Planting

November

Thu 18	Northern Ireland Clinic

JEANS FOR GENES APPEAL

Palliative Care in an Educational Setting

My Experience with Nathan, a Child with Sanfilippo Disease

Editor's Note: This article has been extracted with permission from a talk given by Jane Terry at the MPS Annual Conference in June 2003

I first saw Nathan at his mainstream school playing in a shared infant resource area. He was very restless and vocal, but enjoying the book being read to him by his Learning Support Assistant. Nathan was described as having communication difficulties, loss of skills, and challenging behaviour.

It was becoming increasingly difficult to give Nathan what he needed in a mainstream setting and a lot of time was spent in the SENCO's room. Nathan's parents had already visited our school and decided on an appropriate placement. He was transferred in September 1999 by which time I had already passed on as much information as I could about Sanfilippo Disease to the staff prior to his arrival.

During the early days of his placement we spent time finding activities that Nathan enjoyed. He coped with most sessions but only for a limited time and it was hard to sustain his interest. Nathan enjoyed his books and often joined in repeating lines of a story. He also enjoyed music and often sang snippets from current songs which were clearly his parents' favourites or TV adverts!

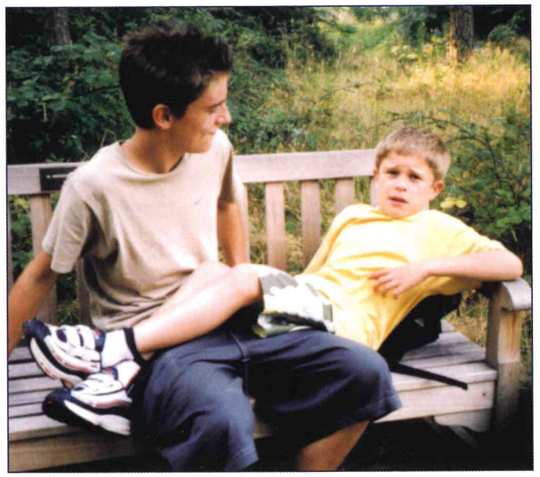
Helpful and supportive parents gave us as much information and help as they could and always kept us informed of any developments.

After half term a letter was received from the school doctor to say that Sanfilippo disease was confirmed. This came as an enormous shock to us. We could only begin to imagine how his parents must be feeling.

A class meeting was held and it was decided that, essentially, our approach would not alter. We owed it to Nathan and to his parents to continue trying to achieve our targets for him. What did alter was our thinking. We knew we could not change his behaviour.

We also knew there would be a loss of skills, but we had no idea of the timescale involved. As we got to know Nathan, and through regular meetings and observations, we were able to agree how best to meet his particular needs.

In the longer term, we would need to begin involving other specialists, such as music therapy. Informing staff and obtaining as much information as possible were also essential. Nathan's music therapy began in Spring 2000.



Nathan (MPS III) & his brother Stephen

To find out about Sanfilippo Disease we looked through published material which was very useful, but we still needed advice on how to respond effectively within school. We were put in touch with the Deputy Headteacher at a nearby school where another pupil with Sanfilippo Disease attended.

This was a huge help. My staff and I all regarded this as a turning point and it was in our meetings following this visit that we really began to understand that more than anything we needed to accept and be ready to respond to change.

After that we found we could take changes on board far more readily. We would record and discuss anything we noticed, consult the parents, and then adapt our approach and programmes accordingly, often guided by their response.

I then attended the MPS annual conference in 2000 with our music therapist. This helped in developing strategies and programmes further, including working with Nathan's parents via the telephone, meetings and a Home/School record book, speech and language therapy, occupational and music therapy, special programmes such as soft play, setting targets and individual education plans, managing Nathan's behaviour and concentrating on comprehensive 'whole school' approach.

To summarise, what helped us most in working with Nathan and his specific needs were working together, being flexible, getting advice and information, creating an appropriate environment and enjoying the moment.

The Role of the Speech and Language Therapist

Editor's Note: The information in this article has been taken from a variety of sources suggested by Rebecca Howarth, Senior Speech and Language Therapist at the Royal Manchester Children's Hospital who spoke at the Society's Annual Conference in 2003.

As well as being trained to assess verbal and non-verbal communication and speech and language development in individuals, Speech and Language Therapists can also provide input on developing feeding and drinking skills. This will include giving advice on positioning during feeding, which food types and textures are appropriate, the use of feeding utensils and facilitating communication during feeding.

Other medical factors also need to be carefully considered as well as the nutritional and safety aspects of the swallow, therefore the Speech and Language Therapist will work in liaison with a multi-disciplinary team.

Many families are concerned about the swallowing difficulties their children experience. This article has been written to give an overview of how a Speech and Language Therapist can help.

Swallowing Difficulties & MPS Diseases

Swallowing is a significant problem for a lot of individuals who have an MPS disease. In addition to the narrow airways, common in MPS disorders, an individual with physical disabilities will need more support with feeding and swallowing as their co-ordinated movement is affected. This will inevitably have a direct impact on their feeding skills.

Feeding

Feeding is a complex process which involves a combination of actions and a degree of co-ordination to ensure that breathing and swallowing do not occur at the wrong time.

In infancy three main processes are undertaken which include sucking, swallowing and breathing. These need to be carefully timed to ensure safe and efficient feeding and prevent the aspiration of food (where food enters the lungs) and the swallowing of air.

Normal Swallowing

There are three stages involved in the normal swallowing process including oral, pharyngeal (swallow) and oesophageal (food moving to the stomach). A breakdown can occur at any, or all three, of the stages.

Stages of Progression in MPS Diseases

Initially, the oral stage resulting in reduced chewing ability and longer mealtimes, then pharyngeal difficulties, displayed initially by inconsistent swallowing, borderline aspiration, and/or chronic aspiration, followed by increased pharyngeal difficulties resulting in recurrent aspiration of fluids and solids.

Clinical symptoms of a swallowing difficulty and aspiration include recurrent chest infections or history of frequent upper respiratory infections, regular spiking temperatures, choking or coughing during feeding and inability to manage oral secretions.

Assessment

Once a swallowing difficulty (dysphagia) has been identified the individual can be referred to a Speech and Language Therapist for an assessment of the problem. Speech and Language Therapists have specialist knowledge in the anatomy and physiology of the head and neck structures and can therefore assist with an assessment and the subsequent management of the feeding and swallowing difficulties.

The Speech and Language Therapist will assess the individual's feeding and swallowing skills, usually during a bedside examination. This is also likely to include liaison with nursing staff, parents and other professionals involved in the individual's care. The assessment may include an observation of the individual's behaviour while feeding, and changes in heart rate and SATS may also be monitored to give an all-round picture of the individual's ability to feed.

Videofluroscopy

This may also be used in an assessment of an apparent swallowing difficulty. The Videofluroscopic Swallow Study (VFSS) is a high-tech device which is often used to get a closer look at a person's swallowing reflexes. It gives a clear view of all stages of the swallow and can detect aspiration if it occurs. It can also identify the impact which different food textures and positioning strategies may have on the safety of the swallow. The examination lasts around 10-15 minutes and is simply an x-ray of how well the swallowing mechanism is working.

Cervical Auscultation

This sounds complicated but essentially refers to a method whereby the different acoustic sounds heard during respiration and swallowing can be determined. It can be very effective in accurately diagnosing dysphagia. Despite being a new technique in England it has been confirmed as effective by further videofluroscopy examinations.

Management

The Speech and Language Therapist will agree an appropriate treatment plan with the family designed to help manage the dysphagia. Factors considered in the management of a feeding disorder include modifying the diet, avoiding high-risk textures, slowing the rate of feeding, using thicker liquids: thickening powder rather than naturally thick drinks, reducing volume (aspiration may increase with amounts taken) and non-oral feeding such as a gastrostomy.

High-Risk Foods

These include stringy, fibrous textures such as pineapple, runner beans, celery, lettuce; vegetable and fruit skins including beans (broad, baked, soya, black-eye, peas, grapes); mixed consistency foods including cereals which do not blend with milk such as muesli, mince with thin gravy, soup with lumps; crunchy foods like toast, flaky pastry, dry biscuits, crisps; crumbly items such as bread crusts, pie crusts, crumble, dry biscuits; hard foods like boiled and chewy sweets and toffees, nuts and seeds; and husks including sweetcorn and granary bread.

If you have concerns about your child's swallowing ability ask your GP for a referral to a Speech and Language Therapist. They will be able to advise you how to best manage your child's feeding and will support you if a further study into their swallowing ability is needed. ■

Jeans for Genes Research Grant Update

Since 1996 the Society has been one of four partner charities in the Jeans for Genes Campaign. The money raised on J4G Day is restricted to funding scientific research into genetic diseases and Advocacy Support projects.

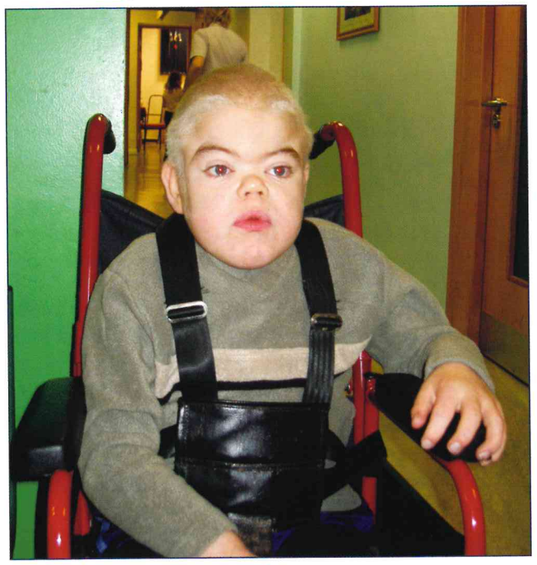
The Society is delighted to announce that from the proceeds of the 2003 J4G Day, it has awarded grants to three new research projects:

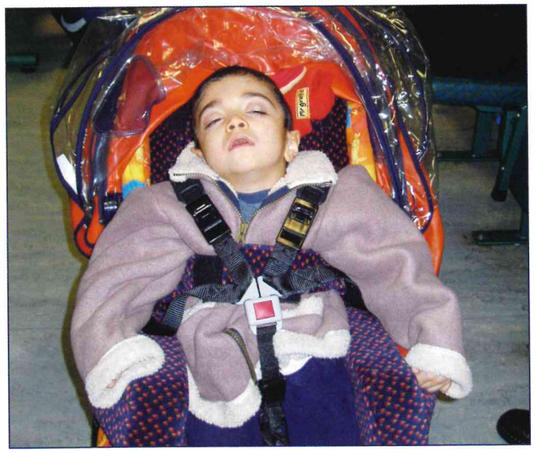
- a five-year lectureship in stem cell biology applied to inherited metabolic disease, principally the Mucopolysaccharidoses at the University of Manchester, Institute of Science and Technology - Dr Rob Wynn, Dr Ed Wraith
- an investigation to identify biomarkers for the Mucopolysaccharidoses Institute of Child Health, London - Prof Bryan Winchester, Dr Clare Beesley
- an 18-month paediatric secondment programme to gain further experience in the clinical management and treatment of MPS and Related Lysosomal Storage Diseases. Addenbrookes Hospital, Cambridge - Dr Uma Ramaswami .

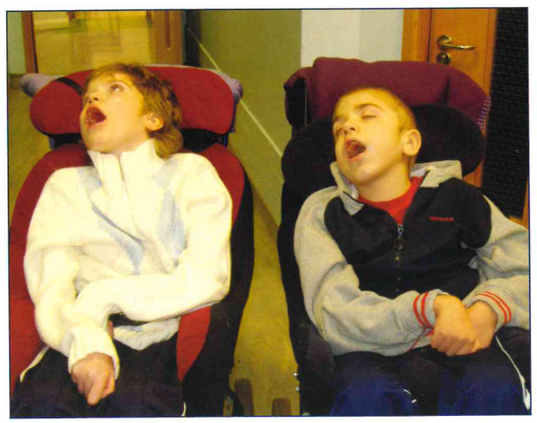
Two Advocacy Support projects, the Financial Assistance Scheme and two Advocacy Support workers, are to receive Jeans for Genes funding totalling £70,000.

MPS Regional Clinics Birmingham and Newcastle

Sam Vaughan



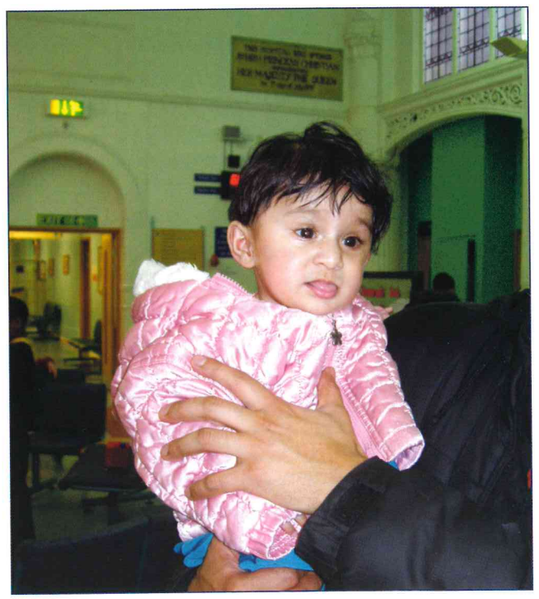




Another month, another clinic (or two)...

On a wet and miserable day in January, I managed to drag myself out of bed at 5.30am for a trip up to the North. It was Birmingham clinic day, and despite the early start and dodgy weather, the clinic ran incredible smoothly without any hitches, bar the wild goose chase Sophie and I had around Birmingham hospital, as different people pointed us in different locations to find the MPS clinic!

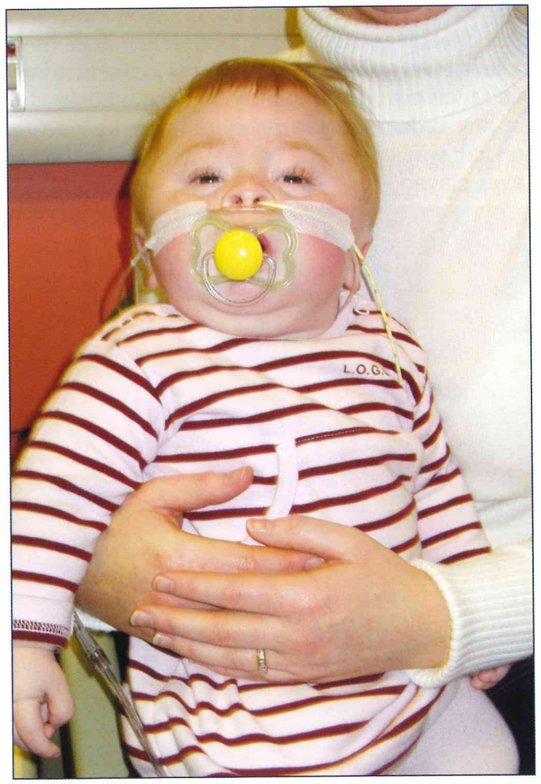
Thankfully, a few weeks later, the Newcastle clinic followed suit, and also ran like clockwork (minus the goose chase)!

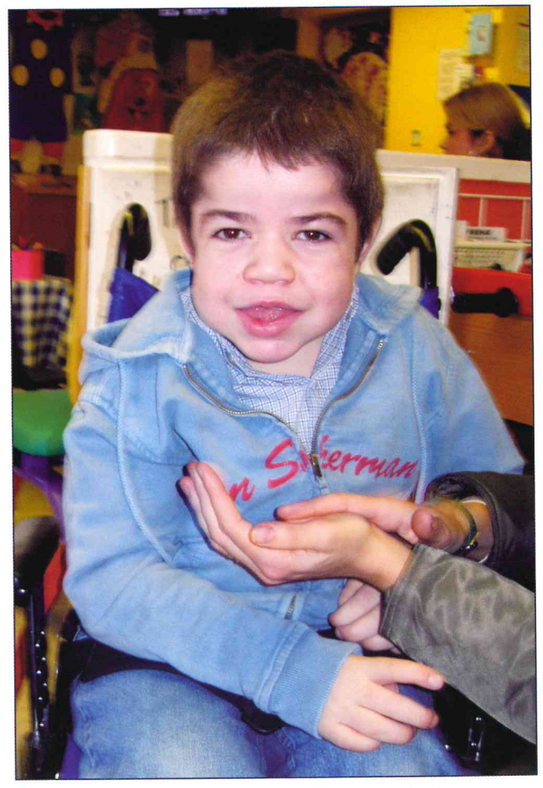


Birmingham MPS Clinic clockwise from top left: Ben Williams (MPS II), Sumaira Begum (MPS IV), Marium Hussain (MPS IV), Kim and Dwain Caines (MPS III), Pavan Tailor (MSD).

The nurses were very accommodating and we had our own little booth in the main waiting area complete with sales stall, TV and a football table, which was very popular with everyone visiting! Plus, we were plied with tea and biscuits all day which was great... well, not so great for the waistline, but it did keep us going through the clinic!

It was great to meet with everyone and put some more faces to names. And I must also extend my thanks to the doctors and nursing teams involved with both the clinics, as their continued support is invaluable and greatly appreciated.

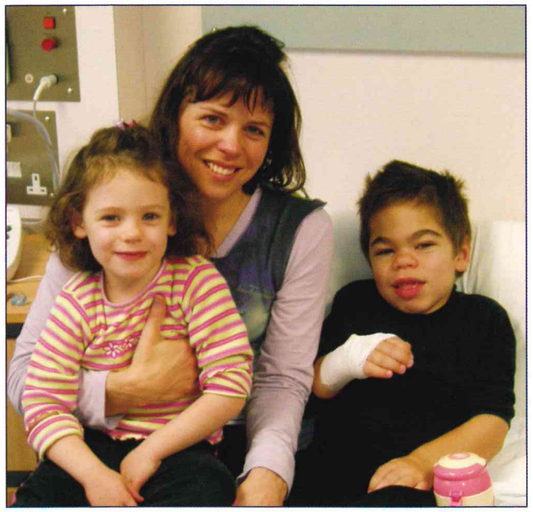


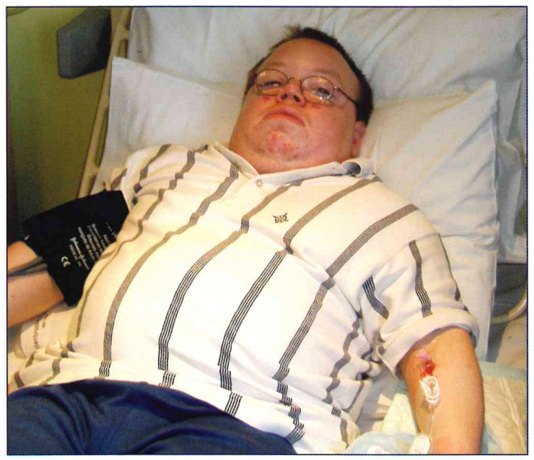


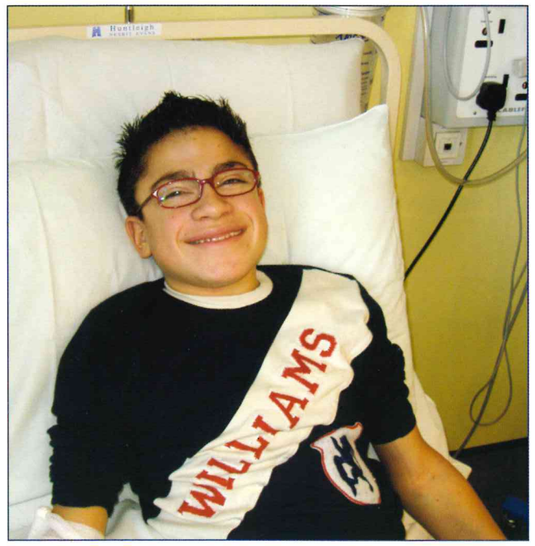
Birmingham MPS Clinic clockwise from top left: Fahim Hussain (MPS III); Newcastle MPS Clinic clockwise from top right: Callum McDonagh (MLIII), Daniel Muers (MPS II), Evie Mason (ML II).

MPS II ERT Clinical Trial

Sophie Denham







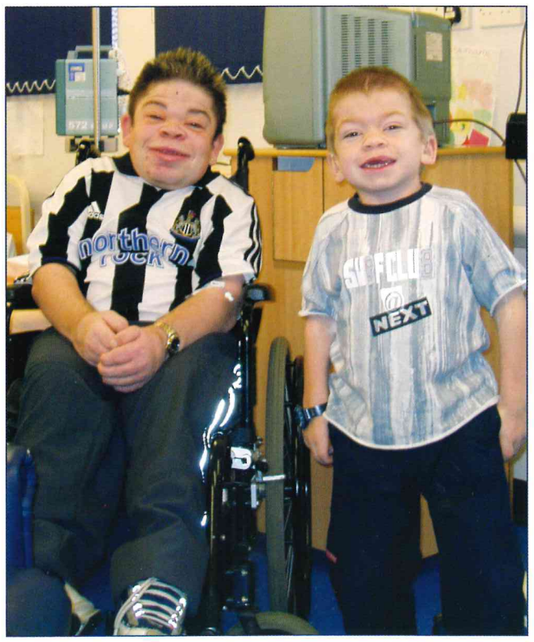
Week One - Baseline Study

(5 Jan 2004 - 11 Jan 2004)

I arrived at the Willink Unit bright and early on a Monday morning. I was there to support the start of the baseline study for the first MPS II Enzyme Replacement Therapy Trial.The Baseline study would take place throughout the coming week and individuals taking part would have to participate in various tests to see if they would be eligible to take part on the trial. Out of the ten individuals who took part in the Baseline study nine met the requirements to be put forward for the study.

In the two days that I was supporting the trial, most individuals and their families were able to find out if they had got a place on the trial. It was very difficult not to become involved in their worries and I found myself biting my nails and making numerous amounts of tea (tea being a comfort for any anxiety, so my nan used to tell me anyway!!) as we waited in anticipation to hear a 'Yes, you're in' or a 'Sorry, you didn't make it.'

All the families on the study stayed at a nearby hotel and it gave them a chance to talk and reflect away from the confounds of the hospital. A few glasses of wine and beer helped ease the nerves for the adults. For the children there was the chance to play on a playstation and have a few novel games of pool. Novel in the fact they had their own rules, numerous people could play in any order and the only cue stick had no tip on it! It was one of the best games of pool I have ever played!!!



Children, adults, parents and siblings on the MPS II ERT Clinical Trial

Week Two - 1st Infusion for MPS II Study

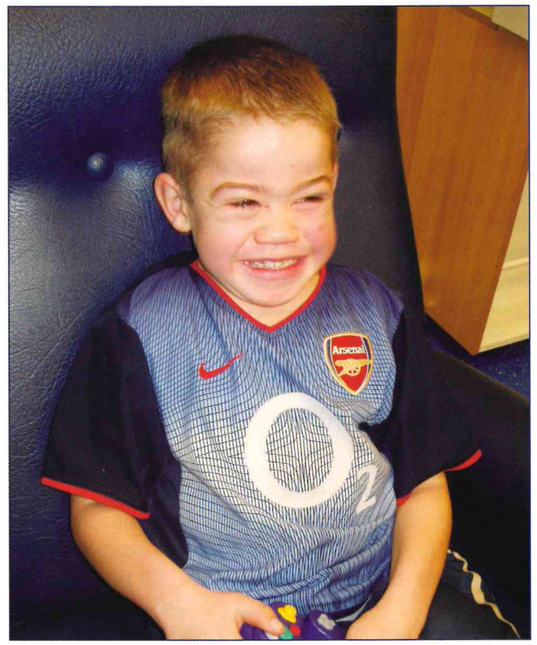
(First group 15 Jan, Second group 16 Jan)

The days had arrived for the first infusions to take place and I was again faced with a room full of people who were nervously trying to occupy themselves while they waited for the last checks to be made and to be linked up to the machines which would either administer enzyme or placebo. Once all the machines had been checked, double checked, and checked again, it was countdown and all the machines were switched on at the same time.

There were many sighs of relief that this day had finally come and I think every person either smiled like a cheshire cat or shed a few happy tears. The two infusion days were very different and the noise level on day one reached points higher than those reached at a real football match. In the confounds of the Willink, on two separate play stations we had two of the longest football matches in history being played. Manchester versus Newcastle and Liverpool versus Newcastle. It nearly came to the point where the dads (you know who you are!) were nearly shown the red card by the nurses for noise disruptions.

Day two was a very different story. There was no playing of the playstation until school homework was completed. Lessons did not, however, stop with the children. The nurses and myself learnt how to sign our names which I'm trying very hard not to forget.

The first infusion day took a total of six hours from start to finish to allow the nurses to monitor each individual closely. Subsequent infusions should, however, only take about four hours from start to finish which everyone was extremely happy about. The first infusion was probably the longest day most had experienced, but one definitely worth waiting for.



Children and nurses on the MPS II ERT Clinical Trial

MPS II ERT Clinical Trial

Information Days

Alison West

In the first month back after Christmas there was a sudden rush for knowledge at our local Children's Hospices. Within the space of a week I gave two information training sessions: one at Helen House in Oxford and the other at Naomi House in Wiltshire.

The sessions were informal and aimed at carers, nurses and all those involved in the care of children with an MPS disease who use these hospices for respite. The staff were able to ask any questions they felt were relevant throughout the sessions and the discussions which followed the presentations proved to be a very informative part of the training. This interactive set-up worked very well and, hopefully, everyone found something of interest to take away with them.

As the staff already work with some children who have an MPS disease I focused mainly on the conditions they knew, looking at ways of managing the ever-changing care needs of these children. We discussed some actual situations that had happened and I provided them with case studies and some more in-depth information aimed at helping staff to adequately meet these children's care needs.

The MPS Society runs these sessions at a variety of places including hospices, schools and hospitals. If you feel an Information Day or training session would be useful at your place of work please contact the MPS office.

Fabry Patient Meeting 2004

Rob Kenton

The third Fabry Patient Meeting took place this January in The Atrium at The Royal Free Hospital in North London. The origins of these meetings stems from the first get-together which was in reality a Christmas Party for the patients who took part in the early TKT trials. The organisers then were the staff at the Clinical Trial Centre at the Royal Free, and although they have now mostly moved on to new areas, the meetings continue to thrive under the current organisation of the Lysosomal Storage Disorders Unit.

The hard working staff on Crowley Ward where they are based, had brought together many patients and their families collated from the past four years or so, to meet and discuss various aspects of Fabry Disease with a number of interested parties. Most of the material was refreshingly new and the result of considerable progress over the past year.

After initial introductions and a welcome to the meeting we settled down to a highly technical and most interesting resume of recent work carried out by Dr. Derralyn Hughes. A small blood sample has been taken when patients visit the centre which is then analysed to find out how accumulation of the abnormal material, GB3, relates to problems with heart, kidney and other organs in Fabry patients.

It seems, from microscopy of these samples, that anomalies are appearing in the way these white blood cells (monocytes) are being affected, the changes being different from the way in which cells with normal enzymes work, and even between the cells from Fabry males and females; the question is why is this happening? Most of it was simplified for us to understand but it left me feeling impressed at the amazing complexity of just one cell and the work that it has to carry out.

Professor Bryan Winchester then gave a talk on the way in which Fabry disease is being monitored under ERT. This was particularly interesting for many of those present, as he has been working with the Fabrazyme drug and had results possibly not seen before by those patients using the Replagal alternative; the two drugs are produced by different companies who specified their own monitoring techniques during the initial trials and subsequently.

The most common result with the introduction of Fabrazyme appears to be an immediate reduction in lipid storage, followed by a period of stabilisation to near normal levels which looks encouraging.

One of the main problem areas for physicians dealing with Fabry Disease is the remarkable diversity between patients; level and type of organs affected, age and sex can all vary making accurate results difficult.

Before the lunch break Dr. Patrick Deegan had word of a new trial being presently discussed whereby patients currently receiving ERT would be asked to modify the amount and frequency of the drug being administered. In Fabry Disease, the lack of a particular enzyme caused by a genetic mistake is what leads to the build-up of GB3 within the organ cells. With ERT, these lipids are now being dealt with by the infused

Fabry Patient Meeting 2004 cont.

Rob Kenton

enzymes every fortnight but they soon lose their effect, so more frequent infusions are possibly the way to increase performance. Currently users of Replagal are infused with 0.2mg per kilo (weight), which would be doubled or infused weekly to try and find improvements in results.

Lunch brought the opportunity not only to satisfy one's stomach (and very nice it was too!), but being of the buffet variety allowed some chat with others and to catch up on news and progress of fellow patients, a sharing of experiences always proving beneficial and thought provoking.

Dr. Atul Mehta, resident Consultant Haemotologist at the Royal Free, recalled us to the meeting and gave us news of the latest results from the Fabry Outcome Survey (FOS). This collection of data is vast and very detailed; it has been compiled from questionnaires completed by Fabry patients from all the participating countries in Europe since the beginning of the trials. The results show signs of improvement for many patients and are encouraging.

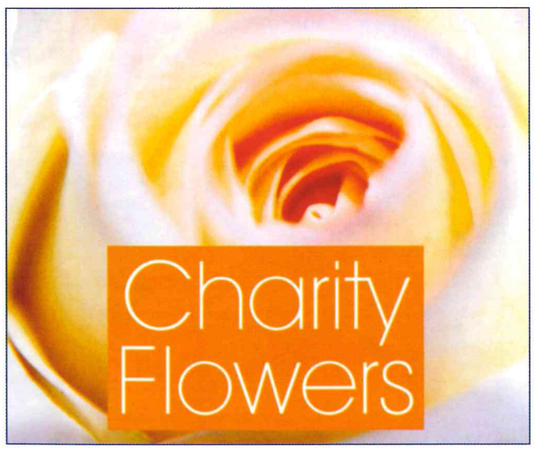
The penultimate speaker was the Society's very own Ellie Gunary, a name you will of course recognise, and the subject was the familiar one of funding for ERT. It seems that although both drugs have been licensed for some time now there are still a couple of areas where Primary Care Trusts (PCT's) are not willing to co-operate, and Ellie explained the procedures and methods the Society is using to help patients and families who are faced with this problem.

For our final speaker it was back to the medical books with complications to the Ear which can occur in Fabry patients. About 80% of us have these and Dr. Daniel Hajioff went into detail to explain why we suffer from these problems. It seems that ERT is helping for some but the problems which exist are so varied that again, results are not conclusive.

For the time remaining to us we were invited to ask questions to the whole panel of speakers now chaired by Professor Tim Cox and joined by several other physicians working closely with Fabry Disease, who attempted to answer a number of problems presented by the assembly. Each question was answered by whoever was deemed to be appropriate by a method of 'pass the microphone', until the satisfactory outcome was reached.

Looking at the row of speakers, I felt quite proud that this tiny malfunction of gene my mother had passed on to me could create such interest and enthusiasm among these people; and fortunate that I am living in an age where we have the knowledge and ability to do something about it.

From myself, and I'm sure I can speak on behalf of all the fellow Fabry patients and families, thanks to Alan, Sian and Linda and all those who organised and contributed to make the day a success. ■



Sale!

	Adult	Children
Sweatshirts	£7	£4.50
Poloshirts	£6	£4

Limited stock, assorted colours
Contact Gina for availability

Charity Flowers

15% of the retail price of each order goes to Charity. Order your flowers through Charity Flowers, quote MPS at the time of ordering and they will make a donation to the MPS Society.

A Holiday in India

Ellie Gunary

A promise I made to myself as 2003 (yes 2003) dawned was that this was going to be my year to begin travelling on holiday and visit new countries.

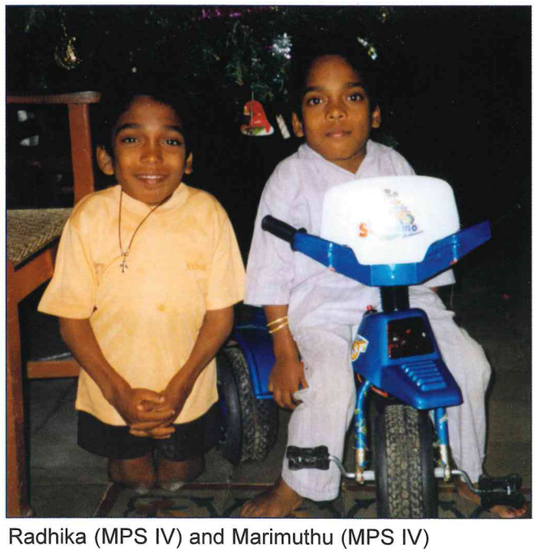
Growing up my parents had always encouraged me to write to children in other countries including the children they sponsored in Africa. Learning of these children's lives and following closely the politics of working in developing countries I grew up with ambitions of living in and learning more about developing countries, other cultures and ways of life.

Life, however, takes many different turns and as things have turned out my life choices since have placed me well and truly in Great Britain.

An invitation to spend the new year of 2003/2004 in India with a friend's family was therefore a dream come true. Whilst on holiday in Chennai I took some time out to visit two children. Radhika and Marimuthu who had Morquio Disease.

Elizabeth Herridge who accompanied her husband, the British High Commissioner for a four-year posting to Chennai (Madras) in Southern India had written to the MPS Society in June 2003 about three children, Manikandan, Marimuthu and Radhika who she had become friends with.

Elizabeth first met the three children in a centre, which offers orthopaedic surgery, physiotherapy, occupational therapy, speech therapy, education, residential facilities, vocational training and



Radhika (MPS IV) and Marimuthu (MPS IV)

Radhika, Dr Ramaswami, Marimuthu & staff member

rehabilitation. I also visited this centre whilst in Chennai and spoke with the staff there who remember all three children very well.

Elizabeth wrote of her first meeting with Manikandan at this centre. 'He was new to the hostel, sat in the corner and everyone fell over him or trod on him… I was in the process of bringing in wheelchairs from England and I could see immediately that he would benefit from one. 'When the wheelchair 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 years of his life inside a palm hut on a local beach, which was his home.'

'But… then I saw a movie about a young man with Morquio Disease and I knew that Manikandan had the same disease. I searched out a geneticist and it was confirmed. Unfortunately it was too late for any treatment. Later Manikandan was moved to the Cheshire Home in Chennai and he is now attending a school for children with disabilities.'

The following year Elizabeth met a further two children, a brother and sister, Marimuthu and Radhika at the same centre who had similar symptoms to Manikandan. The member of care staff on duty when I visited explained that the three children know they have Morquio Disease but are not aware of the details of this.

Marimuthu's and Radhika's parents live in a small village two hundred miles away from Chennai where they are daily labourers. Marimuthu and Radhika were also confirmed as having Morquio Disease and now also live in the Cheshire home. All three children are funded to live there for the rest of their lives.

When I visited the children one evening, Manikandan was visiting a friend so unfortunately I did not meet him. I did however have the pleasure of being welcomed by the huge, warm smiles of Radhika and Marimuthu. All three children help each other out and pursue many activities the Cheshire Home provides.

Marimuthu can no longer walk but gets around on a tricycle funded by a UK charity. Radhika is the organiser and looks after the two boys keeping their clothes in order, however as it was stressed to me, all the children help each other out in their own ways.

The member of care staff on duty when I visited explained that the three children know they have Morquio Disease but are not aware of the details of this. It would not have been right for me to tell the children more and then not be around

Dr Ramaswami, Radhika, Marimuthu & Ellie Gunary

to follow up any questions they had. Instead I shared information with the staff at the Cheshire Home so they will be ready should any of the children in the future want to ask more questions.

My lasting memory of Radhika and Marimuthu will be their warm welcome to a stranger who turned up out of the blue from a country many miles away. ■

Sofia's Story

Editor's Note: This article is taken from an e-mail sent to the Society by Jorge Cruz Villalba who lives in Spain.

We were very glad to have met representatives from the UK MPS Society and would like to thank you very much for the information you sent us about the work of your organisations and the different MPS Societies throughout Europe

This is our story…

When Sofia was 2 and a half years old a paediatric examination revealed that her liver and spleen were larger than normal so, to find out the reason for this, Sofia underwent a series of tests. When she was 3 and a half, we were aware that her behaviour in school was abnormal, she did not integrate with the other children and she did not take part in class activities. She did her own thing. Soon after her fifth birthday Sofia was diagnosed with Sanfilippo Disease from a skin biopsy.

This news was very hard to bear. Not knowing what Sanfilippo Disease was, we started to search and search for information about the illness. It was very difficult to find out what to expect and we began to realise that there was not sufficient information on this Disease in Spain.

That was when we decided to do something about it. We got in touch with the UK MPS Society and then ADAC in Seville, attending various meetings about the Mucopolysaccharidoses and there we met a number of families in the same situation.

We are now trying to spread information and raise awareness about the disease via the media (television, radio, the press) and, for mutual support, making contact with associations round the world.

Sofia (MPS III)

BioMarin Demonstrates Normalization of Carbohydrate Storage In Brain Tissue using an MPS I Model. Data Warrants Further Study of Intrathecal Injection of Enzyme for Treating Neurological Symptoms of MPS I.

NOVATO, Calif., Sept. 4 / PR Newswire FirstCall / BioMarin Pharmaceutical Inc. (Nasdaq: BMRN; SWX) announced results from studies indicating that intrathecal injection of recombinant human alpha-L-iduronidase (rhIDU) can reduce carbohydrate storage in brain tissue in the canine model of MPS I (mucopolysaccaridosis I). Emil Kakkis, M.D., Ph. D., Senior Vice President of Business Operations at BioMarin, presented the data at the ninth International Congress of Inborn Errors of Metabolism, on September 3rd in Brisbane, Australia.

Results from the studies are summarised below:

Intrathecal injection (injections directly into cerebrospinal fluid) of rhIDU once-weekly for four weeks delivered high levels of enzyme to the brain leading to normalization of glycosaminoglycan (GAG) carbohydrate levels in all four MPS I dogs treated. Results were based on a quantitative measure of GAG carbohydrate levels in addition to histological analysis of stained sections of brain tissue in treated MPS I dogs and normal dogs.

Penetration and activity of rhIDU in canine brain tissue was dose-dependent as indicated by enzyme analysis of brain tissue from treated dogs.

Delivery of enzyme via intrathecal injections also penetrated the scarred and thickened meninges (outer members of the brain and spiral cord). In MPS I dogs resulting in substantial reductions in GAG storage. GAG storage in the meninges can lead to spinal cord compression, a debilitating symptom of MPS I. Injection of rhIDU into the cerebrospinal fluid in rats resulted in significant penetration and activity in brain tissue as well as uptake into neurons confirming results observed in the canine model. Dr. Kakkis commented 'This evidence suggests that administration of alpha-L-iduronidase directly into the cerebrospinal fluid delivers enough enzyme to normalize GAG accumulation in the brain and meninges of the canine model of MPS I. Currently, bone marrow transplantation is the only available treatment that possibly addresses the neurological components of MPS I, but it is associated with significant morbidity and mortality.'

BioMarin Brain Disease Research

BioMarin is evaluating two approaches to treat the neurological components of lysosomal storage diseases including direct injection of enzyme into the cerebrospinal fluid (intrathecal administration); and use of the company's proprietary NeuroTrans™ program that uses carrier molecules to actively transport enzymes across the blood-brain barrier.

NeuroTrans may also be applicable to a variety of other diseases in which the inability to deliver adequate doses of intravenously infused drugs to the brain limits efficacy. BioMarin develops and commercialises therapeutic enzyme products to treat serious, life-threatening diseases and other conditions.

This press release contains forward-looking statements about the business prospects of BioMarin Pharmaceutical Inc; including without limitation, statements about: current preclinical research related to alpha-L-iduronidase and NeuroTrans and expectations regarding further pre-clinical research in such programs. These forward-looking statements are predictions and involve risks and uncertainties such that actual results may differ materially from these statements.

These risks and uncertainties include, among others; actual results and timing of current and future preclinical trails: the results of possible future clinical trails related to alpha-L-iduronidase and NeuroTrans; the content and timing of decisions by the FDA, the European Commission, and other regulatory authorities; and those factors detailed in BioMarin's filings with the Securities and Exchange Commission, including, without limitation, the factors contained under the caption "Factors That May Effect Future Results" in BioMarin's 2002 Annual Report on Form 10-K and the factors contained in BioMarin's reports on Forms 10Q and 8K. Stockholders are urged not to place undue reliance on forward-looking statements, which speak only as of the date hereof. BioMarin is under no obligation, and expressly disclaims any obligation, to update or alter any forward-looking statement, whether as a results of new information, further events or otherwise.

BioMarin's press release and other company information are available online at:

http://www.BMRN.com.

Information on BioMarin's website is not incorporated by reference into this press release.

Genzyme and TKT Form Collaboration to Develop and Commercialize I2S for Hunter Syndrome in Japan. Companies Sign Global Legal Settlement

Cambridge., Mass., Oct.8 / PR Newswire FirstCall / Genzyme Corporation (Nasdaq: GENZ) and Transkaryotic Therapies, Inc. (Nasdaq; TKTX) today announced the companies have entered into an agreement under which Genzyme will develop and commercialise iduronate-2-sulfatase (I2S), TKT's enzyme replacement therapy for the treatment of Hunter Syndrome, in Japan and other Asia/Pacific territories and a broad litigation settlement agreement. TKT has retained all rights in North America, Europe and Latin America, and intends to commercialise I2S directly in those territories.

Under the terms of agreement, Genzyme will pay TKT approximately $3 million in upfront payments and other consideration. TKT also has the potential to receive up to an additional $8 million relating to certain regulatory and commercial milestones, primarily related to a regulatory submission and approval in Japan. TKT will manufacture the bulk drug substance for commercial sale in Genzyme's territories and will receive payments that will equal approximately one-third of net sales in those territories. In addition, Genzyme has options to obtain rights to certain other research programs being developed by TKT in the territories where Genzyme holds rights to I2S.

Henry A. Termeer, Chairman and Chief Executive Officer of Genzyme Corporation, stated 'We look forward to working with TKT to improve the lives of these desperately ill patients with Hunter Syndrome. While the two companies will continue to offer competing therapies for Fabry Disease, we will work collaboratively to address the very serious unmet medical needs for Hunter patients.'

Michael J. Astrue, President and Chief Executive Officer of TKT said 'We believe Genzyme's expertise in working with products of this profile in these territories will add significant value to our Hunter syndrome program.'

In addition, Genzyme and TKT have signed a global legal settlement involving an exchange of non-suits between the companies. As part of this exchange, Genzyme will withdraw from the patent suit brought against TKT in July 2000 involving Replagal™ (agalsidase alfa), TKT's enzyme replacement therapy for the treatment of Fabry Disease. TKT has agreed not to initiate any patent litigation relating to Aldurazyme® (laronidase), Genzyme's enzyme replacement therapy for the treatment of MPS I, which is being commercialised in a joint venture between Genzyme and BioMarin Pharmaceutical Inc. However, in both situations, licensors of the patents at issue to Genzyme and TKT, respectively, could continue or commence legal actions despite Genzyme and TKT's legal settlement.

MPS II Clinical Trial

Transkaryotic Therapies Inc. (TKT) started their worldwide 12-month MPS II (AIM) pivotal clinical trial in January 2004. The AIM study is a randomised, double-blind, placebo-controlled, clinical trial to evaluate the safety and efficiency of iduronate-2-sulfatase (I2S) in 90 boys and young men with MPS II. These children and adults have been enrolled at nine sites worldwide. There are three infusion sites in the United Kingdom, the Royal Manchester Children's Hospital, Great Ormond Street Children's Hospital and Addenbrookes Hospital, Cambridge with22 MPS II sufferers enrolled. Nine of the twenty-two trial patients are from outside the UK. If the results are positive TKT expects to file applications for market approval in the United States and Europe in 2005.

TKT previously conducted a 6 month phase I/II randomised, double-blind, placebo-controlled clinical trial to evaluate the safety of I2S and its clinical activity in 12 children affectedby MPS II. Three doses were studied and within each dose group three MPS II sufferers were randomised to receive the I2S enzyme and one patient to receive placebo bi-weekly for six months. Results of the study showed that I2S was generally well tolerated and demonstrated evidence of clinical activity in several clinicial aspects of Hunter Disease, including respiratory function and functional capacity.

Further information can be found at the TKT Inc. website: www.tktx.com.

MPS IV A

TKT is beginning enzyme replacement therapy research for MPS IV-A to determine the impact on the bones. BioMarin Pharmaceutical Inc. currently is not actively involved in developing this therapy.

At this point there is no timeline for a human clinical trial. Studies in MPS VI and VII animal models suggest that, if given early, enzyme can potentially change the outcome of bone and cartilage disease.

Psychosocial Outcomes of BMT for Individuals Affected by MPS I

Research Update – Cheryl Pitt

Cheryl Pitt

The pilot study is now complete! Eight families very kindly took up our invitation to take part in the pilot study exploring the psychosocial outcomes of BMT for their children affected by MPS I Hurler Disease. We are very grateful to them for giving up their time to talk so candidly about their children and their families. At the end of 2003 the families were visited in their homes and interviewed about their experiences of living with a child with MPS I post-BMT and about their children's progress in life.

The information that the families generously provided has enhanced our understanding of the ways in which the children cope with and adjust to the different challenges in their lives. It has also showed us the great rewards and joy brought to families by their children, as well as the worries and concerns families have to deal with at different stages in their children's lives.

The aim of the pilot study was to get an overall picture of the life experiences of families of children with MPS I following bone marrow transplant, and to explore how the children are adjusting and developing socially and emotionally. It was important to explore these issues in an informal way, before embarking on a larger main study, as we had to make sure we were 'barking up the right trees' before inviting all those of you concerned to take part.

Thanks to the efforts of our pilot families, we now have a clear idea of the issues that are pertinent to MPS IH children and their families, and we are now ready to extend the study to include all families of children affected by MPS I Hurler disease post-BMT in the UK.

This main study is currently in development, and details of what will be involved will be sent out to you later this year. The method being proposed for the main study will differ to that employed in the pilot study. It will be more questionnaire based, and your children affected by MPS I will also be invited to take part. This is very important research and we do hope that you and your children will take the time to participate.

Call for Research Participants

Bone marrow transplant is no longer the experimental intervention it used to be, it is now a standard treatment for MPS I, and has given affected individuals the opportunity to live full lives.

However, as you know, BMT is not a cure for MPS I and brings with it some unique challenges for individuals and their families. Although our knowledge about health and disability outcomes is greatly improved, and families have a clearer idea of what to expect with regard to these issues, we still know very little about the psychosocial outcomes of BMT.

One question that remains unanswered is: What are the factors that contribute to healthy adjustment and quality of life post-BMT? We have a unique population of people growing into young adults affected by MPS I Hurler Disease, and young infants with the condition continue to be treated by bone marrow transplant. It is therefore essential that we explore the factors in their lives that determine their social and psychological well-being, so that these outcomes can be better understood, and in turn, enhanced. I am sure you will agree that as much as possible should be done to ensure these children's lives are as full and as happy as possible.

Your help in this research is therefore greatly needed. Although the study is still in development, you will be contacted during the summer of this year and invited to take part in the research. You will be sent details about your involvement should you decide to take part, and you'll be asked to volunteer. If you decide to take part you will be assured of confidentiality and anonymity.

If you would like to find out more about this research, please call Cheryl at the MPS offices.

The Computer Revolution!

Computers are one of the greatest modern inventions enabling us to communicate with others around the world and access a vast amount of information at amazing speed and with a minimum of effort.

For many individuals with a disability, access to computer has revolutionised their lives. Activities which most people take for granted, but which have presented enormous difficulties for children and adults with disabilities, can now be done using a computer.

Writing with a pen, or holding a book or newspaper to read, can be very difficult for many individuals affected by an MPS or Related Disease because of the physical manifestations of their disease. Painful, stiff joints, lax joints and weak wrists make it enormously difficult to grip writing materials, let alone be able to do this for a significant period of time.

However, computers don't always behave! Sometimes they have a mind of their own and do what they want to do, not what you tell them! Furthermore, for individuals with a disability they may present with other problems. Many of the more simple problems can be overcome without too much time or expense.

What if the text on the screen is too small to see? What about changing the settings in Windows to change the colour scheme or alter the size of the icons? Software is getting more sophisticated too with special tracker balls and keyboards designed to meet different needs.

There are many organisations out there which can provide advice, guidance and information on products and services that may be able to help you. Here are a selection:

Keytools

Keytools is a company that provides a range of computer equipment that promote communication, independence and develops existing skills. Much of their work involves matching the needs of individuals. You can contact Keytools on 023 8058 4314 or look at their website www.keytools.com.

Ability Net

Ability Net is a charity whose aim is to make the benefits of using computers available to disabled children and adults whether it be at home, in education or in employment. They provide a range of services including information and advice, assessment opportunities, equipment supply and support, professional awareness training and services to employers. For further information check out www.abilitynet.co.uk or phone 0800 269545.

The Aidis Trust

The Aidis Trust is a charity which provides computer equipment to individuals with physical disabilities for use in their own home. They supply equipment with a variety of adaptations to suit individual needs. If you would like to apply contact the Trust on 01202 695244 or check out their website www.aidis.org.

Can You Help?

Have you found a particular product or organisation that has been really useful or helpful? If so, do let us know. Perhaps other readers would benefit!

Means Test is Abolished for DFGs for Children

In December 2003 it was announced that the means test has been abolished for parents of children who apply in Northern Ireland for a Disabled Facilities Grant (DFG) for help with adapting their homes. This abolition is due to take effect from early 2004.

Whether the rest of the UK will follow suit is subject to debate. This issue may be covered in the Government's spending review which occurs in the Spring. The MPS Society understands that the government's review of the DFG may also include whether the grant, which is already discretionary in Scotland, should also remain mandatory in the rest of the country. Although abolishing the means test is a positive step, there are concerns about the 'doing away' of the mandatory grant.

The Society is working with other disability organisations to lobby the government on this issue and is asking its members to write to their MP.

If you would like to write to your MP requesting that the rest of the UK follows the example set in Northern Ireland by abolishing the means test, and also that the DFG remains a mandatory, not discretionary, grant we would love to hear from you. We can also help you compile the letter if you would like us to.

Contact the Society for further help or information.

Sam Wheeler (MPS IV)

Campaign Launch from the DWP

In Spring 2004 the Department for Work and Pensions is launching a campaign on the duties of businesses under the Disability Discrimination Act 1995.

This is part of a broader drive to improve general understanding over disability and the difficulties faced by people with disabilities.

For more information visit www.dwp.gov.uk.

The Perfect Armchair?

Mark Wheeler (father of Sam, MPS IV)

For any readers searching for the perfect chair, this is a photo of Sam in his new armchair.

The chair cost £40 plus VAT from Makro. It is an ideal size for Sam and looks much more grown up than many of the other children's chairs currently available so Sam is happy to use it. Perhaps it would suit other children like Sam too?

Do you Want to Take Part in a Video Project?

The Child Bereavement Network is undertaking a video project highlighting positive stories about coping with bereavement. They are looking for young people aged 13-19 to participate.

If you are interested, check out www.ncb.org.uk/news.

Update on Direct Payments

The Government has confirmed that disabled people can use their direct payments to pay close relatives, who do not live with them, for care services. In exceptional circumstances, direct payments may also be used to pay relatives who live with the recipients. More information on direct payments can be obtained from the Society.

Made to Measure Clothing

Sally Summerton (mother of Sophie, MPS III)

One of the problems encountered with Sophie was that she would play with and sometimes remove her nappy. This was often an unpleasant experience!

Through the Disability Living Foundation I found a small company that makes made to measure T-shirts and vests with poppers at the crotch, similar to the ones available for babies. This has eased the situation for us at bedtime and during the day as it prevents Sophie from getting to her nappy. Products are very good quality and are available in various colours.

The company is called Revolution – Underwear. Tel: 0116 277 5316 or E-mail office@revolution-underwear.freeserve.co.uk. They supply a brochure on request.

Signed England Football Shirt

Would you like to win this?

Glass-framed signed authenticated England football shirt donated by the Stewart family which was won from Sky Sports. The original bid winner fell through.

If so, please send in your bids with your name and address and return to the MPS Society by Friday 30 April 2004.

Please note there is a reserve price of £200 so the Society can obtain maximum funds for this item.

Donations

The Health Foundation
Mrs J Cottam
Mr John Scott
Helen Corcoran
Deborah Burniston
Mr & Mrs E Nelson
Mr & Mrs Whettem
Anne & Mick Palmer
Masterfoods, a division of Mars UK Ltd
Kate Gollop
Tilford Toddlers, Farnham
TKT Europe 5S AB
Gluckstein Charity Trust
Bristol Brunel Lions Club
Electrolux

Stamps and Foreign Coins

Mrs C Garthwaite, Jersey
Mr and Mrs Selvaranjan
Mrs Florence McConnell
West Wiltshire Primary Care Trust
(Adult & Community Services)
Molly Griggs
Nichola Gray
Sue Lowry
ICCS MacGregor

Fundraisers

The Boomerang Golf Society, Ely
Roxspur Measurement & Control Ltd
(In lieu of Xmas Cards)
Mr & Mrs Ingram, In lieu of Xmas Cards
Maria Murphy, Bristol (Half Marathon)
Global Direct(Xmas Raffle & Company Matching)
Marina Foster (Charity Shop in Bristol)
Northgate Information Solutions (Recycled Paper)
River Wear Social Club, Sunderland
Encore Direct
Washington Envelopes
Crawford Transport
PDQ Engineering
Platen Press
Dennis Jones Hackney Taxis
Roker View Private Hire Taxis
Princess Royal Barracks, Germany (Auction Night)
Sue Lowry (Coffee Morning)
Dunlop Aerospace (Who Wants to be a Millionaire?)

Collection Boxes

C.M.L. Jones & Partner, Dispensing Chemist, Swindon

Money in Memory

Jack Waddington
Gethin Robins
Thomas Birch
Enzo Dorso
Matthew Hodges

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Just Call Me Rock 'n' Roll!

Alison West

The Towersey Morris Men

Oh yes, I can live it up with the best of them! Wild parties, all night raves, an evening with the Towersey Morris Men...No, don't turn the page just yet. Hear me out.

Admittedly Morris dancing isn't everyone's cup of tea, and I *did* approach the evening with a fair degree of scepticism, but I have to say...

I had a great time!

The Towersey Morris Men (or TMM for short) have been regular supporters of the MPS Society for the last 13 years and in that time have raised over £6000 for us.

On this particular evening I was presented with a cheque for £500 and even got to participate in a couple of the dances. From the moment I arrived I was made to feel very welcome and was immediately invited to join in (anyone who knows me at all will believe me when I say that I was in my element!). Despite a slightly dodgy experience in which I momentarily lost my cool (note the blind panic on my face!) the evening was great fun.

As I said before I did approach the evening with some scepticism as I didn't really know much about Morris dancing, except for what I'd seen in an episode of Dad's Army. I wondered just what could make a grown man want to dance around in a strange white costume with bells on his legs, waving scarves and banging large wooden sticks with other similarly attired men. Oh, and facial hair appears to be obligatory too!

As I got to know the different members of the TMM it first became clear that this was just like any other club, where people from all different walks of life can get together with a shared interest and talk about things other than 'the daily grind'. Let's face it Morris dancing is about as far removed from the daily grind as you're going to get! There is also the opportunity for them all to share their enjoyment with the 'mere mortals' when they perform their dances at various pubs in the area throughout the summer season.

You may have noticed the word 'pub' in that sentence and it is on the strength of this that I am seriously considering becoming a Morris Groupie. Apparently the only qualifications you need for that are to appreciate the energy and skill of the dancing, reciprocate the fun and friendship and, most importantly, be able to hold your beer. Trust me, this I can do! You can even laugh at the ridiculous costumes - I actually think they rather like the attention!

To round off the evening it was down the pub for a swift half (!) of real ale, which I am assured is all part of the tradition. Who could ask for anything more?! So if you're looking for a more enjoyable way to spend a beautiful summer evening than moaning, as usual, that 'There's nothing on the telly!' might I suggest a trip to see the Morris Men? You never know, you might surprise yourself.

For details on their 2004 programme go to the TMM website @:

www.towersey-morris-men.fsnet.co.uk

Winter Warmer Festival

Jeff Bawden

The Caversham Charity Folk Festival has loyally supported the MPS Society for ten years. This year, for the first time, they had a Winter Warmer Festival, run across three pubs in Caversham.

All the musicians gave their time free of charge and the music was great from the Blues to the Skiffle Concert.

The MPS Society would like to thank everyone involved in the Festival and, in particular, Mike Tierney and Maureen Jerrett.

The Caversham Charity Folk Festival

River Wear Social Club

Members of the River Wear Social Club in Hendon raised a total of £1,950 on 16 October 2003 by holding a 70s night. This money was presented to Nichola Gray, whose daughter, Evie, suffers from ML II. The money has been given to the Society to continue funding for research to help Evie and others like her who suffer from this very rare condition.

Sgt. Jason Gray, Evie (ML II) and Nichola Gray

Nichola Gray and Michael Mason

Coffee Morning & Auction at the Princess Royal Barracks in Germany

Nichola Gray recently sent the Society a cheque for £1323.38. This money was raised by her brother, Sgnt Jason Gray and his colleagues at the Princess Royal Barracks in Germany. They work for the British Armed Forces and the money was raised by holding a coffee morning and an auction night.

'Who Wants to be a Millionaire?' at Dunlop Aerospace

During last year, members of the Sales Team of Dunlop Aerospace raised money through various activities. One of the main activities is a daily 'Who wants to be a Millionaire?', where staff contribute to answer questions. The money is then split, and the Society was one of the five chosen charities to receive £200 in memory of Matthew Hodges, whose sister Sarah contributed to the proceeds.

Raffle at the Robinson Library, Newcastle University

Sandra Fish is mother of Laura and Jessica who have Morquio Type B disease.

Sandra's hobby is cake decorating and as a way of fundraising to say thank you for the information and help she has received from the Society, Sandra made and decorated a Christmas cake to raffle at the Robinson Library, Newcastle University, where she currently works.

Sandra and her friends and colleagues raised a total of £91 through this raffle. We extend our thanks to everyone who bought a ticket!

Christmas Cake Raffle

Fundraising Can you help?

The Society for Mucopolysaccharide Diseases relies heavily upon voluntary donations in order to continue its high level of service to individual members and their families, raise awareness and promote research into MPS and Related Diseases.

We need to raise at least **£600,000** this year in order to continue our work. You can help us reach this target in a number of ways. Just check out the ideas below.

Through the government's gift aid scheme we can get more money from your donations at no extra cost to you! **Last year this scheme raised us an extra £4,200!** If you are a UK tax payer and complete a gift aid declaration the Society will receive an extra 28p per £1 donated. This also applies to sponsorship forms so encourage your sponsors to tick the necessary box.

Tobogging, Bungee jumping, Mountain climbing, Computer gaming... Darts night, Belly dancing, Swimming, Pub quiz, Dog walking, Bingo, Cycling, Cake eating, Fashion show, Goal scoring, Head shaving, Throwing pies, Leap

Organise and get sponsored for almost any type of event, such as a bowling tournament, raising money for MPS.

We can supply sponsor forms, posters, collection boxes and t-shirts. All you need to do is ensure your event is safe, and make it clear for whom you are raising the money.

These are just a few ideas of ways you can raise money for the Society. If you would like help or advice in organising an event, or for further information, just give us a call. We hope you can help...

mps@mpssociety.co.uk

☎ **01494 434156**

Don't stash your cash.
Let go of your dough.
Fundraise for us...
... just give it a go!

Plan an idea
(However funny!)
Anything goes
Just raise us some money

You don't have to take part
You can sponsor a mate
Help raise awareness
And help educate

Working as one
We might raise enough
Awareness and money
To make life less tough

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For more information
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