Newsletter

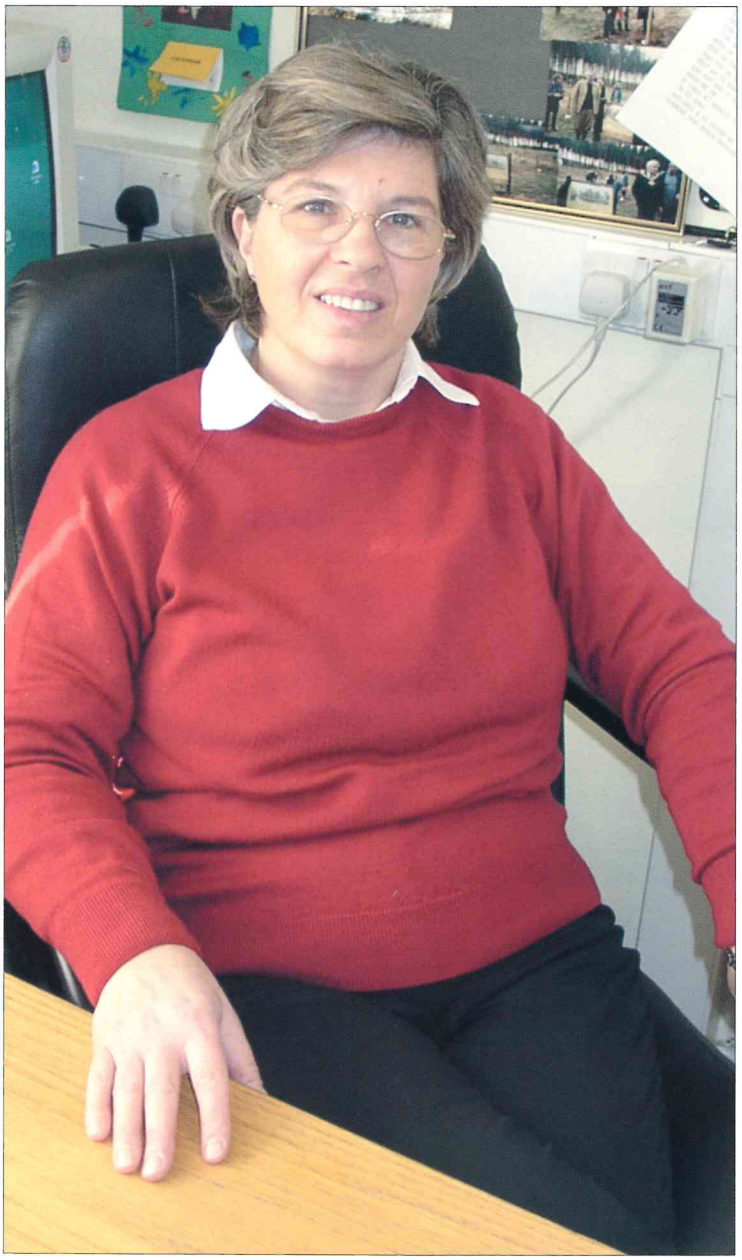
The Society for Mucopolysaccharide Diseases

National Registered Charity No. 287034

Winter 2001

MBE for Christine!

The Director of the MPS Society, Christine Lavery, has been awarded an MBE in recognition of services to the MPS Society in the Queen's New Year Honours.



Christine founded the charity nearly twenty years ago and has since worked tirelessly raising awareness of MPS diseases, supporting hundreds of affected families, as well as encouraging fundraising to promote vital scientific research. Dr Ed Wraith, who has worked closely with the Society for many years, said, "I doubt there has ever been a more deserved award", and her colleagues at the MPS office were obviously delighted with the news.

The award ceremony will take place at Buckingham Palace later in the year.

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'CARE TODAY, HOPE TOMORROW'

What is the Society for Mucopolysaccharide Diseases?

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

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

How does the MPS Society meet these Aims?

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

Telephone Helpline
Includes out of hours listening service

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

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

Regional Clinics, Information Days and Conferences
Information days and 11 regional MPS clinics throughout the UK

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

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

Information Resource
Publishes specialist disease booklets and other literature.

Quarterly Newsletter
Containing information on disease management, research and sent to members free of charge.

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

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

CHAIRMAN'S REPORT

The past twelve months have been both challenging and most rewarding and finished with the news of our Director, Christine Lavery becoming a Member of the British Empire, MBE, for her services to the Society for Mucopolysaccharide and related diseases in the Queen's New Year Honours List.

I know, that on behalf of every member of the Society, I can say congratulations and well done to Christine on the news of her becoming a Member of the British Empire, M.B.E. It has been awarded to her for her hard work, dedication and professionalism in the way she has developed a Society that really cares for and looks after its members. We all have a lot to thank Christine and her staff for and now the Queen will be doing it for us, in a most appropriate way. Congratulations Christine Lavery M.B.E.

The new board of Trustees got down to work soon after the Annual General Meeting and have met twice to set policy and agree the budgets and work plans for 2001/2002.

Support to those suffering from Mucopolysaccharide Diseases, their families and carers remains a prime focus of the Society's work. The Society's ability to provide a consistent level of advocacy to an increasing number of individuals and families has received a great boost recently when the Society succeeded in demonstrating the importance of this work and was awarded a grant of £30,000 per year for four years to fund a Development Officer.

I was very pleased to be involved in making the successful presentation to the PPP Healthcare Trust alongside, Christine Lavery and fellow Trustee, Wilma Robins.

Many of you have been following with interest the making of the MPS Video. I am pleased to say that this 48 minute video is now available from the MPS office. At the same time I am delighted to tell you of a brand new publication 'I've Got Morquio's' written and designed for children who have Morquio Disease, their brothers and sisters, and friends. However those who have reviewed the booklet suggest it is also a must for older siblings, parents and grandparents as well as professionals supporting children with Morquio Disease.

As you will read further on in this Newsletter Jeans for Genes has been a magnificent success this year with over £2.5 million already banked. The Society has already received a first payment enabling it to confirm its commitment to the second year of a 3 year project grant of £560,000 to the Institute of Child Health, London and the Willink Laboratory, Royal Manchester Children's Hospital. In January the Trustees will meet to consider new grant proposals for 2002.

Mindful that Christmas and the coming year may bring mixed blessings for those affected by any of the MPS or related diseases the staff and Trustees wish you all a peaceful New Year.

Barry Wilson
Chairman

Urgent Press Release January 3rd, 2002

TKT GRANTED ORPHAN DRUG STATUS FOR HUNTER DISEASE TREATMENT IN BOTH EUROPE AND THE UNITED STATES

Transkaryotic Therapies, inc. (NASDAQ: TXTX) today announced that the company's Niche Protein® product, Iduronate-2-Sulfatase (I2S), an enzyme replacement therapy for the treatment of Hunter disease, has been designated an orphan drug in both Europe and the Untited States.

www.tktx.com/news/c_press_HunterOrphan.htm

NEWS FROM THE MANAGEMENT COMMITTEE

The Trustees met in September and November to consider the following matters

Trustee Appointments
Newly elected Trustees, Adam Turner, Sue Peach, Judy Holroyd and Barry Wilson were congratulated on their appointments to the Management Committee. Barry Wilson was elected Chairman, Steve Butler and Judy Holroyd were elected Vice Chairpersons and Judith Evans elected Treasurer. Chris Holroyd was co-opted to the Society's finance sub committee. The Chairman expressed the Society's appreciation to Sarah Long and Angela Brown who retired as Trustees at the AGM.

Staff Changes
The Trustees agreed the post of Project and Information Officer with special responsibility for the organisation of the International Symposium on Mucopolysaccharide Diseases, 21-23 June 2002 to be held in Paris.

Policies
At the Society's Budget Meeting Trustees considered and agreed all the current policies. In line with recent legislation the Trustees also agreed the need for additional policy statements in areas of e.mail and internet usage, quantified risk management assessments and health and safety.

MPS Business Plan
The Trustees also reviewed the third year of the Society's Business Plan and agreed a work plan and budget for financial year beginning 1 November 2001. The Society will continue to prioritise its advocacy support to individuals affected by MPS and related diseases, their families and carers. The Society will also continue to provide an educational role through its publications, information days and conferences as well as support the development of new treatments and therapies. The Society will continue to fund innovative and vital research into Mucopolysaccharide and related diseases through the Jeans for Genes appeal.

Donations and Fundraising
The Trustees were delighted to learn that following a written application and subsequent presentation of the work of the Society by Barry Wilson, Wilma Robins and Christine Lavery the PPP Foundation has awarded the Society a grant of £120,000 over 4 years to help fund the Society's advocacy support programme.

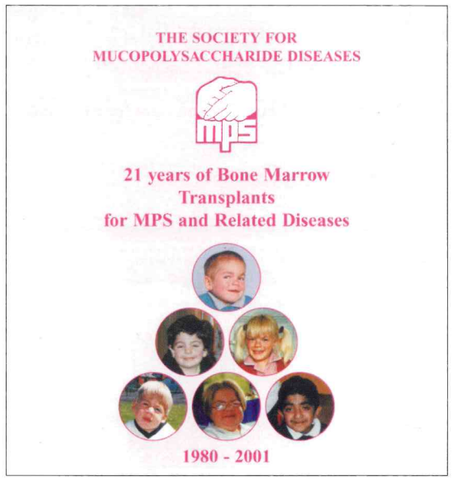
Jeans for Genes
Trustees received the excellent news that the October 2001 Jeans for Genes appeal had reached over £2 Million .

Report on 21 Years of Bone Marrow Transplants for MPS and Related Diseases

Available from 1st February 2002. Full report on the proceedings of the conference "21 Years of Bone Marrow Transplants for Mucopolysaccharide and Related Diseases".

This report includes the papers presented by each of the speakers as well as addressing questions asked by the audience.

UK	£10 inc. p&p
Europe	£12.50 inc. p&p
Overseas	£15 in. p&p



NEWS FROM THE MPS OFFICE

MPS Video

The long awaited "MPS Video" is now available. It was shown to families attending the Clinic in Northern Ireland and definiately got unanimous approval.

The video, through Dr Ed Wraith, Dr Ashok Vellodi, Professor Bryan Winchester and Dr Fiona Stewart, gives a detailed overview of the range of Mucopolysaccharide and related diseases including approaches to clinical management, treatments and future therapies.

Families living in Scotland, Northern Ireland, Wales and England share their experiences and help to depict the activities and support offered by the MPS Society.

Without the generous support of all these people and the BBC Breakfast news presenter, Jeremy Bowen, who kindly did the narrative, the making of the video would not have been possible.

For those wishing to purchase a copy of the video please find enclosed with the newsletter a flyer and order form.

Order your Video Now While Stocks Last

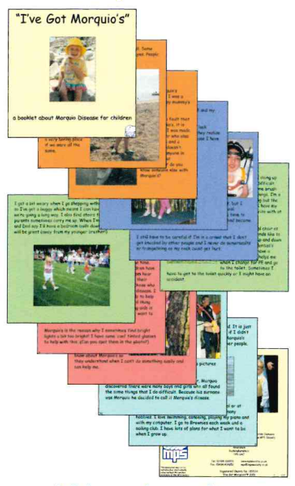
United Kingdom:
£5

Europe:
£7.50

Rest of the World:
£12

Postage and packing included in the price

New Morquio booklet for children



The MPS Society with the help of Claire Garthwaite, has produced the first in a new series of MPS booklets designed for children.

This colourful booklet, 'I've Got Morquio's' was written by Claire specifically for young people affected by the disease (and their siblings). It compliments the current Morquio booklet which is designed for adults and professionals.

The MPS Society is currently producing the 'I've Got Hunter's' booklet and will eventually try to cover the whole MPS disease range.

Fiona Stewart reviews the Society's latest publication, 'I've Got Morquio's'.

Dr Alex McGee and I have both read the booklet and we love it. It is clearly written in language that children could understand. We like the fact that it is colourful and has lots of pictures. Although it talks about the problems encountered by children with Morquio it is upbeat and should not limit their aspirations. We give it a big thumbs up!

Dr Fiona Stewart
Department of Medical Genetics

Fiona Woodcraft

Who's coming with us to Paris in June? If you are, it'll be me who you'll be contacting with all your difficult questions! I joined the MPS Society in mid November, with my main responsibility being the International Symposium on MPS and Related Diseases. I liaise closely with Association VML (Vaincre les Maladies Lysosomales) - our counterparts in Paris - as well as Christine and other members of staff here at MPS, and the job involves all aspects of the event's organisation. I've been lucky enough to go to Paris several times already, giving me the opportunity to familiarise myself with the conference venue and attractions on the social programme and to work face to face with staff at VML. There's so much to organise, but I enjoy a challenge! I do hope that you are looking forward to the event as much as I am.

Fiona Woodcraft
Project and Information Officer

PARIS 21st - 23rd JUNE 2002

7th International Symposium on MPS and Related Diseases

This conference promises to be a very interesting and enjoyable event and we want to help as many of our members to attend as possible. The programme includes internationally renowned scientists, clinicians and representatives from the pharmaceutical industry, enabling delegates to find out about the very latest clinical management, research and therapies. There will also be some exciting activities for the children, supervised by our own, English-speaking and trained volunteers, and the opportunity to meet other affected families from across the world.

Convinced of the importance of this conference, the Trustees have agreed that a **National MPS Conference** will not be held in September 2002, and that more funds will be devoted to helping individuals with MPS and their families to participate in the Paris conference instead. The Society is therefore able to offer fifty MPS individuals and their families the opportunity to go to Paris at the same cost as was charged last year for the Northampton conference. In addition, **travel bursaries** of up to £200 per family will be available which may, depending on the method of transport chosen and area of the country in which you live, cover the full cost of travelling to the conference.

The prices for registration and accommodation at the Professional and Family Symposium (Friday 21st to Sunday 23rd June) are as follows:-

	Full price	UK MPS price
1st Parent/carer/adult sufferer	£300 (480 €)	£150
2nd Parent/carer (sharing)	£100 (160 €)	£50
3rd Adult (sharing with other two)	£100 (160 €)	£100
Child 0-3	£20 (32 €)	£10
Child 4-12	£50 (80 €)	£25
Child 13-17	£55 (88 €)	£30
MPS sufferer 0-17	As for other children	Free

NOTE:
These subsidised prices apply exclusively to members of the UK MPS Society.
An adult sufferer of an MPS disease who participates in the conference programme can be accompanied by only *one* additional adult at the subsidised rate. An adult sufferer who participates fully in the children's activities will be charged as for a child aged 4-12.

The Society is able to guarantee full financial help to only the first 50 families so it is vital to **book as early as possible**. Included with this newsletter is the Final Announcement for the Symposium, giving further information about the event and registration forms. Please send your payment in pounds sterling by cheque or credit card to the MPS office. If you would like to request a travel bursary, please indicate this on the appropriate form.

We are continuing to pursue the option of a visit to **Disneyland Paris** prior to the conference. This will involve travelling to France on Wednesday 19th June, spending Thursday 20th in the park, and transferring to the Sofitel on the Friday morning. For those in the south-east of England, we hope to hire a coach with wheelchair access to take us to Paris, and we have researched group rates for travel by air from locations further north. We will probably stay in the 3 star Novotel outside Disneyland itself, as the hotels in the park have proved to be expensive. It is likely that the coach option will cost approximately £230 per adult and £180 per child (aged 3-11), to include 2 nights' bed and breakfast, travel from a couple of suitable pick-up points in the London/Kent area, and entrance to the park. Unfortunately we cannot give precise figures at the moment for prices excluding coach travel.

Please indicate on the form if you would like to extend your visit to France in this way, although the trip will take place only if there are sufficient numbers. Travel bursaries will still be applicable. You will be contacted with further details once we have an idea of bookings.

Fiona Woodcraft
Poject & Information Officer

FAMILY NEWS

Tara Murphy

Tara and sister Kate

Tara on Weymouth beach

Tara

Hello everyone, I am Tara Murphy's mum and my name is Maria. My husband is called Ivan and we have another little girl named Kate who is 6 years old.

Tara was diagnosed with Hurler on 22nd June 1998 aged 8 months. Probably like all MPS parents being told that first initial diagnosis was the most heartbreaking news we had ever been told. The next few days were spent in a complete haze with lots of tears and heartache. Then we were told that a bone marrow transplant may help Tara. Our older daughter Kate, aged 2fi at the time, was a perfect match and the BMT took place 5 weeks later, on 4th August - Tara was 9 months old.

Tara was absolutely remarkable throughout the chemotherapy and transplant and was allowed home on 28th August (their grandad's birthday) where she has continued to make great progress. Tara started nursery school at 2 years old and will start primary school next September with, hopefully, the help of a statutory assessment.

It still amazes us how well Tara has progressed and how well she has caught up with her classmates in this last year at Nursery. Obviously we still have constant hospital appointments together with continued check-ups with MPS doctors but we never forget how lucky we are to have two lovely, beautiful daughters Kate and Tara. I thought you would like a few photos especially Tara posing on Weymouth beach this summer - totally chilling out.

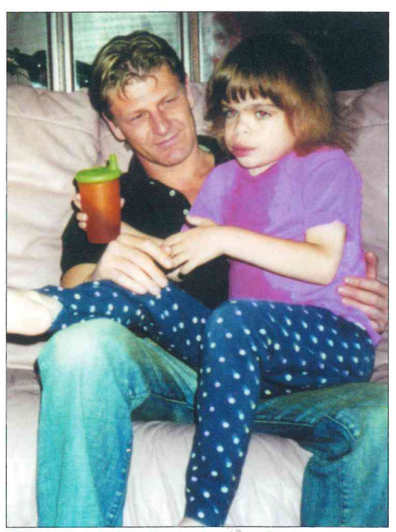
On 28th September we held a charity night in support of the MPS Society. We had a disco and Irish singer cabaret. It was an excellent night especially when our daughter Kate performed her Irish dancing in front of everyone. We raised £2060.

On 9th October I ran the Bristol Half Marathon. It was my first attempt at running a half marathon and to say I was nervous was an under statement. If that wasn't bad enough the weather was totally appaulling. It was torrential rain and gale force winds but determination and support (uterally dragging me along!) from 2 friends I managed to complete the course in two and a half hours and raised £350.

We would like to thank the MPS Society for all the help and support that you have given to our family.

Maria & Ivan Murphy

Lisa's Famous Footballer Grandad

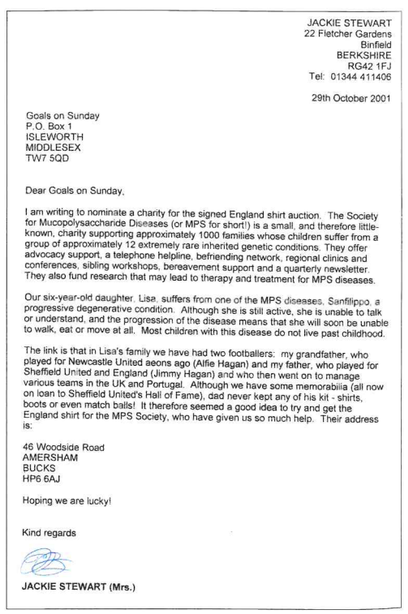


Lisa with Lord of the Rings star, Sean Bean

Lisa's mum, Jackie took part in a competition held by Sky TV, 'Goals on Sunday' to win an England football shirt signed by all the England football team. Amazingly Jackie's letter was chosen from thousands as the winner and the MPS Society is to receive this prestigious shirt to be used to raise funds.

If any one has any ideas on how to raise the most money or contact to help us, please do let MPS know.

Well done and thank you to Jackie.



JACKIE STEWART
22 Fletcher Gardens
Binfield
BERKSHIRE
RG42 1FJ
Tel: 01344 411406

29th October 2001

Goals on Sunday
P.O. Box 1
ISLEWORTH
MIDDLESEX
TW7 5QD

Dear Goals on Sunday,

I am writing to nominate a charity for the signed England shirt auction. The Society for Mucopolysaccharide Diseases (or MPS for short!) is a small, and therefore little-known, charity supporting approximately 1000 families whose children suffer from a group of approximately 12 extremely rare inherited genetic conditions. They offer advocacy support, a telephone helpline, befriending network, regional clinics and conferences, sibling workshops, bereavement support and a quarterly newsletter. They also fund research that may lead to therapy and treatment for MPS diseases.

Our six-year-old daughter, Lisa, suffers from one of the MPS diseases, Sanfilippo, a progressive degenerative condition. Although she is still active, she is unable to talk or understand, and the progression of the disease means that she will soon be unable to walk, eat or move at all. Most children with this disease do not live past childhood.

The link is that in Lisa's family we have had two footballers; my grandfather, who played for Newcastle United aeons ago (Alfie Hagan) and my father, who played for Sheffield United and England (Jimmy Hagan) and who then went on to manage various teams in the UK and Portugal. Although we have some memorabilia (all now on loan to Sheffield United's Hall of Fame), dad never kept any of his kit - shirts, boots or even match balls! It therefore seemed a good idea to try and get the England shirt for the MPS Society, who have given us so much help. Their address is:

46 Woodside Road
AMERSHAM
BUCKS
HP6 6AJ

Hoping we are lucky!

Kind regards

JACKIE STEWART (Mrs.)

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FAMILY NEWS

Joanne Evans' report on the Free2B Conference

Earlier this year I was invited to join the Kidz Board of the charity Whizzkidz, which provides mobility aids for disabled children in the UK. I have really enjoyed being involved with the charity and I attended the Free2B Conference held on 22nd October. This was Whizzkidz' first ever conference and was a great success. Over 80 disabled young people aged 11 to 18, their brothers, sisters, friends and parents from all areas of the UK attended at the Ocean, in Hackney, London -a truly wheelchair accessible venue!

The aim of the day was to highlight the negative attitudes faced by young disabled people and to stress the importance of having independent mobility. Whizzkidz had arranged a varied programme of speakers, question and discussion time and workshops during which all the delegates had the opportunity to express their views, concerns and questions.

The conference was chaired jointly by ITV broadcaster, Alastair Stewart and the Chairman of Whizzkidz Kidz Board. We started with some of the Kidz Board members and their friends and siblings who spoke about the way in which having independent mobility has changed their lives and relationships with their able-bodied and disabled friends and siblings.

Next was Ade Adepitan, whom many of you will know as a member of the Great Britain wheelchair basketball team, and TV presenter. Ade told us of his battle to overcome the barriers he had faced in establishing himself in his chosen sport. He inspired us all to persevere with whatever we want to achieve in our lives.

Andy Berry, a disibility rights campaigner from SCOPE, talked about the practical ways of overcoming people's preconceptions, and he was followed by actor and visABLE model, Luke Hamill, who spoke of the lack of representation of disabled people in the media - a subject close to my heart as I would like to pursue a career in that field.

After a break for lunch BBC Breakfast presenter, Sophie Raworth and another of the Kidz Board members chaired a series of workshops to discuss further the issues raised during the morning - this was a really lively session and everyone certainly had a lot to say on a wide variety of topics!

At this point in the proceedings, our parents, who had attended their own conference programme, joined us for the remainder of the day. They were in time to hear practical advice from Disability Rights Commissioner, Dr. Philippa Russell CBE, who explained how the new legislation in the form of the Disability Discrimination Act will affect us and hopefully ensure that disabled youngsters have the same opportunities as their able-bodied peers.

The final speaker of the day was truly inspirational actress, comedian and winner of the Daily Telegraph Open Mic Award in 2000, Francesca Martinez, who gave an amusing and informative talk on how she challenges negative attitudes by maintaining a positive attitude and enabling people to see past her disability. Again, many of you will remember Francesca from her starring role in Grange Hill.

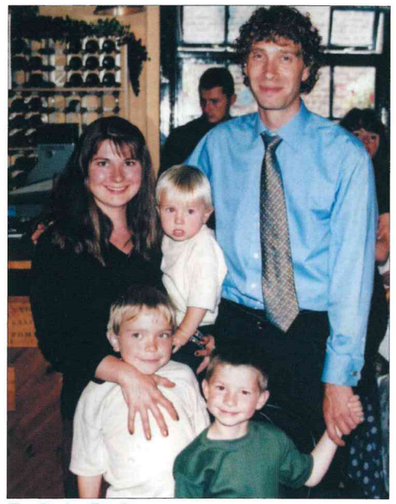
We all agreed that the conference had been a huge success and an inspiration for us to achieve great things in our lives.

Joanne Evans

Whizzkidz can be contacted on 020 7233 6600. www.Whizz-kidz.org.uk

FAMILY NEWS

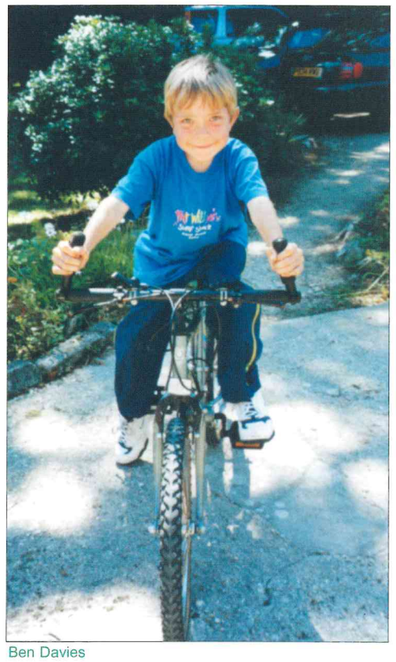
In a Land of Giants



The Davies Family

I may as well start as I mean to carry on, and without honesty I will be doing no one any favours. That stab of pain in the heart, that feeling of having your insides ripped out when you find out that your child has a disease that is incurable, with symptoms that can only get worse, that pain never goes away, or hasn't yet. Don't get me wrong, you may not find yourself curled up in a ball of pain, or plotting some cunning way of ending it all for your whole family three years down the line, but the pain is still there. You just deal with it differently.

I felt that my life had effectively come to an end. I had dreams for my family. We had high hopes. You know the ones, famous football players, rugby players, airline pilots, whatever. I just hadn't included a disabled, wheelchair bound son in the equation. I knew that I had been pretty naughty at times as a child, and even a young adult, but I was sorry. This was no way to make me understand. Yeah, I had got the picture, but this was serious. Someone else shouldn't be paying for my faults; that just wasn't fair. Or maybe it was. After all, seeing someone you love from the depths of your heart degenerate before your very eyes is pretty effective punishment. Or maybe there is no such thing as fairness. Fairness is a pretty human concept. It doesn't really make sense when you think about it.



Ben Davies

So, fairly screwed up, I faced the future. I faced the future with a large white plastic grin. I was looking ahead with my eyes tightly shut. This may not have been the perfect way of dealing with it but at least my head was pointing in the right direction.

I am tall; I had always been tall; I was tall at junior school. I had spent my life looking over other people's heads. I loved being tall. My feet were a long way from my head, and always had been. Tall people had an advantage, somehow. Ben would not be tall. He looked up to me and probably would forever. I just hoped that he would manage to look up to me metaphorically as well as physically for a long time to come.

I started to see small people everywhere. I started to see small people in town and on the television. I started to see people with all manner of disabilities. It isn't that I hadn't really seen them before, just that I hadn't really thought about them much. I had never thought about them much because their disability didn't really impact upon me. Now I couldn't think about them because disability did impact upon me and I wasn't sure how I was going to cope.

Still, facing the future with a brave face I knew that we would all have to cope. Ben, me, my wife, and Ben's younger brother Patrick. We would all help each other come through this. Come through this, what was I thinking? There will be no 'coming through this'. This is the way it will be. This is the way it will be until Ben dies. And by that time we will be gone ourselves. I had only just got used to having children and now I was thinking about the end of theirs and our lives. What sort of disease was this? I wasn't coping particularly well.

I made an effort to avoid contact with support groups. Support groups were for people that needed support and I certainly didn't. I was facing up to things and planning for the future. I was being sensible and logical. I was being sensible, logical and miserable. I didn't feel sorry for myself, nor did I once think 'why me?' so therefore I was coping. I flew a banner that stated that 'Ben's attitude to the disease would be a reflection of mine', so I made sure that my attitude was positive. On the outside I was positive. On the inside I was contorted with grief.

Ben grew. He didn't grow very quickly but he grew. He carried on walking. He didn't walk very quickly but he walked. He played football, swam, canoed and rode his bike. He talked, and my word he talked. I never expected so many questions. His wit and intelligence amazed me. His reaction to his, now obvious, set of disabilities made me burn with pride. I had never figured that Ben would appear to be facing his 'problem' so

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FAMILY NEWS

positively. I was also acutely aware that my grief was based on how I imagined Ben would feel about this disease, and in reality there was no way that I could foresee how he would feel. With Ben feeling positive we could all feel positive.

I plucked up courage and decided to attend the Conference. Yes, we had heard of the Conference and even seen the photographs, but had never wanted to go. I really didn't want Ben to see how things might turn out. I didn't want to see how things might turn out. I did, however, want to hear how research into the control of the disease was progressing.

We went.

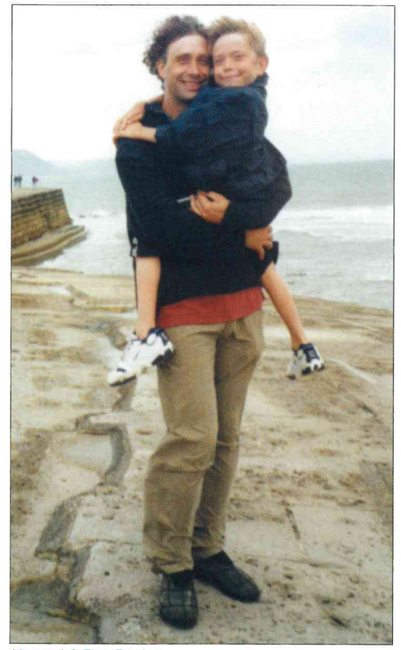
We met other people with Morquio's disease; we met people with all manner of MPS diseases. We met parents and carers. We met specialists. We met Dr. Tomatsu. In speaking to people we found support. I found support, and only then realised that we had always needed it. Not in any cathartic way, just to know that we weren't alone. And we weren't. We found hope. We found inspiration. We met people that were devoting their lives to the cure and management of MPS diseases.

I met short people and I met very short people. I wondered just how short a person could be. I talked to people with disabilities. I drank with people with disabilities. I realised that we all have disabilities. My disabilities may not be physical, but I sure have them. For me to ever assume that physical stature had any bearing on anything at all was ludicrous. How many times have I let preconceptions creep into my thoughts and actions? I will never know. I do know, however, that my inability to see the person beyond the body has been a disability. If I was tall I was surrounded by giants.

Ben has Morquio's disease. That's the way it is. He has a disease. A disease that at the moment is incurable. We are a family. We are not your usual family. One of our three boys has Morquio's disease. Ben knows that he has Morquio's disease. We will never know how he really feels about this disease deep down inside. He probably really hates it. I hate it. There is nothing that we can do about it so we mustn't let it eat us up. We can, however, learn to live with it. It is not always negative.

To quote from 'The Prophet' by Kahlil Gibran:

And a woman spoke, saying Tell us of Pain.
And he said:
Your pain is the breaking of the shell that encloses your understanding.
Even as the stone of the fruit must break, that its heart may stand in the sun, so must you know pain.
And could you keep your heart in wonder at the daily miracles of your life, your pain would not seem less wondrous than your joy;
And you would accept the seasons of your heart, even as you have always accepted the seasons that pass over your fields.



Howard & Ben Davies

We have all come to know Morquio's disease, but none more closely than Ben. He amazes me and I love him deeply. I will always look up to him.

Howard Davies

Ben with his brothers, William and Patrick

Ben Davies

BIRTHS, NEW FAMILIES AND IN REMEMBRANCE

Births

During the past 3 months the families of 12 affected children and adults with a Mucopolysaccharide Disease and 5 individuals affected with Fabry disease have sought the support of the MPS Society. The following families have requested their details to be included in the Newsletter.

Helen and Anthony Corcoran's son, Jake, has been diagnosed with Hurler Disease. Jake is 11 months old. The family live in Yorkshire.

Mr and Mrs Oakley's son, Nathan, has recently been diagnosed with Hurler disease. Nathan is nineteen months old. The family live in the Midlands.

Congratulations to Angela and Graham Ratcliffe on the arrival of Madison Tabitha on the 31st October 2001. Angela is currently on maternity leave from her post as Development Officer with the Society.

Congratulations to John and Jo Allen on the birth of their son Thomas born 20th October 2001. A brother for Jasmine and Bethany.

In Remembrance

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

Steven Grandidge who suffered from Sanfilippo Disease
15 February 1989 - 27 October 2001

Jamie Hicks who suffered from Hunter Disease
27 December 1985 - 3 November 2001

Elizabeth Murphy who suffered from Sanfilippo Disease
27 April 1984 - 5 November 2001

Shannon Mongan who suffered from Hurler Disease
27 August 1998 - 12 December 2001

Jade Robinson who suffered from Hurler Disease
24 May 1990 - 22 December 2001

Daniel Croghan who suffers from Hunter Disease
22 October 1986 - 29 December 2001

In Memory

Katie Louise Martin, went to sleep
January 8th 2001

The happy hours we all shared
How sweet their memory still
You've gone and left a vacant place
That we can never fill

There's a face that we remember
A smile that we recall
A memory we will always treasure
Because you were loved by us all

We miss you so much
Love you always

Mummy, Daddy, Lisa & Amie

MPS Newsletter Winter 2001

CHILDHOOD WOOD

Remembrance Day

On 26th October 2001 families arrived at the Clumber Park Hotel in Nottinghamshire to remember and celebrate the lives of seventeen children and adults who have lost their lives from an MPS or related disease.

The opportunity to plant a tree is always open to bereaved families but this year as part of the end of the Society's eighteenth birthday celebrations all bereaved families who had not planted a tree in the wood were specifically invited to do so. Whilst families shared memories over lunch in the company of the Lord Lieutenant of Nottinghamshire, Sir Andrew Buchanan, and Councillors Nellie Smedley and Maureen Tewson the rain came down outside. After lunch Sir Andrew Buchanan welcomed everyone and explained his role of representing the Queen at this and other events he attends within Nottinghamshire.

With trepidation everyone then prepared to face the elements outdoors. To say it was raining would be an understatement. Whilst walking through Sherwood pines we all hopefully looked for a break in the clouds but to no avail. We were not going to be spared. Once at the wood everyone looked enviously at Chris Bray, the forest ranger, who was the only person wearing suitable waterproofs for the occasion. Sheltering beneath the trees, which gave a little protection but which seriously hindered any talking as the rain bounced very loudly off the leaves, the plaques were handed out. Moving away from the shelter a little more could be heard. It was still a struggle to hear Paddy Tipping MP read "Remember" and then Sir Andrew Buchanan reading the names of the children and adults being remembered. I can only imagine most people's thoughts were along the lines of "I'm wet, wet, wet!" It was wet, wet, wet. It was so wet it rained into our wellies. The children did not need to splash in the puddles. The puddles were taking over. Chris in his appropriate wet weather gear rushed round and planted the trees and then each family chose a tree by which to put their plaque.

As you will see from the photos umbrellas were an essential accessory but even umbrellas provided little protection against this type of rain. It was a memorable day but on a serious note the opportunity to remember at what is normally a very peaceful, tranquil part of Sherwood Pines was not really possible. It is hard to reflect when soaked, unable to hear and battling through a deluge. I hope those families present gained an impression of the wood to imagine it on a dry day and even if not experienced were able to imagine its tranquillity and beauty. As soon as possible a retreat was made to the café and whilst I am not at all sure the café staff appreciated the traipsing in of "drowned people" spreading mud across the floor their warm drinks were very welcome before making the journey home.

Ellie Gunary
Assistant Director

Sir Andrew Buchanan, Maureen Tewson with Lisa and Amie Martin

Lindsey Devine and her father Brian Petch

Lisa and Amie Martin

Laura Devine

Clumber Park Hotel

THE SOCIETY FOR MUCOPOLYSACCHARIDE DISEASES

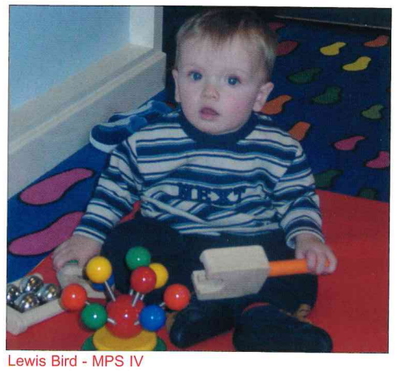
FRIDAY 26TH OCTOBER 2001

CHILDREN AND ADULTS BEING REMEMBERED

Paul Blanchard
Tracy Ann Borrett
Lee Brown
Timothy Roy Chambers
Billy Cody
Katie Devine
Shane Dickson
Tom Fuzzard-Tucker
Jason George
Stuart Lawrie
Philip Lewis
Katie Martin
David Maitland Morgan
Shaun Osment
Annette Puddy
Dennis Rowan
Annie Stokes

CHRISTMAS PARTY - NORTH EAST

Yorkshire and East Coast Christmas Party, 2nd December 2001


Lewis Bird - MPS IV


David Seymour (MPS III) and mum Angela


Eileen Smale and Amie Martin

Venue Kaleidoscope is a brand new purpose built building for children with disabilities. I have been on the steering group to get a Children's Resource Centre for eight years. Often I was the only parent in the group, but would not give up the idea of a one stop shop for the benefit of families who have a child/children with disabilities. N.C.H. Action for Children, have provided us with a lovely building and I thought what better way to give it a trial run than to have an MPS party there. If it meets the needs of our families then it would cover the majority of families.

The day started frosty, but lovely and bright. We had already delivered most of what we needed to Kaleidoscope on Friday afternoon, because it would be difficult to get David in wheelchair, 20 inflated balloons, 20 gifts for children, paper plates, cups, spoons, extra mugs etc. My younger son Christopher, who is now a teenager, and believes he is past the age of parties, decided it would be a good idea to take a TV. and his Playstation so that he and the other teenagers wouldn't get bored. The Playstation was installed in one of the interview rooms. Success! We had to remind them to come out to eat and visit Santa for their presents.

We are very fortunate to have several conference volunteers living locally, who gave up their time to come along and entertain the children. Eileen Smale took it upon herself to dress as a clown, - the outfit she wore was inflatable and wobbled, and she also spent a large amount of her time face painting. The children loved her. Eileen's partner Max got the joyous job of donning the big red outfit and doing a large amount of ho, ho, ho'ing. He too did a splendid job. A lot of the entertainment was provided by a man called Kev Brown. Kev is a teacher's aide at the local S.L.D school. He came along with a guitar and Karaoke machine. There was a large amount of sing along, pass the parcel, and budding young stars giving it their best shot at a singing career (popstars eat your heart out).

A superb buffet was provided by a local pub, plenty of crackers were pulled, sweets were eaten, drinks were drunk, conversations were had and laughter was heard through the building. Before we knew it the afternoon was at an end, children were beginning to get tired and it was getting dark. Everyone seemed to have had a good afternoon and we were soon discussing the possibility of holding another event in the spring in this comfortable, suitable building.

It was a tiring day, the cleaning up took about an hour, but it was all worth it. It's so nice to catch up with old friends and maybe could be talked into organising a spring event. Watch this space.

Angela Seymour
angie@seymour.freeserve.co.uk


Meeting Father Christmas


Matthew Ingram MPS I (BMT) and Jack Bird MPS IV

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CHRISTMAS PARTY - SOUTH EAST

South East Christmas Party, 1st December 2001

The South East Christmas Party was held on 1st December at the Maidstone Hilton Hotel. As it was Thomas Birch's 9th Birthday the same day it was agreed to combine the two events.

Six MPS families and some special guests of Thomas' swelled the numbers. After a really excellent buffet lunch all the children and young adults including those with MPS played musical chairs and pass the parcel. There was a prize for nearly everyone in between the layers.

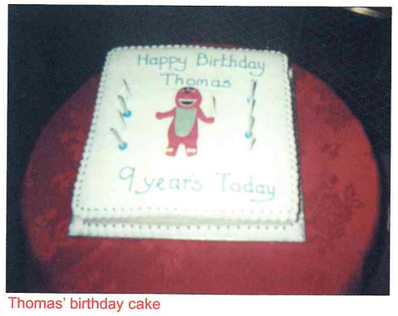
At 2 o'clock the clown arrived and to everyone's amazement kept the young ones and not so young ones amused until 3.30 when Thomas blew out his candles and everyone sang 'Happy Birthday'.

Father Christmas made a timely appearance and had a gift for each child in his sack.

Christine Lavery
Director


Thomas recieving his gift from Father Christmas


Thomas and friends



Thomas' birthday cake


Musical chairs


Musical chairs


Spinning plates is simple when you know how

CHRISTMAS PARTY - MIDLANDS

Midlands Christmas Party, 1st December 2001

Sinaed and friends

Let's drink and be happy

What a success! The Midlands MPS Christmas party added a special touch to start off what I hope will be a great Christmas. New and old faces were welcomed to the Birmingham Metropole Hotel to come together and have a great afternoon.

The party started off with a spread of very good food and excellent hotel staff to serve it to us. The staff really entered into the spirit of our party. We all did lots of talking and getting to know each other. Entertainment followed with balloons, bubbles and birds of the magic show. Laughter could be heard all round the room as the magic words 'abracadabra' were being shouted from all quarters and the magician created puppies, swords etc, from a colourful collection of balloons.

When the magic show came to an end, party music took over. The magician left his bubble-making machine on full blast with the children making the most of dancing around them and generally trying to catch as many bubbles as possible. Suddenly, just for a moment, silence filled the room, followed by the sound of 'ho -ho- ho'! Little and not so little faces looked up with anticipation. The door opened and there stood 'Santa'. Swinging his large sack off his back, Santa sat down and waited as each child at the party came up to receive their present.

Alas, my first MPS Christmas party came to an end. It was a great opportunity for local MPS families to get together and to celebrate the coming of Christmas. We all helped each other in whatever ways were necessary, in the knowledge and unspoken words that we all understood. A great afternoon, good fun, good friends and a great support! We all look forward to existing and new Midlands families joining us next year! Merry Christmas everybody.

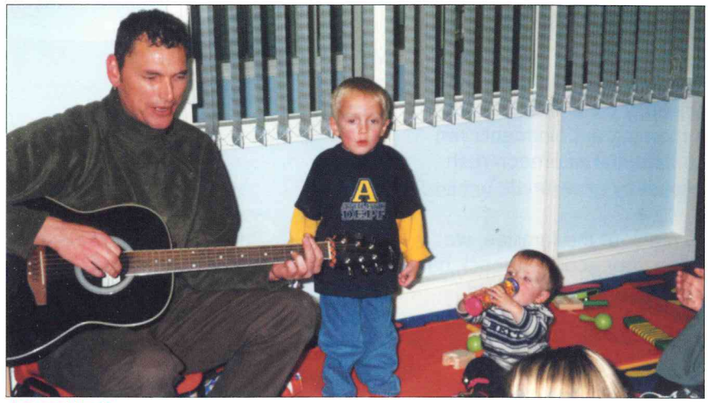
Joanna Wilson

Balloon modelling

Joanne picking a raffle prize

Hannah helping the magician

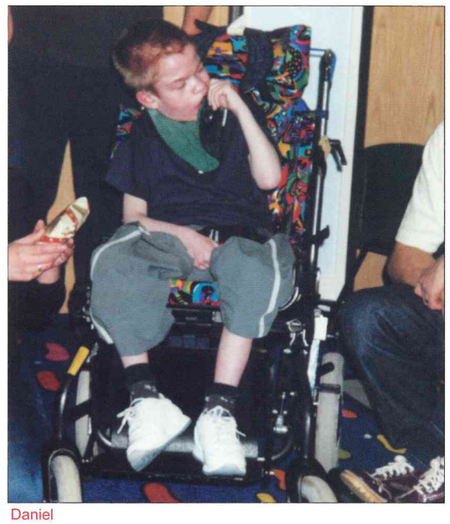
CHRISTMAS PARTY GALLERY



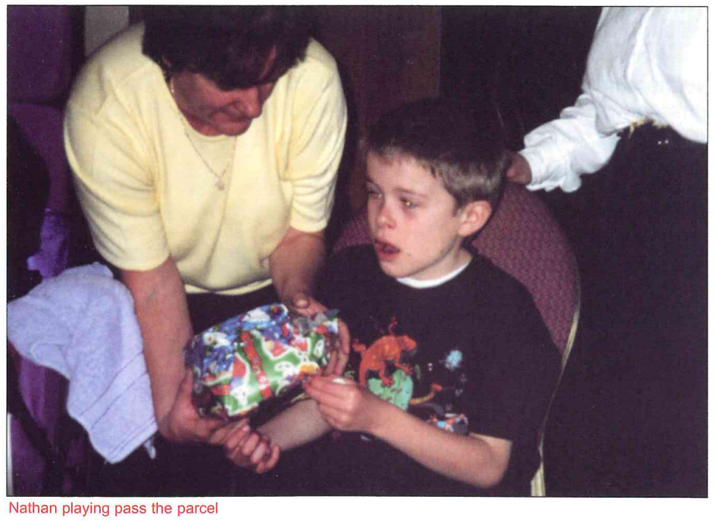
Jack and Lewis

Hannah trying her hand at magic

Jack with Eileen Smale (face painting)



Daniel



Nathan playing pass the parcel

Meeting Father Christmas

REGIONAL CLINCS

Bristol, Thursday 18th October

Alison and I attended the Bristol MPS Clinic at Frenchay Hospital. Normally this is a very busy clinic and even though the morning appointments ran fairly well to time, the afternoon rush soon got underway after lunch.

In talking to many of the families, we obtained some very good feedback but we also welcome suggestions and comments on how to improve our regional clinics and the evaluation forms sent out after each clinic are invaluable. The main problem in previous Bristol clinics has been the parking, or rather, lack of it, but the MPS Society managed to reserve a couple of parking spaces for this clinic which were appreciated.

Much of the success of this clinic should, of course, be attributed to the doctors. We would like to extend our thanks to Dr Philip Jardine and Deirdre at Frenchay Hospital for all their help in organising the clinic, and to Dr Ed Wraith for his continued support for the MPS regional clinic programme.

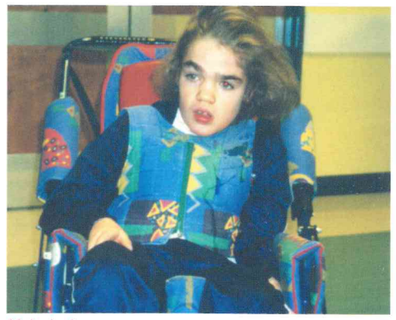
Antonia Crofts
Asst. Development Officer

Cardiff, Friday 19th October

Joseph Coleman

After a full Bristol clinic the day before, Alison and I travelled to Cardiff to attend the Welsh MPS Clinic. Despite adverse weather conditions (very, very heavy rain!) many families battled through the elements to reach the warmth of the hospital waiting room.

It was lovely to see so many families attending the clinic and taking advantage of the opportunity it provides to meet other families and see specialist consultants. Our thanks, as always, go to Dr Graham Shortland and Dr Ed Wraith for yet another successful Cardiff MPS Clinic.



Melanie Jones

Antonia Crofts
Asst. Development Officer

Northern Ireland, Friday 30th November

Lucia MPS I with her mum and grandfather

With the Northern Ireland MPS Clinic now taking place twice a year it was time for Ellie and I to return to the Hilton, Templepatrick. Compared with when we were there in May, November was cold and wet but this didn't dampen the spirits of those attending the clinic, several of whom for the first time. The atmosphere was relaxed and with two children too ill to make the journey there was time for Dr Ed Wraith and Dr Fiona Stewart to have a preview of the MPS Video. Their support and help in making the video was invaluable.

On behalf of those in Northern Ireland affected by an MPS or related disease and their families, I'd like to thank Ed and Fiona for making these clinincs possible.

Christine Lavery
Director

John ML II with his parents, Dr Ed Wraith and Dr Fiona Stewart

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MPS ADVOCACY SUPPORT PROGRAMME 2002

Christian MPS I and his father

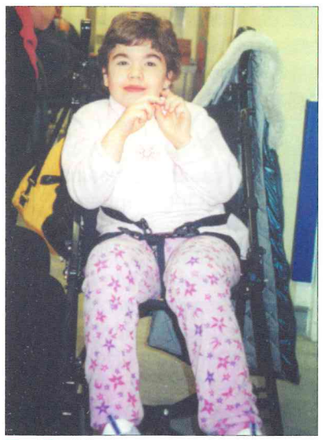
JANUARY
Friday 25th January - **Birmingham clinic**

FEBRUARY
Tuesday 5th February - **Newcastle clinic**

MAY
Friday 10th May - **Northern Ireland clinic**
Thursday 30th May - **Bristol clinic**
Friday 31st May - **Cardiff clinic**

JUNE
Friday 7th June - **Scotland clinic**
Friday 21st June - **International Symposium Paris**
Saturday 22nd June - **International Symposium Pais**
Sunday 23rd June - **International Symposium Paris**

Matthew Ingram



Faye Barnett

JULY
Sunday 7th July - **Childhood Wood Remembrance Day**
Friday 12th July - **Birmingham clinic**

OCTOBER
Friday 4th October - **Jeans for Genes day**
Friday 25th October - **Childhood Wood Planting**
Thursday 31st October - **Bristol clinic**

NOVEMBER
Friday 1st November - **Cardiff clinic**
Friday 29th November - **Northern Ireland clinic**

Jake with his mum Carol and sister in the Childhood Wood

DATES TO BE CONFIRMED

East Anglia clinic

The East Anglia clinic has traditionally been held in January at the Norfolk and Norwich hospital. The Norfolk and Norwich Hospital has in the last year moved and due to this relocation it has been known for some time that the annual MPS clinic would not be able to take place at this hospital until later in the year. All individuals and families using this clinic were consulted and the feedback received was conclusive. Individuals with MPS, their families and carers would rather wait until later in the year for a clinic at the Norfolk and Norwich Hospital rather than consider an alternative location.

South East clinic

It is hoped that a clinic will be held in the south east of England. As soon as progress has been made to secure the support of a local consultant and a date arranged with Dr Ashok Vellodi all the individuals and families who would benefit from this clinic will be contacted.

Regional Days

A programme of regional days is being planned to include events throughout the UK at different times of the year including Christmas Parties.

National MPS Conference

After considerable thought and due to the superb programme being offered at the International Symposium in Paris, it has been agreed to provide additional help to get as many individuals with MPS and Related Diseases and their families to the Paris conference as possible. As a consequence the Society will not be holding a National Conference in 2002. If you want to come to the Paris conference but are concerned about travel and costs please call Fiona now.

INTERNATIONAL CONFERENCES

International Conferences


Craft Workshop at German MPS Conference


Maria and Paul with friend at Hungarian Conference


Richard & Elizabeth Volk with Christine at German MPS Conference


Anna and Paul Ubleis at Hungarian Conference


MPS Conference, Germany

To celebrate their 10th Anniversary the Hungarian MPS Society held a conference and meeting of the International Working Party on Mucopolysaccharide and Related Diseases at Lillifured from 11th to 14th October 2001. Following the events of 11th September a number of invited speakers and representatives from European MPS Societies didn't make it. However Ellie and Christine presented papers on Advocacy Support and the MPS European Registry and attended the International Working Party meeting. There were useful discussions on disability and terminology as well as the European MPS Registry. Dr Michael Beck from Mainz, Germany provided an update on current and future therapies for MPS and related diseases and Delphine Genevaz from VML spoke about the International Symposium to take place in Paris from 20th to 23rd June 2002.

Less than 12 hours after arriving back in the UK Christine and Ellie left for San Diego in the US to participate in a meeting to discuss Enzyme Replacement Therapy and clinical trials for MPS II Hunter disease. Whilst in San Diego they also attended a meeting on Fabry Disease hosted by TKT.

In October 2002 the Society and Dr Ed Wraith will be supporting a clinic and conference for patients who have a Mucopolysaccharide or related disease in Norway. In preparation for this clinic and conference and en route to Sweden, Christine met with Kjersti Grindal and Odrun Gronvik at Frambu in Norway to develop their plans.

On the island of Marstrand the University of Gothenburg had organised an International Postgraduate course on Lysosomal Storage Diseases. Christine was invited to attend and spoke on the role of patient organisations.

In November the German MPS Society held their 15th Weekend Conference at Wurzbach in the forest of Thuringen, south of Leipzig in former East Germany. This was the biggest MPS conference, outside the International Symposiums, ever held with over 100 families attending. Dr Emil Kakkis came from California to speak on Enzyme Replacement Therapy and during the weekend Christine presented him with a framed photo of Dr Ed Wraith's team and patients who took part in the clinical MPS I trial.

As you will see from the photo below, Winter Wonderland came early in that part of Germany and the children were kept busy with indoor activities that included drama workshops, painting, pottery and a creche. On the Saturday evening prior to the Gala Dinner the children that took part in the drama workshops performed their pieces to the parents and guests. The children clearly enjoyed their moments of fame and it was wonderful for their parents too.

There were very useful discussions with the Trustees of the German MPS Society as well as an opportunity to meet and learn from the families. During the weekend Christine also visited in hospital one of the oldest ladies to be suffering from Morquio Disease. Christine has met Heideros many times since they first met at the Austrian MPS conference at Sharding in April 1986 and was saddened to see her so unwell.


Christine at the German Conference in Wurzbach

JEANS FOR GENES DAY

Jeans for Genes reaches a record total - £2,513,545

We extend a very big thank you to everyone who supported Jeans for Genes in 2001. With your help a record £2,513,545 had been raised by December 2001.

It is now time to begin planning for the 2002 campaign. Your support, whether it is handing out Jeans for Genes Fundraising packs, organising an event or providing a story to the media, is vital in making this campaign a success. Enclosed with this newsletter is a short questionnaire, which we are asking every individual and family to complete and return to the MPS office. Anyone who is able to speak to the media will be contacted by a member of the Development Team and provided with support. The Jeans for Genes Management Team is frequently asked by local newspapers for the type of stories that could be provided. With the help of our members and supporters we hope to provide the Management Team with potential stories (which at this stage will retain the anonymity of the families and individuals) for this purpose. At no time will any personal details be given without your agreement and contact from the Development Team first.

Monies raised through the Jeans for Genes campaign enable the MPS Society to carry out vital research into MPS and Related Diseases and fund advocacy support projects including the equivalent of a Development Officer post and the Regional clinic programme.

Please help us to help you make Jeans for Genes 2002 an even bigger success by completing the short questionnaire enclosed.

Ellie Gunary
Assistant Director


Alison, Ellie, Antonia and Angela on Jeans for Genes Day

Ben Richarson appears in the Rugby Advertiser promoting J4G Day

A Mother whose toddler son was diagnosed with a terminal genetic disease is hoping to raise public awareness through this year's Jeans for Genes day.

Ben was diagnosed with an MPS disease known as Sanfilippo one month ago. Mrs. Richardson, Ben and his one year old sister Georgia, who attend the Noah's Ark playgroup in Northampton enjoyed various fundraising activities this week in a bid to boost charity funds.

Mrs. Richardson told the Advertiser: "I had never heard of MPS before Ben was diagnosed and expect most other people have not either. So many people take part in the Jeans for Genes day and are unsure about what they are helping towards and I would just like to help to raise public awareness."

Children with Sanfilippo disease are missing an enzyme essential in helping to break down mucopolysaccharides. Without this enzyine the mucopolysaccharides remain stored in the cells causing progressive toxic damage. Physical problems such as deafness, bowel problems, seizures and epilepsy and hip disease may affect sufferers. "I always knew something was not quite right with Ben from an early age, call it a mother's intuition," she said. "He was hard to potty train, milk intolerant and he would always be falling over and bumping into things. He constantly needs to chew things and was in and out of hospital from a very young age. When we were told it was a massive shock and you realise that all those nice things you had planned will all have to be done that much earlier." She added, "But you have to carry on, stay positive and hopeful really. You put it to the back of your mind. He is well at the moment and that is what you have to stay focused on."

Mrs. Richardson hopes that if no cure is found in Ben's lifetime then maybe funds can go towards helping others like Ben. "We are working towards the future. Maybe we can help other children in the future and their families."


Ben and Mrs Richardson

Article and photo reproduced by kind permission of the Rugby Advertiser

RESEARCH UPDATE

BioMarin and Genzyme to file for marketing approval for Aldurazyme based on Phase 3 trial results

Based on the strength of the trial's results, the companies plan to meet with US, Canadian, and European regulatory authorities to discuss applications to market Aldurazyme.

The following is a press release dated Nov. 2,2001, announcing positive results from a preliminary analysis of data from the Phase 3 MPS I enzyme replacement therapy trial.

The Phase 3 trial enrolled 45 patients at five sites in the U.S., Europe, and Canada in a randomised, double-blind, placebo-controlled study designed to evaluate the safety and efficacy of Aldurazyme in patients with MPS I. During the 26-week evaluation period, 22 patients received weekly intravenous infusions of Aldurazyme and 23 patients received weekly placebo infusions. The 45 patients ranged in age from 6 to 43, with an average age of 15 years old. All patients completed the trial and have elected to receive Aldurazyme in an open-label extension study.

Patients were evaluated at defined intervals to assess progress in meeting two primary endpoints. The preliminary data analysis showed a statistically significant increase in pulmonary capacity (p=0.028), and demonstrated a positive trend in endurance as measured by a six-minute walk test (p=0.066).

Among other endpoints measured in the trial, the main findings of an earlier open-label study of Aldurazyme were confirmed: a reduction in liver size and a reduction in excretion of urinary glycosaminoglycans (GAGs), the carbohydrate substances that accumulate in patients with MPS I. Data from the earlier trial were published in the New England Journal of Medicine in January, 2001.

The safety profile was comparable between the treatment group and the placebo group. There were no Aldurazyme-related serious adverse events. The most commonly reported reactions were fever (14 patients on the placebo and 10 in the treatment group); headache (16 on placebo and 11 in the treatment group); rhinitis (10 on placebo and 8 in the treatment group); and rash (5 on placebo and 8 in the treatment group).

"The data described confirms and expands upon the results demonstrated in our first clinical trial," said Richard Moscicki, M.D., senior vice president and chief medical officer at Genzyme. "We have amassed the body of evidence that supports the potential benefits of Aldurazyme for patients with MPS I."

"This milestone in the development of Aldurazyme represents a critical step toward delivering a much-needed therapy to MPS I patients," said Stuart J. Swiedler, M.D., Ph.D, BioMarin's vice president, clinical affairs. "We look forward to discussing the next steps with worldwide regulatory agencies."

Developing New Therapies

MPS IV-A

BioMarin is developing a programme to give the missing enzyme, galactose 6-sulfatase, to individuals with MPS IV-A. Studies in animal models suggest that, if given early, the enzyme can change the outcome of bone and cartilage disease. At this time, there is no clinical trial planned.

MPS VII

Drs. Emil Kakkis and William Sly have received a grant to develop enzyme replacement for MPS VII. The initial studies will be conducted on the MPS VII mouse and, if successful, application will be made to the FDA for a clinical trial in individuals with MPS VII.

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RESEARCH UPDATE

NORD awards over $300,000 in Lysosomal Storage Disease Research Grants

New Fairfield, Conneticut, 11th October , 2001. The National Organization for Rare Disorders (NORD) has awarded the first NORD Roscoe Brady Lysosomal Storage Disease (LSD) Fellowships, totaling $337,680, to five physician researchers from the US, Japan and Brazil. This year's award winners will explore the causes of Fabry, Krabbe, Hunter, Gaucher ("go-shay") and Tay-Sachs disease. Abbey Meyers, NORD's president, says, "Our grant recipients will help solve the mysteries behind the genetic defects and enzyme deficiencies that lead to these orphan diseases."

Lysosomal storage disorders are categorised by an abnormal build-up of various toxic materials in the body's cells that affect the skeleton, brain, skin, heart, and central nervous system. People living with one of these genetic diseases may lose function in more than one system of the body with severe medical consequences and often shortened life spans. There are currently no effective treatments for the vast majority of these diseases.

Meyer's says, "The NORD Roscoe Brady LSD Fellowship Program funds young scientists at the beginning of their medical careers and encourages them to specialize in lysosomal storage disorders."

All grant recipients will work under the guidance of senior mentors in the fields of genetics and neuroscience. NORD also hopes that the fellowship program will entice established researchers to direct their attention to LSDs.

Million Dollar Gift Creates Fellowships

This spring, Transkaryotic Therapies (TKT), a Cambridge, Massachuetts biotechnology company dedicated to improving the lives of present and future generations by developing products to treat rare diseases, committed $1 million to NORD to encourage scientific research on lysosomal storage diseases. This extraordinary gift, together with additional support from Genzyme Corporation and individuals interested in LSD research, allowed NORD to create the Roscoe Brady LSD Fellowships. The Fellowships are named to honour Dr. Roscoe Brady, the scientist who discovered the metabolic basis of several lysosomal storage disorders and who pioneered effective enzyme replacement therapies for Fabry and Gaucher disease.

Dr. Brady is Chief of the Development and Metabolic Neurology Branch of the National Institute of Neurological Disorders and Stroke, and continues to pioneer our scientific understanding and treatment of these rare diseases.

Research Projects receive Funding include:

Dr. Makiko Yasuda, also from Mount Sinai, will study regulatory elements that control the defective gene that causes the enzyme deficiency identified with Fabry disease, hopefully generating information for potential gene therapies.

Dr. Ida Schwartz, a scientist at Hospital de Clinicas de Porto Alegre, Brazil, will concentrate her research efforts on analysing the biochemical and genetic characteristics of patients with Hunter disease, and of women who may be carriers of the disease. She seeks to find a correlation between the internally coded, inheritable information which programs the machinery of the cells, and the observable physical structure, function or behaviour of people living with Hunter disease.

RESEARCH UPDATE

Web Based Patient Registry for MPS VI (Maroteaux-Lamy Syndrome)

The Women's and Children's Hospital (WCH) in Adelaide is pleased to announce its participation in a web site Registry dedicated to patients affected by MPS-VI (Maroteaux-Lamy syndrome).

What is the purpose of the Registry?
The Registry's main purpose is to create a world-wide database of people affected by this disorder that provides information about its prevalence, the range and severity of clinical symptoms, and up-to-date information for patients about research results, clinical trials and new treatments.

What information will be collected?
Participants will be asked to answer a series of general medical questions relevant to MPS-VI. They will also be asked to provide their names, addresses and contact numbers. Contact details for their physician will also be required. The results of laboratory or other tests will not be included in the Registry.

What is the relationship between WCH and BioMarin for the purposes of the Registry?
BioMarin and the Women's and Children's Hospital have entered into a special arrangement to establish this Registry.

The project supervisor at the Women's and Children's Hospital is Professor John Hopwood. The project has been approved by the Women's and Children's Hospital Ethics Committee.

Under the arrangement, BioMarin is contractually obligated not to access individually identifiable patient data, nor make it available to others who may have an interest in obtaining the data. BioMarin has agreed to be bound by this obligation on an on-going basis.

BioMarin will, however, have access to the information in deidentified (anonymous) form, as will the general public. For example, this will allow BioMarin (and the general public) to obtain information from the Registry that tells them how many people in the world are affected by MPS-VI, but it will not tell them their names or give them any other information that may identify an individual.

The Women's and Children's Hospital will be involved in the Registry data collection for a period of 12-months. After this initial establishment phase, BioMarin intends to transfer the database and the task of on-going data collection to a non-profit organisation dedicated to alleviating suffering of MPS-VI patients. This organisation will be under the same obligations of confidentiality as the WCH.

Who will have access to the information?
The only people who are permitted access to information that identifies patients will be the Registry Coordinator and several other staff at the Women's and Children's Hospital who are directly involved with this project. These individuals are under strict obligations of non-disclosure.

BioMarin will have access to the information to enable on-going maintenance of the web site.

Who owns the information?
The web site and Registry database are owned by BioMarin Pharmaceuticals, Inc. BioMarin is based in California, USA, and is developing therapeutics for MPS-VI. The information contained in the database is owned by BioMarin, but each individual who registers is granted a license to access their own data only.

How secure is the web site?
Whilst strict security measures have been incorporated into the design of the web site by an independent third party, and both the Women's and Children's Hospital and BioMarin are under strict obligations of confidentiality and non-disclosure, neither is able to guarantee that the site and its contents will not be breached by unauthorised users (i.e. 'hackers').

Are there any costs involved?
Participation in the Registry is free of charge.

MPS Newsletter Winter 2001

RESEARCH UPDATE

Can patients withdraw their information from the Registry?
Patients have the right to withdraw their participation at any time by completing the appropriate document on the web site.

Who can register, and how?
Anyone affected by MPS-VI can register by accessing the web site (http://www.mpsvi.org). Patients will be asked to identify the physician who manages their symptoms associated with MPS-VI. The physician will then be required to verify the information provided by the patient before registration will be deemed complete. Patients under the age of 18 years will require the approval of their parent/guardian before being able to register. Once the process of verification is complete, the Registry Coordinator will issue each patient with a special code number to maintain confidentiality and privacy.

The web site outlines the Terms and Conditions of Use and provides a Privacy Statement which details what information is being collected, circumstances under which it might be released and the measures in place to ensure its security.

If internet access is not available, the required documents can be obtained in paper form by contacting the Registry Coordinator on the appropriate toll-free number listed in this leaflet.

Further questions?
Special toll-free numbers have been established for people to obtain more information about the Registry from the Registry Coordinator. **Please note that English is the preferred language for the use of this service**.

Voicemail has been arranged for calls that may be received outside of normal business hours. In this circumstance, please leave your name, contact telephone number and the country you are calling from, and the Registry Coordinator will return your call on resumption of business the next day.

Contact Details:
MPS-VI Registry Coordinator
Department of Chemical Pathology
Women's and Children's Hospital
72 King William Road
NORTH ADELAIDE SOUTH
AUSTRALIA 5006
AUSTRALIA.

Toll-free numbers:

United Kingdom	0800 7314 485
Fax:	(08) 8161 7100
International prefix:	(618)

Email: oatess@senet.com.au

Global MPS VI Registry

MPS Classifications

Syndrome		Enzyme Deficiency
MPS I H	Hurler	alpha-L-Iduronidase
MPS I S	Scheie	alpha-L-Iduronidase
MPS I H-S	Hurler-Scheie	alpha-L-Iduronidase
MPS II	Hunter	Iduronate sulfatase
MPS III A	Sanfilippo A	Heparan ***N***-sulphatase
MPS III B	Sanfilippo B	alpha-***N***-Acetylglucosaminide
MPS III C	Sanfilippo C	Acetyl CoA: alpha-glycosaminide acetyltransferase
MPS III D	Sanfilippo D	***N***-Acetylglucosamine 6-sulfatase
MPS IV A	Morquio A	Galactose 6-sulfatase
MPS IV B	Morquio B	beta Galactosidase
MPS VI	Maroteaux-Lamy	***N***-Acetylglucosamine 6-sulfatase
MPS VII	Sly	beta-Glucuronidase
MPS IX		Hyaluronidase
ML II	I-Cell	***N***-acetylglucosamine-1-phosphotransferase
ML III	Psuedo-Hurler polydystrophy	***N***-acetylglucosamine-1-phosphotransferase

INFORMATION EXCHANGE

Genetics and Insurance

The use of genetic information by insurance companies has been a hot topic this year. In our view too much emphasis has been placed on genetic information revealed by DNA tests at the expense of other forms of predictive information such as family history. GIG has campaigned hard to broaden the debate so that the relevance of all types of genetic information can be looked at, not just the results of DNA tests. We have also argued for the need for proper social welfare benefits for all those affected by genetic disorders as a must, rather than relying on private insurance as a means of providing for individual wealth and well being when genetic disease occurs.

Alistair Kent, Chief Executive of GIG is a member of the UK Forum of Genetics and Insurance and the Association of British Insurers Genetics Committee. He has engaged in an educational role with underwriters and geneticists to create better mutual understanding of the way in which both of these groups go about their business.

Reproduced with kind permission of the Genetic Interest Group

Disability Rights - New Ban on Genetic Discrimination

Fears of a future "genetic underclass" were allayed last week by the announcement of a five year moratorium on the use of DNA test results by insurance companies.

The ban on the use of genetic tests will begin in November, but insurers will still be able to use genetic test results authorised by the Government's genetics and insurance committee (GAIC) for life assurance policies of more than £500,000 and critical illness, income protection and long-term care insurance of greater than £300,000.

Alzheimer's Society chief executive Harry Cayton has welcomed the agreement between the government and the industry trade body, the Association of British Insurers (ABI). "We hope that the moratoriurn will halt genetic discrimination and encourage families to participate in genetic research without fear," he said.

The only test approved by GAIC so far is for Huntington's Disease - a hereditary degenerative disease which attacks the central nervous system - in the context of life assurance cover, although the ABI has applied to the committee to use two genes associated with familial early onset Alzheimer's disease.

Cars for Disabled People

The disability website www.youreable.com has teamed up with the Bank of Scotland in a new scheme aimed at helping disabled people get cars.

The scheme will particularly help people not eligible for higher rate mobility component of Disability Living Allowance. This group is not entitled to cars through the Motability scheme which supplies about 150,000 vehicles a year.

Youreable chief executive Stephen Harpin said: "Those that do not qualify for the scheme often find it difficult to get credit. If people go through this scheme, the Bank of Scotland will now take into account state benefits when they assess the credit risk."

Under the new scheme which is a partnership with Bank of Scotland Freeway, disabled people can pay a deposit and monthly payments for two or three years. The disabled person can buy the car, leave the scheme, or opt for another vehicle at the end of the contract.

Bank of Scotland Freeway, tel: 0845 7697243, youreable web site: www.youreable.com

MPS Newsletter Winter 2001

INFORMATION EXCHANGE

EQUIP

EQUIP (Electronic Quality Information for Patients) www.equip.nhs.uk is a new NHS website of quality information for UK patients and carers.

Information is gathered on medical topics and filtered through quality criteria, so that only good quality, useful websites are selected to appear on EQUIP.

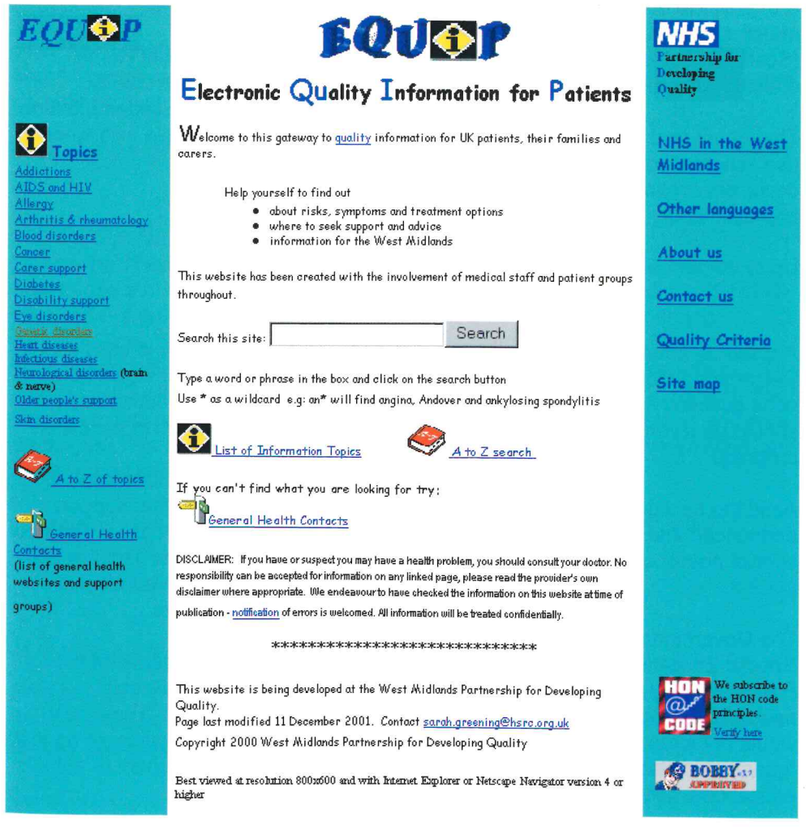
Each medical topic has 5 sections:
1. Links to information-rich websites that have either been written or checked by doctors
2. Links to other useful websites on that topic that have been filtered through a quality toolkit
3. National support groups and helplines
4. West Midlands contacts and support groups
5. Other useful information - ie: leaflets or videos available

For medical topics not covered yet, there is a page of general health website links and general support contacts and helplines.

There is also a selection of information resources in languages other than English and a section on West Midlands NHS services.

Patient support groups for major topics have been involved in discussions about the content and format of the website. EQUIP is being used as a primary resource for all NHS Direct call centres in UK and is being recommended by many health professsionals. Although it has some additional West Midlands Information, most of the website is appropriate for all of the UK.

The content of the website is being expanded and reviewed constantly and we welcome support groups to contact us with information for the website. I hope that you will have a moment to take a look at the site and any feedback would be very welcome.



INFORMATION EXCHANGE

Capital Limit Change helps Independence

LEGISLATION

Up to 1,000 disabled people could benefit significantly from changes to the independent living fund announced by the government last week.

As part of the annual changes to benefit rates coming into effect next April, the Department for Work and Pensions said the capital limits for application to the ILF would increase from £8,000 to £18,500 and the earnings of beneficiaries and their partners would be disregarded in assessments.

Each of the two changes will affect about 500 people, said disability minister Maria Eagle, making ILF recipients £130 per week better off on average.

"While this helps only a small number of people, it helps some of the most disabled people," added Eagle. "It fits in very well with our approach of helping people to help themselves to work." Among the other benefit changes announced by the DWP were a £5 increase in the disabled child premium to £35.50, and rise in the standard rate of statutory maternity pay and maternity allowance from £62.20 to £75 per week, rising to £loo per week in Apri12003.

New Scottish Housing Law

Ownership Options in Scotland

A new housing law now forces local authorities in Scotland to keep records of local housing suitable for disabled people.

All 32 Scottish local authorities also have to consider the needs of people with physical disabilities in their housing plans, thanks to the Housing (Scotland) Act 2001, which came into effect in November.

They must also explain how they will use schemes to help disabled owner occupiers organise repairs and improvements.

Up to 20 per cent of disabled people in Scotland are believed to live in unsuitable housing.

Jackie Baille, Scottish Social Justice Minister, believes that increasing the amount of suitable housing will allow disabled people to live where they want and in the tenure they choose.

Meanwhile, a new booklet Home Truths, published by the charity Ownership Options in Scotland, offers advice to disabled people who want to own their own homes.

To get hold of the booklet tel: 0131 661 3400 or email oois@talk21.com

Education U-turn

LEGISLATION

A disability education group has welcomed a Government U-turn on a code outlining how special support should be given to disabled school children.

Amid the backdown, ministers also announced that funding to help meet special needs would be £91 million in 2002-3.

The Government withdrew a draft Special Educational Needs Code of Practice in July after pressure from campaigners who were angry that it would not force education authorities to "quantify" in statements of special educational needs how much support, such as speech therapy, a child should be given. They feared it would be used as a loophole allowing authorities to make the bare minimum in provision.

MPs approved an amended code in November which says that support should normally be quantified. The new code comes into force in January 2002 along with the Special Educational Needs and Disability Act.

John Wright, chief executive of the Independent Panel for Special Education Advice, said: "They must now enforce this. If they cannot do that we will be back asking them to put it into law."

MPS Newsletter Winter 2001

INFORMATION EXCHANGE

£3m Boost for Carers

£

The charity Carers UK will head a £3m project to help carers find work and get training. The scheme, backed by the European Social Fund, is called Action for Carers and Employment and is expected to help 2,000 carers over a three year period.

Tel: 020 7566 7843

Mobility Assist Group (MAG)

They offer advice and assessment on the range of equipment available to enable less mobile people of any age to live independently.

Tel: 0845 6000 166

Citizens' Advice Bureau

citizens advice bureau

adviceguide

CAB have put information on a website which is updated regularly and covers everything from housing, debt and the legal system to tax, benefits, family matters and consumer rights.

www.adviceguide.org.uk

British Bankers' Association

BBA BRITISH BANKERS' ASSOCIATION

They have produced a free information sheet with the following title, "Signing your name, how to overcome the difficulties". This could be useful for people with poor co-ordination or sensory loss.

www.bba.org.uk

Free Computer Training for children in the south West

BIBIC

Children with learning difficulties aged 13 - 19 can now receive free training on how to use a computer, thanks to a grant from the European Social Fund. BIBIC received the grant to teach children with any form of disability or learning difficulty. They do not have to be registered disabled or have a statement of special educational need and carers are also welcome to participate free of charge. Staff will take children through whatever they need to learn, ranging from the usual packages (eg Microsoft Office) to the internet and specialised software packages. This is to enable more effective communication. Adaptive devices can be arranged if needed.

Contact Pam Brookes at BIBIC (Bridgewater, Somerset) Tel: 01278 684060
email: info@bibic.ora.uk or see www.bibic.org.uk

It's Your Call

This is a new guide to telephone services for disabled people giving the special services that are on offer, eg, free directory enquiries, alternative bill formats and priority fault repairs together with a comparison table.

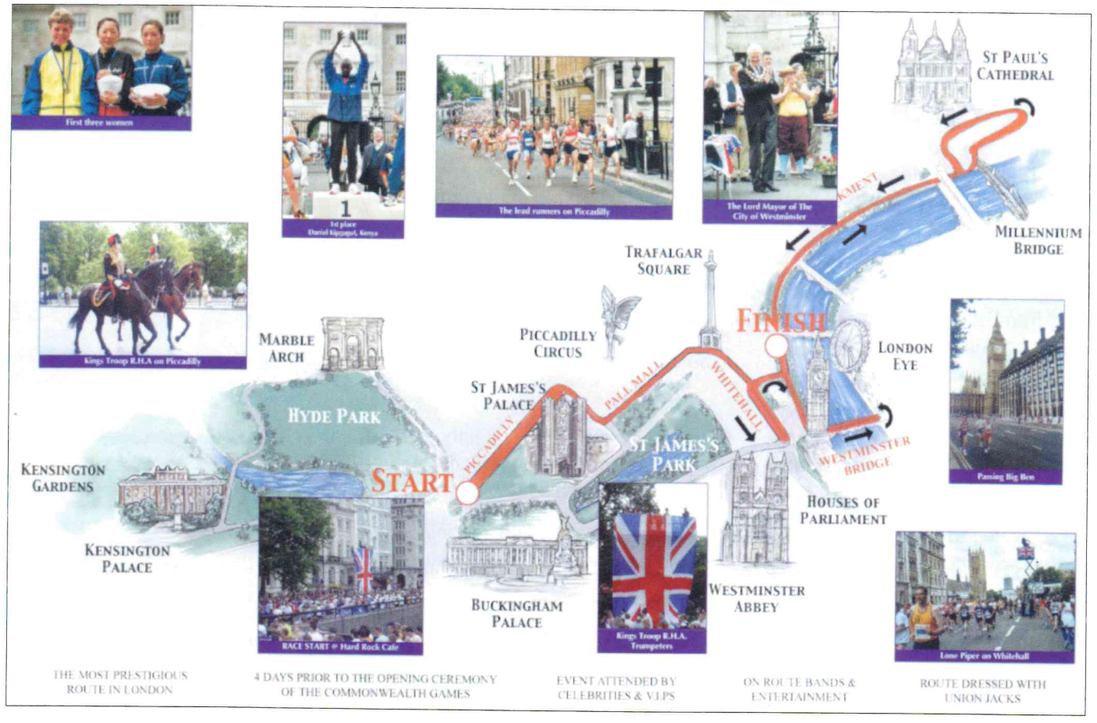
Tel: 0207 427 2460

FUNDRAISING

10K Open Road Race

Last year's 10K Road Race was such a sucess with £2,300 being raised and still money to come, we have decided to enter again.

We have secured 24 places for this year's event to be run on 21st July 2002. Entry forms are available from the MPS office and have to be completed and returned to the MPS Society by 15th April 2002.



Payroll Giving

The Inland Revenue has come up with a good scheme that allows you to give donations to the MPS Society and save on tax at the same time.

Two thirds of the population give to charity but better use could be made of this money if it was deducted from your gross pay before tax. This means that the money that you would have 'given' to the taxman will go straight to the MPS Society.

Under the scheme if you pledge to give us £10 through 'Payroll Giving' then it will only 'cost' you £7.80 but it is worth £11 to a charity. A big difference. This is 41 percent more than if you put the money into a collection box. For higher rate taxpayers £10 through the payroll would cost the individual £6 but be worth £11 to a charity -83 percent more. This is because until 2003 the Government is adding 10 percent to all donations made in this way.

Remember the Government's additional 10% is only available until 2003. Any PAYE employee can donate through Payroll Giving as long as your employer is signed up for the scheme. And if not, why not?

www.inlandrevenue.gov.uk/parollgiving
or telephone Gina at the MPS office

DONATIONS & FUNDRAISING

The Society is grateful to the following who made donations

Anne Hill
Mr S J Hill
Lesley Edwards
Val Challen
The Oddballs Golfing Society
Mrs E A Armstrong
Caversham Folk Festival
John Hogg Butchers
Mrs C Gooch
F E Woods
Mrs P Harris
Joyce Wilson
David Wraith
Mrs D West
Linda Browning
Vickie Brockie
Mrs C Rockett
Mrs M Howells
Community Mental Health Team
Pat Shroff
Bank of England
Christopher Laing Foundation
Mr Dickey
Joseph Strong Frazer Trust
Anthony Andrews Ltd
Shazia Chaudry
Loreto Grammar School - Omagh
Genzyme Corporation
Entertainment Software Charity
John Jackson Memorial Cup Darts League
Raspin Family
Hardy Family
J Davis
St Mary;s Centre - Haddenham
Methodist Ladies Fellowship - Haddenham
Long Marston & Puttenham Horticultral Society
The Fitton Trust
Andrew's Family & Friends
Mrs Dagnall
Herbert & Peter Blagrave Charitable Trust
S Blanch - Norwich

The Society is grateful to the following who held fundraising events

A R Mellor & Associates
Jane Coney, Alison Thompson, Erica Coney,
Kim Coney - Sponsored 20 Mile Walk
Church Council of St Lawrence Parish
Parish of Cirencester with Watermoor - Charity Lunch
Charity Flowers
Maria Murphy - Half Marathon
Stuart Gascoyne - Open Country Event
Tresidder Family
Kingswood & Hanham Bowling Club
Margaret McMorran/Janice Corden - Sponsored Famine
M I Stimpson - Webb Ivory Sales
Mrs L Hutley, Mrs J Timms, Mrs F Thomas and
Mrs E O'Hea - Wintershall Tennis Tournament
The Crystal Bingo - Deeside
Gala Bingo - Wirral
The Royal Stewart - Gretna Halloween Disco
Andy Whettem - Goodwood Bathrooms
Sponsored Walk (106 miles)
The Terry School of Dance - Evening of Dance
Denis Otway - Great North Run
Langley Ward Conservative Club - Oldbury
Stonelaw High School - Sponsored Swim

Collection Box

Kirkinner Inn - Kirkinner

Stamps

Norman J Wigley & Partners
Hardy Family
Johnson & Johnson
Martin Family

Foreign Coins

Johnson & Johnson

Marathon Runner

Rosemary Nurse's nephew Neil Carey is running in the 2001 London Marathon for the MPS Society. If anyone would like to sponsor him for this event please contact either Gina at the MPS office or directly to Neil at:

87 Glendale
Swanley
Kent
BR8 8TP

MPS GRAND DRAW 2001

£500 Cash
J Bates - Chalfont St Giles

Samsung DVD & £100
P Scannett - Belfast

Dual Cyclone Dyson
S Johnson- Shrewsbury

Kids Bike & Helmet
no name - Essex

Polaroid Camera
G Kershaw - Frodsham

Casio Baby G Watch
P Hughes - Stockport

Mini Electronic Organiser
K Rowe - Kent

£30 Argos Voucher
E Nicholls - Cheshire

Tasco 8x Binoculars
A McNeil - Southampton

Dancing Rudolph
J Cooper - Lincolnshire

Signed Samantha Mumba T-Shirt
P Orchard - Herts

Milk Frother
W McConnor - Co. Down

Games In Leather Case
M Herbert - Bristol

Taylors Port
J Slates - Londonderry

£10 Wyvale Voucher
Leaver - Radstock

£10 Sainsbury Voucher
D Jones - Derby

2x Guest Passes UCI
K Lattam - Derbyshire

Euro Calculator
Gremo - Kent

£8 Marks & Spencer
D James - Glamorgan

£10 Alldays Voucher
M Fasey - Notts

MPS GRAND DRAW 2001

2x Guest Passes UCI
D Murphy - Kent

Mini 24 Piece Tool Kit
L Lee - Portsmouth

Lego
R Jenkins - Hereford

Lego
JL Edwards - Cardiff

Lego
Kembrey - Bristol

Lego
E Quinn - Glasgow

Toiletry Bag
B Osborne - Birmingham

Flash Light Key Chain
E Porker - Belfast

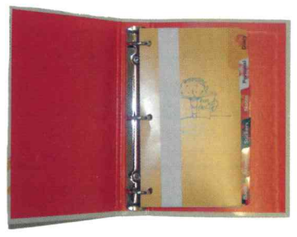
Document Bag
Pitt - Colchester

Heart Stationery Set
N Harvey - Oxford

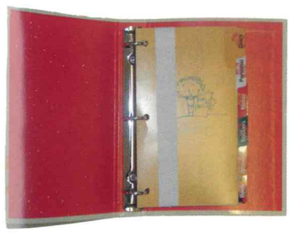
2 Bottles White Wine
Harris

Photo Frame Clock
Wragg - Worthing

Eurocamp Rucksack
M Milo - Dedham



Filofax
S Hodges



Filofax
S Booth - Middleton

Eurocamp Rucksack
S Brown - London

Please note that prizes may differ slightly to those shown in the photos.

CONTACTING THE SOCIETY

MANAGEMENT COMMITTEE

Chairman	Barry Wilson	
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Treasurer	Judith Evans	
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	Gina Page	Finance Officer
	Alex Roberts	Project & Information Officer
	Fiona Woodcraft	Project & Information Officer
	Mounira Hadj-Rehouma	Development Officer
	Antonia Crofts	Assistant Development Officer
	Alison Britton	Assistant Development Officer

CONTACT US

MPS OFFICE: 46 WOODSIDE ROAD, AMERSHAM, BUCKS HP6 6AJ

Tel: 01494 434156
Fax: 01494 434252

OUT OF HOURS HELPLINE: 07712 653258

email: mps@mpssociety.co.uk
Home Page: www.mpssociety.co.uk
Fabry: www.fabry.org.uk

NEWSLETTER DEADLINES

SPRING
31 March 2002

SUMMER
30 June 2002

AUTUMN
30 September 2002

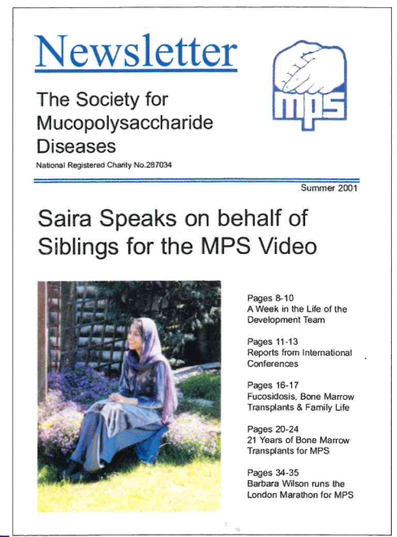
WINTER
17 December 2002

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

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

The articles in this newsletter do not necessarily reflect the opinions of the MPS Society or its Management Committee

The MPS Society reserves the right to edit content as necessary.

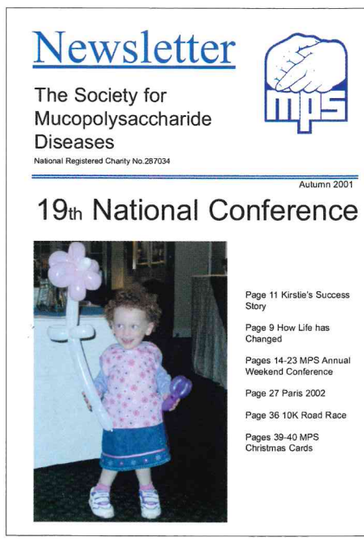

Newsletter

The Society for Mucopolysaccharide Diseases

National Registered Charity No.287034

Summer 2001

Saira Speaks on behalf of Siblings for the MPS Video


Newsletter

The Society for Mucopolysaccharide Diseases

National Registered Charity No.287034

Autumn 2001

19th National Conference

Back Issues

If you have missed an edition of the MPS quarterly Newsletter or know someone who would like one, you can order a back issue. We currently have Summer 2001 and Autumn 2001 in limited stock.

Each copy is £2.50 including postage & packaging. You can pay by sending a cheque payable to 'MPS Society' to the address above, or by credit card over the phone.

7th INTERNATIONAL SYMPOSIUM ON MUCOPOLYSACCHARIDE AND RELATED DISEASES

20th - 23rd June 2002

PARIS. "LA TOUR EIFFEL"

BOOK YOUR PLACE NOW