

Newsletter

The Society for
Mucopolysaccharide
Diseases

National Registered Charity No.287034



Summer 2001

Saira Speaks on behalf of Siblings for the MPS Video



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The Making of the MPS Video Behind the Scenes



Sophie - Sanfilippo



Aaron - Sanfilippo, Dr Fiona Stewart, Dr Alex McGee & Valerie Peacock



Lucia - Hurler & Mum Rosemary



James - Hunter



'CARE TODAY, HOPE TOMORROW'

What is the Society for Mucopolysaccharide Diseases ?

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

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

How does the MPS Society meet these Aims?

Advocacy Support

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

Telephone Helpline

Includes out of hours listening service

MPS Befriending Network

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

Support to Young People and Adults with MPS

Empowering individuals to gain independent living skills, healthcare support, further education, mobility and accessing their local community

Regional Clinics, Information Days and Conferences

10 regional MPS clinics throughout the UK and information days and conferences in Scotland and Northern Ireland

Regional Events

Social events held throughout the United Kingdom for mutual support

National Conference and Sibling Workshops

Held annually and offering families the opportunity to learn from professionals and each other

Information Resource

Publishes specialist disease booklets and other literature.

Quarterly Newsletter

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

Bereavement Support

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

Research and Treatment

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

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A WELCOME FROM OUR VICE CHAIRMAN, STEVE BUTLER

As you will see reported in this newsletter this has been a busy and exciting quarter for the Society. The conference '21 Years of Bone Marrow Transplantation for Mucopolysaccharide and Related Diseases' took place on 8 June 2001. On all accounts it was a great success. 19 families attended, with a majority never having participated in an MPS Conference previously. We anticipated 60-70 parents and professionals attending, Ultimately the number was more than double. We were also very pleased to welcome family and professional representatives from Norway, Ireland, Germany, Italy, Austria and the United States further reinforcing our links with Europe and the rest of the world. As is usual at MPS conferences networking between families and professionals went on late into the night and we are very grateful to Dr Charles Peters from the University of Minnesota who stayed the course sharing the American experience of BMT.

As you will have heard in the last newsletter it is planned to launch the 'MPS Video' in November 2001. Filming is well underway and we are extremely grateful to all the families and professionals who have given their time and shared their experiences for the benefit of the MPS families not just of today but of the future. News just off the press is that Jeremy Bowen, who reads the BBC TV Breakfast news, has kindly agreed to do the narrative for our video.

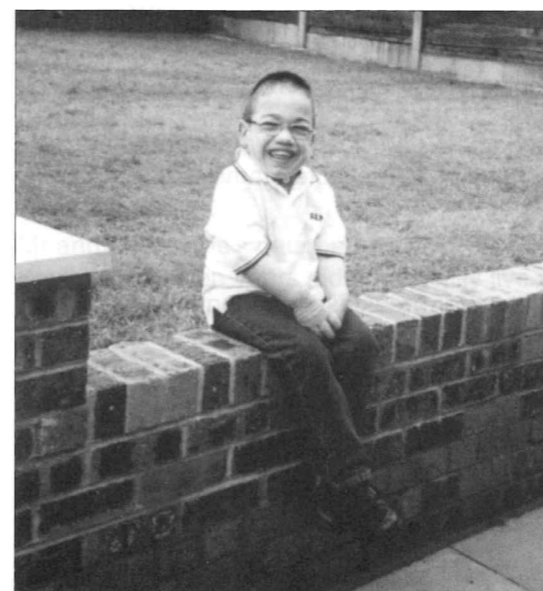
At this point I would like to thank the hundreds of members who have helped the Society in several ways over the last few months. Firstly all the MPS I families who have given so much valuable information on the natural history of their child's disease to

be anonymised and presented to the Food and Drug Administration in the USA. You may find it hard to believe but there is very little collective data on any of the MPS diseases and the Society's Registry of MPS and Related Diseases presents the best source worldwide at present. There are moves to collect such data globally and the Society will be helping in any way it can, but this is still in its infancy. So again thank you to all those that helped in this way. If you still have a questionnaire to complete it is still not too late.

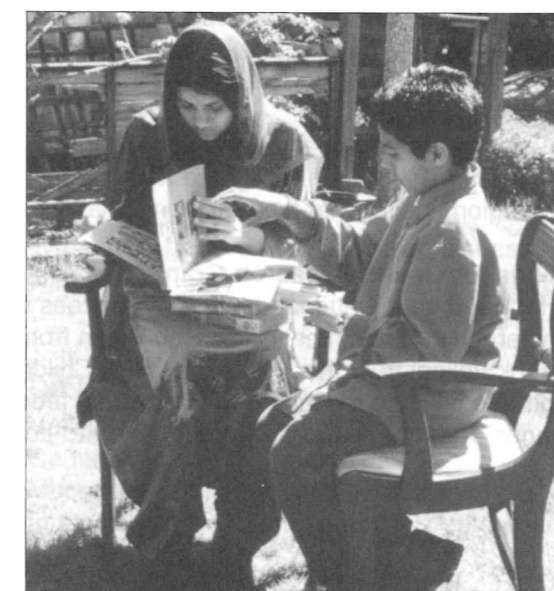
One hundred and fifty MPS mothers of MPS children born between 1993 and 1998 were invited to participate in a social research project to 'Evaluate the effect of the diagnosis of a child with an MPS condition on couples' decisions about having further children' being undertaken by Judy Holroyd as part of her Master's degree in Genetics and Counselling. Again, the response to Judy's questionnaire has been superb and I know she would want us to thank all of you who have helped. It isn't too late to still complete the questionnaire, but do hurry.

Finally we would like to thank all the hundreds of members who have forwarded their membership forms so promptly. Your help with this enormous administrative task is much appreciated by both Trustees and the MPS Staff Team.

Whatever you have planned for these Summer months we wish you well and look forward to seeing as many of you as possible at the MPS Conference 14-16 September in Northampton.



Alex - Maroteaux Lamy



Saira & her brother Taieyyib - Sanfillippo

NEWS FROM THE MANAGEMENT COMMITTEE

The Trustees met in March and considered the following matters.

Staff Changes

Trustees were informed of the appointment of two Development Officers (family support) and an Assistant Development Officer (family support) in addition to a Development Assistant (Research).

Policies

The induction programme planned for new staff was agreed and the Trustees were updated on the Society's occupational health scheme. The Director introduced the MPS Society's draft HIV and AIDS policy for consideration. Amendments were agreed and the policy approved.

Treasurer's Report

In the absence of the Treasurer the Director gave a financial report. She reported that fundraising receipts were extremely encouraging. This was mostly due to one family who had raised over £40,000. The Trustees approved the Society's accounts for the year ending 31 October 2001.

The Society's Constitution

The Chairman asked Trustees to consider the position of Trustees who do not regularly attend meetings. All Trustees agreed that it was necessary to act effectively as a Management Committee. It was proposed to present a motion at the AGM that any Trustee who does not attend three consecutive meetings has to step down. The Director was instructed to approach the Charity Commission for advice on this matter.

Diseases Represented by the MPS Society

The Trustees agreed that the MPS Society include Fabry disease on the list of Mucopolysaccharide and Related Diseases. A Fabry Disease Training Day is planned and agreed.

MPS Promotional Video

The Director informed Trustees that the script to the MPS Video has been agreed and that Christopher Morris of Omnivision is directing the project.

Childhood Wood

Trustees discussed at length the financial risks in the light of foot and mouth disease of running the Bereaved Family Day in the Childhood Wood in July 2001. With regret it was agreed to cancel the Bereaved Family Day for 2001.

Regional Clinics

The Director informed Trustees that families in the South East have asked for an MPS Regional Clinic in Kent. The Trustees agreed that this initiative should be pursued.

Regional Events

Trustees agreed that at least six MPS families must book to participate for an event to be viable. The Trustees also agreed to carry out a consultation with affected families in each of the regions as to what type of events they would prefer.

Jeans for Genes Research Grant

A further meeting attended by three Trustees was held over breakfast following the BMT conference. At this meeting Dr Rob Wynn from the Royal Manchester Children's Hospital, Dr Ashok Vellodi from the Hospital for Sick Children, Great Ormond Street and Elaine Estruch from the Institute of Child Health provided feedback on the first six months of the Jeans for Genes programme grant awarded jointly to London and Manchester.

NEWS FROM THE MPS OFFICE

Naidex Exhibition

On Thursday 17th May, Sarah Pritchett, Andy Hardy and I were given the opportunity to travel up to the NEC in Birmingham to the Naidex exhibition. This is an annual exhibition for organisations who are involved in the provision of care and equipment for people with disabilities. It provided a very good opportunity for the MPS staff to find out about new products on the market, gather up-to-date information and meet exhibitors in person.

The exhibition ran for three days with a number of stalls, demonstrations and seminars running throughout this time. Many different organisations were represented and it was good to get the chance to visit all of them and see what products were on offer.

Jeans for Genes 2001

Thank you to all those families who returned their Jeans for Genes questionnaire. The details you have given on the ways you and your family are able to support the campaign will greatly assist Antonia and Ellie in running the MPS Society's support to the campaign this year. However large or small your contribution it will go a long way to raising much needed funds to support families and adults affected by MPS and research into MPS and related diseases.

New Families

Mr Atkinson's and Ms Topping's son Jack has recently been diagnosed with Hunter disease. Jack is two and a half years old. The family live in the North of England.

Mr and Mrs Coney's son, Thomas, has recently been diagnosed with Maroteaux-Lamy disease. Thomas is five years old. The family live in the North of England.

Mr and Mrs Gallagher's son, Ryan, has recently been diagnosed with ML III. Ryan is nine years old. The family live in Northern Ireland.

Mr Harvey's and Ms Dickie's daughter Brooke has recently been diagnosed with Fucosidosis. Brooke is two years old. The family live in Northern Ireland.

Mr and Mrs Lewis's daughter Kayleigh has recently been diagnosed with Sanfilippo disease. Kayleigh is seven years old. The family live in Scotland

New Baby

Congratulations to Gina and Chris Page on the safe arrival of baby Matthew on 1 July 2001. Gina, the Society's Finance Officer, plans to return to work for the Society following her maternity leave.

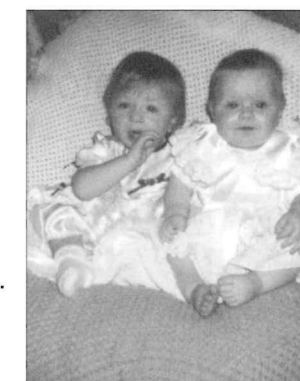
Some we already had knowledge of, but there were some exciting new innovations being demonstrated which we hope will be very useful in the future.

The MPS Society is currently building up a library of information on suppliers of equipment for people with disabilities. Naidex was invaluable in enabling us to gather new ideas so that we can pass these on to families as and when requested. There are so many new products on the market in addition to the old favourites and these can, we hope, only serve to increase the opportunities for people living with disability.

*Antonia Crofts
Assistant Development Officer*

If you are able to support the Jeans for Genes campaign and have not already returned your completed questionnaire then we would please encourage you to do so. Knowing you are supporting the campaign means we can offer you support and ensure you receive all the help you need to make Jeans for Genes day 2001 fun as well as helping raise a record amount of money.

Ellie Gunary - Assistant Director



Brooke & Jade Dickie

Notice of Annual General Meeting

Notice is hereby given that the Annual General Meeting of the Society for Mucopolysaccharide Diseases will take place at the Hilton, Northampton on 14 September 2001 at 7:30pm

A WEEK IN THE LIFE OF THE DEVELOPMENT TEAM

Saturday, VML Family Conference



Vianney Lapointe, MPS II
Hunter, with his dad

While most people are still sleeping Alison and I set off on our journey to Nouan le Fuzelier in France where at la Ferme de Courcimont, VML's family conference is held. We are attending this conference to continue to build working relationships between the staff teams of VML and the MPS Society. Learning how each Society operates and getting to know the staff team is important preparation for next year's jointly run international conference. Thanks to Alison's map reading skills we arrive at our destination two hours after leaving Paris. Despite having listened to French language tapes whilst travelling in the car the previous week my French remains extremely rusty. I reassure myself I can at least say "Hello", "Goodbye", "Please" and "Thank you".

The setting of a farm is ideally suited to the French spring sunshine, which unfortunately evades us this weekend. Having left a beautiful warm sunny day in England it is

something of a surprise to find France cold and wet. At the farm most socialising takes place in the main conference building with the talks and seminars held in a converted barn.

The children's activities because of the poor weather are in wigwams in the grounds rather than outdoors as is usual. Delphine, Jean and Eric who work for VML introduce Alison and I to families and VML's medical professionals. VML supports individuals suffering from a much wider range of lysosomal storage diseases than the MPS Society but despite this supports a similar number of people. We manage to overcome the language barrier to share and listen to experiences. Our ability to communicate is greatly eased by a kind member of VML's board of Trustees who speaks excellent English. After a buffet style evening meal the evening's entertainment consists of traditional dancing. At least French is not needed to understand this.

Sunday, VML Family Conference



la Ferme de Courcimont

Having, as is usual at the MPS conferences reached our beds in the early hours of the morning Alison and I a few short hours later attend Sunday's first session, a seminar for adult sufferers. We are eager to make the most of our visit and learn as much as possible. Even though I am only able to understand the gist of the discussions and not the details I am reminded of how many similar experiences are shared across

countries and cultures. The main area of debate concerns how the lack of information many adult sufferers have about their condition leaves them unable to make informed decisions about treatments offered. Before lunch Alison and I are successful in gaining more data from families for the European Registry and then hurry back to Paris to catch our flight home to enjoy an English evening of warm sunshine.

Monday, Information Day



An Ottobock Buggy

After a busy weekend I am grateful for a 9am start Monday morning. Monday is usually spent in the MPS office with a Development Team meeting held to plan the week's work. Today however I travel north with Andy, a newly appointed Development Officer to present an information day requested by the staff of a residential home where a young girl with Sanfilippo disease is living. After giving an overview of Sanfilippo disease I introduce the MPS Society's model care plan including palliative care and resuscitation policies. Great sensitivity is always needed when introducing palliative care and this is no different when those concerned are a team of residential care staff. Different skills and processes are required when presenting this to a group of twenty rather than on a one to one basis. Back at the office requests for support have

continued to arrive by post and by 'phone. Antonia tells me of a newly diagnosed family who have been in touch and I take their details so I can 'phone them later in the day. She also tells me that a four-month battle for a suitable wheelchair for a child with Mannotosidosis has been successful. The local wheelchair centre had told me that due to lack of funding it could not issue chairs to anyone other than those in the last stages of a terminal illness. Without a wheelchair for Joshua this family are unable to go out as he is very hyperactive and finds walking painful. Without time to spare I had written directly to Tony Blair and was pleased to hear within days from the wheelchair centre that they were reviewing their policy and that a wheelchair would be available for Joshua as a matter of priority. Joshua has now been assessed and the family are very pleased

A WEEK IN THE LIFE OF THE DEVELOPMENT TEAM

with the provision offered. I reflect on how this situation highlights not only the importance of the individual advocacy support but also its limitations.

There is an ever increasing number of families struggling to achieve suitable wheelchair provision for their MPS child or

adult, due in the majority of cases to the fact that models suitable for children with MPS are not standard provision and therefore expensive to supply. The need to address this issue at a national level is becoming very clear and I consider how best to take this forward.

Tuesday, MPS Promotional Video

The second day of filming for the MPS promotional video has arrived. Christine and I meet Christopher Morris (ex BBC journalist), from Omnivision who is leading the production team and travel to Sophie's home. Sophie has recently been diagnosed with Sanfilippo disease and we are to film a retake of the new family visit I had made three months earlier. To make the most of the natural light on what is a beautiful summer's day the film crew consult and we agree to film outside. As the family's garden is rearranged, benches moved, toys brought out and garden chairs wiped down for their first use of the year I try but fail to recall a home visit I have ever been able to make sitting in the garden. Being new to any kind of filming I am very uncertain as to what to expect but my nerves are calmed by thoughts of how much the families involved are giving of their time and energy. In particular Sophie's parents who have agreed to revisit the recent days just after receiving Sophie's diagnosis, a difficult time for all families. There is one star of the day and that is Sophie. She astounds her parents by playing beautifully on the slide and on her bicycle and is not in the least bit phased by a film crew with a camera and sound boom being in her garden.

With plans of making an album of the making of the MPS video Christine takes photographs of each stage of the filming. With Christine holding the camera the funniest shot of the day, Christine falling backwards off Sophie's swing is missed. At least I now have this recorded for posterity in the MPS newsletter!!!!

The next visit is to Taieyyib's home and again

filming takes place in the garden. Taieyyib is an older child suffering from Sanfilippo disease and his parents speak of the difficulties they have had achieving the correct support for their son at school. The star of this visit is Taieyyib's sister, Saira who very eloquently speaks of growing up with Taieyyib, how he used to tear up her homework but how she now attends Grammar school and hopes to be a doctor.

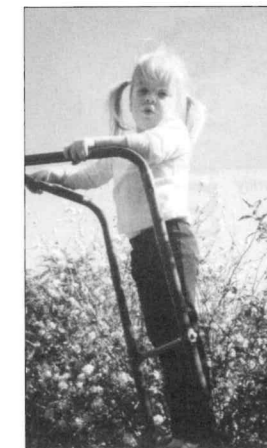
Back at the MPS office we ask those members of staff in the front office if they would mind moving to another office so that Christine can be filmed introducing the MPS Society. Sitting in front of the map surrounded by photographs of MPS children in which a pin represents each MPS child with whom the Society has contact Christine says her piece and filming is completed for the day.

The final hour of the day is spent planning the next stage of the filming, and responding to messages. There are four weeks until the Conference "21 years of Bone Marrow Transplants for MPS and related diseases" Christine reviews with Alex the bookings received and together they confirm that arrangements are running to schedule.

A family with a two year old son with Hurler disease have 'phoned in to ask for support to achieve a supply of nappies from their local Health Authority who are refusing to supply them as Jake is not yet three. I put pen to paper writing not only to the incontinence advisor but also the Chairman of the Health Authority.

information we have on any one issue and identify any gaps.

Christine having heard at the weekend that Hendrick's sister Emily Volk had died is travelling for the second time in nine weeks to Germany, this time with Robin as well as Lucy (her daughter). Hendrick was remembered in the last MPS newsletter.



Sophie



Saira & Taieyyib



Filming at the BMT
conference

A WEEK IN THE LIFE OF THE DEVELOPMENT TEAM

I am thankful that the funeral is today as this is the only day this week that Christine could take leave to attend. Being able to say goodbye and support the many families with whom we have contact to say goodbye is an important part of the advocacy service.

Saying goodbye to only Hendrick and not his twin sister Emily would have been like saying goodbye to half of an inseparable pair. The loss for Christine and her family as well as Hendrick's and Emily's family is also of a place from which the Volk family will now move which holds many treasured memories. My thoughts turn to the Volk family of whom I have heard so much and yet never met, in particular the adjustments ahead of them having spent eighteen years caring for the twins.



Emily

Meanwhile back at the office work carries on as usual. Consultation papers are received

Thursday, Home Visit

I meet with Sarah (Development Officer) first thing in the morning at Heathrow to travel to Northern Ireland. Having triggered an old back injury earlier in the week I am thankful that Sarah agrees to drive. Although I am extremely frustrated not being able to move quickly and easily, I remind myself how small the inconvenience is compared to the difficulties experienced by those suffering from MPS.



Antonia

Considering all the travelling and map reading required in carrying out home visits I contemplate whether a geography degree would be useful. We find the family's home without the benefits of this qualification and spend four hours supporting the family of a recently diagnosed child with MLII. As much as the experiences and emotions shared by families living with MPS can be similar they can equally be very different. Each family and each individual within a family is unique and has his or her own ways of managing and coping. In delivering individual advocacy support it is essential that the focus remains

Friday, Northern Ireland Clinic

After a very busy and successful clinic we travel home. As the week draws to an end my thoughts turn to all the individuals and families who have so readily allowed us to film them, both for the benefit of other individuals and families affected by MPS and related diseases and to raise awareness and promote the work of the MPS Society to a wider audience. I in addition reflect on the

on proposals for a Carers and Disabled Children's Bill for Northern Ireland. Scotland's are expected to follow shortly both expected to be along the same lines as the Carers and Disabled Children's Act recently introduced in England and Wales.

New members of the team are trained to complete DLA forms. Although Antonia and I have become adept at completing three at once in order to meet demand I am thankful that with the new members of staff this should no longer be necessary. An aromatherapist treating a fourteen-year-old girl with Hurler disease (BMT) makes a more unusual request. The therapist is seeking advice on which oils may or may not be used on MPS children. Unable to answer this question I offer to link her with another aromatherapist if one makes contact with the Society.

on the needs of each individual and that nothing is taken for granted.

At the office Antonia is responding to this week's pile of information requests, received from a wide variety of people including students on college courses, medical professionals and interested members of the public. Angela is busy responding to requests for help and information from overseas MPS families.

The evening is spent in Belfast and over a meal with Christine, Dr Ed Wraith and Dr Fiona Stewart we discuss proposals for a twice yearly Northern Ireland MPS clinic. The number of patients, eighteen on the clinic list for the following day makes for an extremely busy clinic. With the large number this year and a potential of forty for a Northern Ireland MPS clinic it is agreed a six monthly clinic is needed. Christine takes the opportunity to brief Fiona and Ed about their roles in the MPS video filming the next day.

different situations and cultures we encounter each week. This week I have visited a family from the Asian community for whom English is their second language, a family from the travelling community and many other families with different values and backgrounds.

Ellie Gunary
Assistant Director

INTERNATIONAL CONFERENCES

New Insights and Future Perspectives, 27-28 April 2001 Seville Spain

Over 300 leading physicians, scientists and patients from 30 countries discussed their research and clinical experience at the 1st International Symposium of Lysosomal Storage Diseases, Fabry Disease: New Insights and Future Perspectives, held April 27-28, 2001 in Seville, Spain. By bringing



Angela Ratcliffe and Christine Lavery with the two posters written for the Seville Meeting by the staff team. The posters will be on display at the MPS conference in September

together the global community of researchers and clinicians dedicated to Lysosomal Storage Diseases with emphasis on Fabry disease, the congress advanced the collective understanding of these devastating and often neglected diseases. The symposium followed the EU's Committee for Proprietary Medicinal Products (CPMP) positive opinion recommending approval of Replagal™ (agalsidase alfa) and Fabrazyme™ (agalsidase beta) enzyme replacement therapies for the treatment of Fabry disease and was sponsored in part as an educational initiative by the Lysosomal Storage Disease Network (LSDN).

"This is truly a hopeful time for people living with Lysosomal Storage Diseases and the physicians and scientists who have dedicated their lives to unlocking the mysteries of these rare genetic disorders. By sharing their experiences from around the world, Lysosomal Storage Diseases experts participating in this symposium made great strides in improving our understanding of Lysosomal Storage Diseases. Combined with the anticipated approval of enzyme replacement therapy in the EU for patients with Fabry disease, we have good reason to be optimistic about the future for patients with this disorder." Roscoe Brady, MD, Chief, Development and Metabolic Neurology Branch, National Institute of Neurological Disorders and Stroke, National Institutes of Health (NINDS/NIH)

The symposium continued with presentations on Fabry and Hunter (MPS II) diseases, ranging broadly from epidemiologic studies to the future of gene therapy. A number of significant themes reverberated in the talks and the poster sessions. Following are highlights of the scientific program:

Fabry Disease

End-stage renal failure is the most common outcome for men with Fabry disease. Mortality and morbidity result from deposition of globotriaosylceramide (Gb3) in the kidney predominately seen in epithelial and mesangial cells in the Bowman's capsules. Lesions are progressive and lead to clinical manifestations of proteinuria and decline of glomerular filtration rate (GFR).

A broad and continuous clinical spectrum of disease exists, with some individuals showing few symptoms and others living with severe manifestations including profound renal and cardiac involvement.

Carrier females may exhibit symptoms of



Fabry disease, yet appear to experience onset of symptoms later in life. Additional studies are needed to learn more about the mechanisms underlying the varying symptomatology of women who are heterozygous for Fabry disease. In an important clinical study conducted in London, Sachdev (et al) found that 4% of male patients presenting with late-onset left ventricular hypertrophy (LVH) also had an a-galactosidase (a-Gal) level that was <1.8% nmol/hr/ml. This low level of a-Gal is considered diagnostic for Fabry. Therefore, atypical cardiac variations, such as LVH, manifested by some people in middle age may be the sole symptom of Fabry disease and may be a key to expanded patient identification.

Christine Lavery - Director

The Society is grateful to TKT for funding Christine Lavery and Angela Ratcliffe to attend this meeting.

INTERNATIONAL CONFERENCES

Strategies for Therapy of MPS and Related Diseases 16th Annual MPS Conference 21-24 June 2001 Los Angeles USA

Mucopolysaccharidoses (MPS) and Hunter Syndrome (MPS II)

Mucopolysaccharidoses (MPS) encompass several types of diseases, including MPS II or Hunter syndrome. Hunter syndrome is an inherited rare genetic disorder caused by deficient activity of iduronate-2-sulfatase (125). Patients affected by Hunter syndrome suffer from a variety of symptoms including skeletal deformities, obstructive airway disease, cardiac failure, and, in severe cases, neurologic decline can occur. New treatments such as enzyme replacement therapy are currently being studied.

The importance of sound epidemiological data was exemplified by the database created and maintained by the European

Registry of Mucopolysaccharide and Related Diseases run by the UK MPS Society.

Hurler Syndrome (MPS I)

Principal Investigator Muenzer (et al) from the United States reported the effect of enzyme replacement therapy with recombinant human alpha-L-iduronidase over a two-year period in 10 patients with MPS I. Muenzer found that using this regime liver volume and urinary glycosaminoglycan excretion decreased significantly. Episodes of apnea and hypopnea during sleep also decreased and amelioration of some clinical manifestations of the disease were reported. In pediatric patients, the rate of growth in both height and weight increased.

European Alliance of Genetic Support Groups 18-19 May 2001 Vienna, Austria

"Putting Patients at the Heart of Genetics"

Having joined the Society I was delighted when I was asked if I would attend a two-day conference in Vienna. The EAGS (European Alliance of patient and parent organisations for Genetic Services) had been given a grant to offer 40 subsidised places for their conference. Having a job in research I thought this was going to be very useful for me to attend.

The topics of the meeting were

- Orphan Drugs and Rare Genetic Diseases
- Ethical and Lay Aspects of the Use of Knowledge of the Human Genome
- Genetic Databases and Common Genetic Disorders
- Genetics and Science Communication
- Consensus Meetings

I thought the MPS members may be interested in a precis of the presentation by:

ELIZABETH MITCHELL - GENETIC DATABASES AND COMMON GENETIC DISORDERS

In the UK the Medical Research Council and the Wellcome Trust are planning to create a DNA database, taking samples from 500,000 volunteers aged between 45 and 65. These DNA samples will be analysed and variations

linked with volunteers' subsequent medical histories in order to identify genetic factors significant or potentially significant in a wide range of common diseases. This collection will be one of over 300 genetic databases in the UK most of which have been set up to look at narrower, more specific issues such as research into gene-environment interactions, or evaluations of particular medical treatments. It will be the largest and in setting it up it is intended to create a resource that will stand the test of time, that makes the best use of UK expertise in genetics, epidemiology and statistics and which capitalizes on the nationwide organisation of the UK National Health Service.

Setting up such a study demands that careful consideration is given to practical, ethical and legal issues as well as scientific and clinical ones if public confidence and support is to be maintained and sufficient volunteers are to come forward and makes the project viable.

Discussion of the issues raised by the proposal has been wide-ranging. Central to the success of the project is the ability to link individuals' medical histories with their DNA sample, yet the fact that this is by definition a research study not a clinical service means the value of any risk factors identified will at least at first be uncertain, creating ethical difficulties in going back to individuals. In an

INTERNATIONAL CONFERENCES

Australian study of breast cancer, volunteers were given the option of having feed back in the event of possibly significant information emerging. In such a case they were advised that a mutation had been detected in their family and it was suggested that they see a clinician under normal clinical service conditions to establish what it was and what, if any, significance it had. This may be feasible in a narrow use data bank (for breast cancer for example) as the fact of joining the databank indicates a knowledge of a possible risk in the volunteer. With a wide ranging study such as the Wellcome/MRC one, where investigations will be over a wide range of disease links unspecified at the commencement of the programme, this is more problematic.

Clearly the consent of volunteers will be essential prior to them joining the data bank. Any consent will have to be wide ranging and because there will be no direct benefit to the individual (particularly in the absence of direct feedback of results) participation will be for altruistic reasons. It is not clear that it would be possible to obtain legally valid wide ranging consent for future (unspecified) uses in all European countries. Indeed social and cultural factors mean that, in some of the EU's member states, there might be a great reluctance to participate in any long term data banking of genetic information or, if they were willing, then they might want to restrict the areas of investigation in the some way. For example, volunteers might be willing to have genetic links to cancer or heart disease investigated, but not mental health problems where there is a fear of stigmatization.

Given that the purpose of DNA data-banks is to improve health care and to aid in the development of new drugs there will be the prospect of a commercial return arising in respect of some parts of the database as the information is used by pharmaceutical companies to develop new drugs. Individuals contributions to the bank will, in isolation be of little value. It is only when they are taken in aggregate that they become statistically significant. Nevertheless considerations should be given to some kind of "stake-holder benefit" being provided in the form of non cash added value.

One possible outcome of such a large scale venture might be the normalisation of genetic information and the realization of the actual role of genetic factors in health and well being for most people - scaling down fears

and expectations based on popular misconceptions about the power and penetrance of genetic diseases in the majority of families.

In all research involving human subjects the question of consent is central. Ideally people participating should give fully informed consent to all aspects of the study, but in practice this is often an unattainable ideal except in narrowly focused studies with a limited range of options and end points. Rather more feasible is that participants feel content with the consent they have given and that they have had the opportunity to inform themselves to the extent that they wish. Many people, particularly those with experience of the real consequences of genetic diseases, feel that there needs to be a balance struck between allowing achievable progress quickly and the power given to other people's fears of abuse, even if they are remote. One option is to impose severe sanctions for unauthorised use of the data-bank and police all users rigorously.

The Conference was very informative and I found the Networking very useful too. I would like to thank Alistair Kent, the Director of the EAGS, for making it possible for me to attend and for his warm welcome. If anyone would like a copy of the whole report please send a s.a.e. (A4) with a cheque for £2.00 payable to MPS to cover copying and postage.



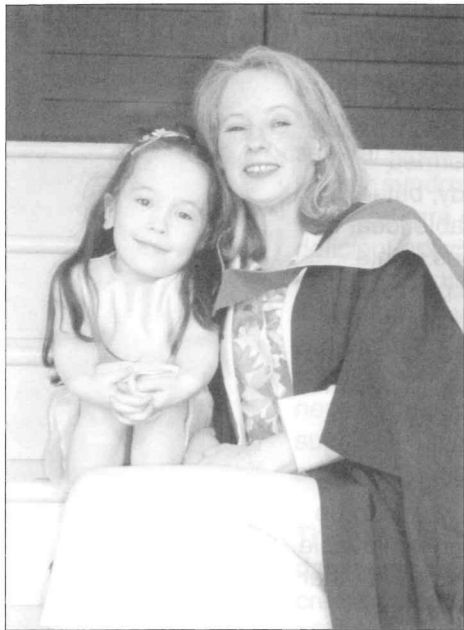
- EAGS -

European Alliance of
Patient and Parent Organisations
for Genetic Services and
Innovation in Medicine

Alison Britton
Assistant Development Officer - Research

FAMILY NEWS

Bernie Drayne achieves Honours Degree



Bernie with proud daughter, Roma - Morquio

Saturday, May 19th was a very exciting day for us all when we attended the Open University Graduation Ceremony at Belfast's Waterfront Hall to receive my award of a 2.1 Honours Degree in Social Policy. My husband Kevin and our three children, Roma, Kilian and Peter were all there to mark the occasion.

The degree took 6 years to complete, and little did we know what was to happen us along the way. Three weeks before the end of the second year exam, our 3 year-old daughter Roma was diagnosed with Morquio. We

were all shattered as we tried to come to terms with it. Life just seemed to stop. Without the encouragement, pushing and support from Kevin, the children and my sisters, I would not have had the heart to continue my studies.

Roma's brother Kilian who was 1 year old at the time of Roma's diagnosis had to be tested for Morquio in the week before that second

Putting Jade in the Picture

Our first team squad were so touched by the plight of two year old Jade Hall that they just had to brighten up her life.

The Beeston toddler suffers from a rare and incurable syndrome and for parents, Simon and Nicola, every moment with her is a precious one.

The couple's young son, Corey, also suffers from the same condition and Simon wanted to buy a camcorder to record the youngsters' early years on film.

Unfortunately, Simon is unemployed and as a life-long Forest fan he asked the team for signed prizes for a fund-raising raffle.

year exam. Whilst others were working hard on their revision, we were chasing Kilian around in an effort to gain a urine sample but we were eventually successful.

I had to go into the examination hall on the morning of the exam worrying about what the result would be. One hour into the exam an invigilator walked up to my table and passed me a little note with a telephone message simply reading 'Your son is OK'. I'm sure she had no idea how important that message was! I shall never forget that moment.

It's been a very long, hard but worthwhile six years. Studying has been very therapeutic especially when coping with Roma's diagnosis. I have gained in confidence, and that's something that us MPS parents really need in our many struggles with authorities and some health professionals. My experience has also encouraged Roma to become a bookworm. She would often remind me to do my homework after I had reminded her to do hers.

The midnight oil has burnt frequently and I have taken the opportunity to study on hospital wards and waiting rooms with Roma at her many appointments. I, for one have been thankful for long waiting times!

When the players got to hear about the situation they insisted on having a whip round and presented the family with a video camera.

Jade and father Simon met with Dave Beasant at the ground and Simon said: "I couldn't believe the players had bought the camera themselves. I just can't thank them enough for what is a marvellous gesture."

And Bes added: "Alan Rogers got to hear about the family's situation first and when he told us, we all wanted to help. I just hope the camera gives them a lot of enjoyment."

This article appeared in issue No. 25 of the Nottingham Forest Matchday Programme.



Jade featured in this article suffers from Hurler Disease, as does her brother Corey.

FAMILY NEWS

My name is Sue Stuart, I'm married to Peter and we have three gorgeous daughters, Jessica 10½ (Hurler), Hollie 6½ and Annie 5.

I thought I would write a short article to encourage MPS parents to achieve their ambitions and not put them on hold because they have an MPS child.

For as long as I can remember, I have loved motorbikes but never did anything about it, especially when Jessica entered our lives. Any ambitions I had, I felt that they had to become



dreams, as I had no chance of realising them, as Jessica was about to take up 110% of my time. By the time Jessica turned 8, I realised that I had to do something for myself and not worry about the girls, they had had my undivided attention (Jessica permitting) all their lives, It was also an age thing (approaching 40).

I booked up a motorcycle direct access course, which guaranteed that I passed my test and would be able to ride any cc motorbike. I didn't need to be an owner of a bike so all I had to do was go on a spending spree (heaven) to get all the right gear! I was able to do the course at my convenience with extra tuition if required. I failed my first test because I put my foot down while executing a U-Turn. Do you see many bikes doing U-Turns? I now know why. I passed my second test in June 2000 and was absolutely overjoyed. I bought a second hand Kawasaki ER500 (the same model bike I was taught on). It was a dream to be able to go for a ride, my bit of freedom from the stresses of having an MPS child, not to mention the other two!

With a bit of juggling with the childcare with Peter, I embarked on several more courses to improve my riding skills. I met lots of people like myself who were "late bikers", also several police motorcyclists who gave valuable information about riding safely.

We discussed many things including bikes that were suitable for the "shorter woman" (me). My next purchase was a set of leathers (jacket & trousers) and a brand new bike - Kawasaki GPZ500 in black & red with a matching red helmet (of course). (See photo).

I now had been bitten by the bug and was itching to do more so I joined the Cambridge branch of The Institute of Advanced Motorcyclists (IAM). They teach you to become an advanced rider by allocating an observer (an advanced rider) who rides with you and gives you feedback on your riding skills. After more discussion about various bikes I have finally found my bike for life, I'm now on my third bike, a Kawasaki ZZR600 - a big boys' bike (lubberly jubberly!). Its silver & bronze so I have had to have a revamp of my gear and now have a super dooper helmet in silver (must have matching accessories!).

I am now at the stage of waiting to hear when my test date is. In less than a year I've had three bikes and am almost an advanced rider but it doesn't stop there, I plan to do my RoSPA gold test with the intention to achieve a good enough grade to enable me to become an instructor.

My next ambition is to learn to fly a plane and or scuba dive But that's another story!

Peter has also realised an ambition apart from his golfing ones. He is now a driving instructor with his own school called "Relax Driving School".



(See photo). We plan to be able to go into partnership teaching both cars and bikes, this is likely to come to fruition once Jessica has passed away.

Caring for a special needs child may use up most of your energy but you also need space and time to do your own thing. Having Jessica is not a prison sentence, she is a very valuable part of my life experience. Your child, your family and most of all, your own self will benefit if you continue to develop as

FAMILY NEWS

Fucosidosis, Bone Marrow Transplants & Family Life

In 1992, we had two children and were told that both had Fucosidosis, a rare lysosomal storage disorder, related to the mucopolysaccharide (MPS) diseases and characterised by the lack of the alpha-fucosidase enzyme. Our elder son, John was three and a half years old, while his brother, Christopher, was only aged six months. John had difficulty walking and had developmental delay. He was already receiving hydrotherapy as well as physio and occupational therapy to keep him supple. Baby Christopher, in comparison did not exhibit any symptoms.

This diagnosis was very distressing news but we wanted to help them. Enzyme replacement and gene therapy were still 'dreams' of the future. A bone marrow transplant (BMT) was considered but this operation had not been performed before for a person with Fucosidosis. It was decided that John was too old but Christopher was of the right age because he would benefit from stopping further damage to his brain and nervous system. We were told that the operation would not correct the problems that would develop for his bones and joints.

Various family members were compared to see if we were a good match for a BMT. Unfortunately, no one was suitable and the search commenced for an unrelated donor, which is greatly helped in the UK by the Anthony Nolan register - a database of thousands of people. A match from an unrelated donor was found and Christopher undertook the operation in March 1992. Figure 1 shows him in the Westminster Children's Hospital. Some three months later, he was allowed to leave and we cared for him and his brother at home.

John's condition deteriorated and he lost his speech, had fits and had to be fed by a nasogastric tube. This continued day and night and was very emotionally and physically tiring. John died in October 1994. Meanwhile, Christopher became stronger and was able to achieve more than his brother e.g. walking and feeding, unaided. Numerous tests were undertaken and it was shown that Christopher's IQ was low. Since starting school, he has had an Educational Statement, - a legal document which defines his special educational needs. He is very happy at school and is able to communicate with a few words and by makathon signing. He particularly enjoys numbers and will tell us the numbers he sees. Figure 2 shows Christopher in 2001, aged 9.

Fucosidosis is a very rare condition and we have yet to meet any family with the same diagnosis, although we have spoken to or been in e-mail contact with families all over the world. In the UK, there are many families who provide complementary support because their child has another MPS condition, such as MPS 1, Hurler or MPS VI Maroteaux Lamy Disease.

On 7 June 2001, a British Charity, the 'Jeans for Genes' appeal provided funds for a one-day conference entitled '21 years of Bone Marrow Transplant for MPS and Related Diseases' which was organised by the UK Mucopolysaccharide Society (www.mpssociety.co.uk). A medical specialist from America as well as clinicians from the UK were able to summarise their experiences. We heard that scientists are making some progress with enzyme replacement and gene therapy, and in the long term BMTs will become a 'dinosaur' treatment. However, the new techniques are still a distant dream.

Participants included parents, several children or adults that had a BMT after being diagnosed with Hurler Disease and staff from all the major UK hospitals involved in this work. Professor Jack Hobbs, who in 1980 undertook the first MPS transplant was able to explain how he had undertaken these transplants and all the clinicians spoke about their lack of knowledge, 'the fog', as they faced the unknown. It was also demonstrated that not all MPS related disorders are helped by a BMT. If the treatment is delayed, the BMT is unable to make a difference to organ damage and progression of the disorder.

Christopher has been lucky with not needing extensive surgery after his BMT. Whereas spinal bones and limb joints are likely to need surgery for Hurler children. Christopher has had a series of MRI scans to check his health and as the years have passed, he has required less medical attention. Because he is unstable on his feet, he requires supporting footwear and to combat infection, he receives a twice daily dose of a prophylactic antibiotic.

Christopher is usually a very happy boy, who always wants to be involved with people and his school work. He loves describing or signing what he sees or wants and has been described as 'delightful' by the clinicians.
Paul & Jeans Leonard

FAMILY NEWS

helping him. Certainly, he has become more tolerant of medical tests and even puts up with having his hair cut!



fig 1. Christopher, aged 9 months, undertaking a Bone Marrow Transplant for Fucosidosis in 1992.



fig 2. Christopher enjoying life, 8 years after his transplant

Free Software for Children with Learning Disabilities

My name is Ian Bean, my family have been members of the MPS Society for many years although we do not attend any meeting or conferences since our son Paul died of Sanfillipo 5 years' ago.

Since Paul's death, I have trained as an ICT teacher and am working in a school for children with severe and profound and multiple learning difficulties in Middlesbrough.

The reason I am sending you this is just to let know about a project I have recently completed which may be of some interest to other members. Over the last couple of months I have worked on the design and implementation of a suite of software for children with PMLD for the Northern Grid for Learning (NGFL).

The software can be used on any PC or Macintosh, is accessible via keyboards, mouse, switches and touch screens. It is progressive offering 140 activities ranging from experiential visual and auditory stimulation through cause and effect, switch building, timed activation and simple scanning all linked to the National Curriculum P levels

for students with special needs. It comes complete with documentation, teaching notes and skills checklists and more importantly it is completely FREE.

The software can be run online or downloaded and used with standalone computers, running in a web browser. It is written 100% in Macromedia Flash giving full screen, high quality graphics and sound effects.

You can run or download the program from... <http://www.northerngrid.org> The program can be found in the curriculum resources/SEN section. The program requires a Flash 5 plug in to be installed which can be downloaded for free at <http://macromedia.com/downloads/> I hope members might find it useful and would be pleased to hear any feedback, good or bad, so we can evaluate the effectiveness of the project.

Very Best Wishes to everyone. - Ian Bean

School Web Site.
<http://freespace.virgin.net/school.priorywoods/>

CLINICS

Manchester - BMT Clinic Friday 30th April 2001

After having spent such a great day with the ERT Trial patients the previous day (Thursday) I was looking forward to meeting the families who were to be attending the BMT Clinic. I wasn't quite sure what to expect as I'd heard the clinics could be quite hectic at times.

As people started arriving, the morning went very smoothly and appointments were running to time. Then came the afternoon which was a bit closer to the 'hectic' I'd been hearing about. It was great to see all the children having so much fun together whether it was having car races up and down the corridors of the Willink, playing with the plastic blocks or just having a go on the computer. The parents also took up the

chance to catch up with old friends and also make new ones. All the children seemed to be making good progress which is great news.

It was a pleasure to meet everyone who attended and I really enjoyed talking with the parents and the children and hearing their views. I would like to say a special thank you to everyone that filled in one of the Information Sheets I sent out or gave up time at the clinic to go through one.

On behalf of all the BMT families and the Society I would like to thank all the staff at the Willink Genetics Unit who made the clinic possible.

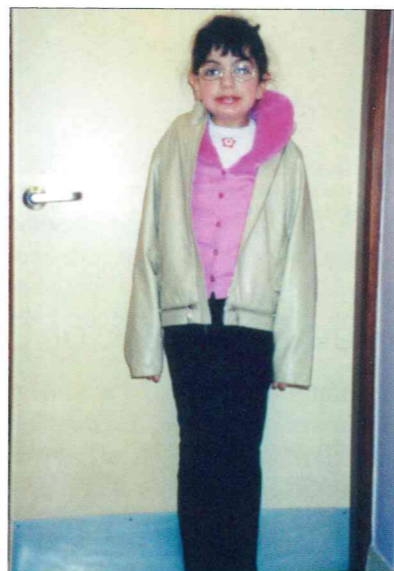
Alison Britton - Assistant Development



Mohammed - Maroteaux Lamy (BMT) and sister, Aliya - Maroteaux Lamy



Alex - Maroteaux Lamy (BMT)



Emma - Hurler (BMT)



Matthew - Hurler (BMT)



Callum - Hurler (BMT)



Issac - Hurler (BMT)

CLINICS

Northern Ireland - MPS Clinic Friday 11th May 2001

This year the Northern Ireland clinic had a different element, it was filmed for the MPS video. This added to what already planned to be a very busy day for Sarah and I who had eighteen families to see, seven of whom had newly diagnosed children as well as ensuring the appointments ran like clockwork. The setting of the Hilton Hotel, Templepatrick offered open countryside and a golf course. As soon as the film crew rescued their equipment after it jammed the hotel's revolving doors they set up outside. In between appointments, the children and adults were filmed playing tennis, golf, reading and drinking at the hotel's bar. Filming at the bar upset a member of the public who was extremely concerned about being filmed. We assured him that we did not want him in the MPS video any more than he

wanted to be in it and James, pictured below continued with his drink. Dr Fiona Stewart from City Hospital, Belfast with Ed Wraith at her side spoke about the role and importance of the MPS regional clinics and two families allowed filming of their consultations. The day was hugely successful with much talking and sharing of experiences. Sarah and I supported families with their advocacy support needs, which included DLA forms and achieving home adaptations. With such a large clinic with so much going on we were very relieved at the end of the day that everything had run smoothly. We continue to be very grateful to Ed for supporting the regional clinics and to Fiona Stewart for all the support she gives both the families and the MPS Society both within Northern Ireland and further afield.



Aiden - Morquio



Ryan - ML III



Jade - Sanfilippo

Scotland - MPS Clinic Thursday 7th June 2001

The Scottish MPS clinic was held for the first time this year at the Royal Hospital for Sick Children, Yorkhill, Glasgow. Dr Peter Robinson, the local consultant who supports this clinic had been able on this one off occasion to arrange for the use of this hospital and an empty ward. Although a little sparse there was plenty of space which many of the children made the most of running around and making plenty of noise.

Nine children were seen in total by the two consultants, Dr Peter Robinson, from Glasgow and Dr Maureen Cleary from the Royal Manchester Children's Hospital. The late start, caused by the inevitable problem at most hospitals of people being unable to find parking spaces was made up for during lunch and everything ran smoothly. Our thanks go to Dr Peter Robinson for supporting the Scottish MPS clinic and for kindly this year

securing our use of facilities at Yorkhill, as well as to Dr Maureen Cleary for travelling to Scotland.

At the clinic I was able to work closely with both doctors and one family in updating their child's care plan and support families to consider any medical issues before their consultation. Feedback has already been received from families attending this clinic and I am sorry to disappoint the family who suggested improvements could be made by supplying an expensive three course meal, alcohol and a babysitting service. The National Lottery Grant we were very pleased to receive which funds the clinic as well as the advocacy support service offered in Scotland does not cover these items. In planning next years clinic all feedback will be taken into account whenever possible. *Ellie Gunary - Assistant Director*

BMT CONFERENCE



A speech by Sue Peach at the Gala Dinner Ladies, Gentlemen and Honoured Guests, Last Sunday Christine 'phoned David and me to ask if one of us would feel able to say a few words about Professor Hobbs. We were honoured to be asked and delighted to do so. David agreed that I should be the one to stand up here tonight, I do tend to be the talkative one of the family, so here I am. This morning I was hoping that the dinner would not be too late as I was at the general election count in Rugby until the early hours this morning. So forgive me if start to nod off, or given the subject, become a little emotional. I would ask Christine to nudge me if I do start to wilt but I happen to know that she was also glued to the set watching the results come in so I'm not sure she would be much help.

Our family first met Professor Hobbs in March 1983. Our daughter, Elisabeth, had been diagnosed as having Hunter's syndrome just a few weeks before, and our lives since then had been a whirl of hospital appointments around the country: Leicester, Cardiff, Great Ormond Street and finally Westminster Children's Hospital. It was in this hospital and with this doctor that we knew Elisabeth would have her only chance of a more normal and longer life. Dr. Rosemary Stevens (formerly Consultant Paediatrician, Great Ormond Street Children's Hospital) had helped us to get that far, but we knew that Professor Hobbs was, as our son Tim would say, the main man.

I remember walking down the corridor to meet Professor Hobbs, Elisabeth running ahead of us. Once inside his office and the formalities over he said, "Well, Elisabeth definitely has the severe form of Hunters." Right, I thought, nothing like getting straight to the point. But that was what almost all of the parents who meet Professor Hobbs were grateful for. His no nonsense, straight to the point, no pussy footing about attitude. By this stage parents wanted facts, options and the truth no matter how unpleasant, this is what Professor Hobbs gave us. He explained clearly the process involved in, firstly, being accepted on to the bone marrow transplant programme and secondly what the transplant would involve, not only for the patient but for the whole family. He was absolutely clear that this had to be a whole family decision and gave us the reasons why. At last we felt that we could make an informed decision as to whether or not we should still opt for a

transplant. Up until then our views had been based on emotion. We would do anything to save our daughter. Professor Hobbs made us think far more carefully about our decision and made us realise that we really did need to talk to all of our extended family as it would certainly affect them as well. We appreciated the way in which he treated us as intelligent people. He did not patronise us and he answered all of our questions with complete honesty. Elisabeth was accepted on to the waiting list. But we knew, that unless a suitable donor was found quickly, she might not be on it for long, because at the time Professor Hobbs was the only doctor prepared to carry out transplants on children with MPS. On top of this, the process was still fairly experimental and there were too many children who needed the treatment. We knew that Professor Hobbs and his team had to make heart-breaking decisions each Tuesday about which children could receive possible life saving transplants and which children were, quite literally, being condemned to death.

We know that this was very difficult for Professor Hobbs, to know that you can save lives but to be fettered by limited resources is an intolerable position to be placed in. We also know that for many members of the medical profession they still have to make these decisions. We do not envy them.

Sadly, Elisabeth died following two transplants, both of our sons being donors. It transpired that the disease had affected her internal organs more than had been expected. However, whilst on Gomer Berry Ward in Westminster Children's Hospital she received nothing but the highest level of care from Professor Hobbs and his team, and as parents we were made to feel part of the team, kept fully informed of everything that was happening and consulted over some of the decisions that had to be made.

Thankfully many MPS children did survive and it is thanks to Professor Hobbs' determination that bone marrow transplantation could help our children that they did so. I've heard it said that Professor Hobbs is a bit of a maverick and a little on the arrogant side, surprising I know. But if being a maverick means pushing against the establishment to get things done, then I'm all for mavericks. And if being arrogant means that you have the self belief to take risks to

A Tribute to Professor Hobbs

BMT CONFERENCE

save others, then I'm all for arrogance.

Professor Hobbs took that first step towards finding, not a cure, but a means of extending life expectancy and improving the quality of life for our MPS children.

Yes, it was risky and experimental but without that first step, without that belief that something could be done to help children who were otherwise condemned to a slow but certain early death, we would not be here today. And without that first step, I believe it would have taken much longer for the medical profession to consider other ways of

helping our children, and others with genetic diseases, through such methods as enzyme replacement therapy and gene therapy.

Although Elisabeth died, we will always be grateful that Professor Hobbs at least gave her the chance of a better life. Many parents, children and members of the medical profession have benefited from his expertise, dedication and determination to push at the boundaries of medical science to help others. Professor Hobbs, on behalf of the MPS Society, we thank you.



Paul Franklin, who presented Professor Hobbs with flowers at the Gala Dinner



The Audience at the BMT conference



The Panel at the BMT conference

Conference Pictures

BMT CONFERENCE



Aiden and Angela Brown, Angela gave this talk at the conference.

Aiden was diagnosed on the 4th of December 1996. He was nearly 10 months old. We were told that his only chance was a bone marrow transplant. We were devastated. The whole time this doctor was telling us our lovely baby was dying before our eyes, I kept looking at Aiden not wanting to believe that this was happening. To me he looked fine, just some small problems or so we thought, but when you put them all together which we had never done before that point, it was very frightening. I was very angry with myself at not being more persistent with the doctors that were looking after Aiden's care. I should have made them do blood tests a lot sooner than they did but at least we might have a chance if we can get a bone marrow transplant. We were flown down to Manchester to meet the MPS doctor, a Dr Ed Wraith. We all went. When we got to the Willink we met Ed and I nearly had a fit. I was expecting the wee doctor with the white coat and we got Ed! Ed was brilliant he explained everything to us again and asked us whether we wanted to go for transplant. I was very sure of my answer, yes. I don't want to lose Aiden without a fight so it was decided that we would have the test. Myself, Jon, Ashleigh and Aaron had our blood tested. Then it was back home to wait for the results. We had about a weeks wait, and were told the results were looking good myself and Aaron providing a match. When we were due to go back to Manchester for the last blood test, the day before flying down Aiden was covered in spots, he had chicken pox. I could not believe it. We had to cancel and then had an agonising 4 week wait to let the infection go out of his system before we could do the next blood test. When at last the test was done we got the news that Aaron was a perfect match and that I was not a suitable donor match.

We sat the kids down and tried to explain to Arron that he was a match. By this time the kids already knew that Aiden had Hurlers and knew everything they wanted to know on the subject. Aaron was only 3 and we were very mindful that Aiden might need two transplants so we did not want to scare Aaron. We told him that if he gave Aiden some of his special blood he would help Aiden a lot. I did not want to tell Aaron that it would save his life because he still had to get through the treatment and we were given a one in twenty chance of loosing him. I did not want to put on Aaron the fear of loosing Aiden. The pressure was great and I did not want any of the kids thinking it would have been thier fault.

When Aiden was admitted to hospital the first thing to be done was his MOT. They checked every part of his body from brain scans to development assessments. It was very hard to listen to all the problems one by one. Aiden had his Hickman line fitted and he got grommets fitted at the same time. Then we were ready for transplant.

We moved into the transplant unit at the beginning of June and Aiden started the 10 day course of chemotherapy. I was terrified. We brought Aaron and Ashleigh with us to Manchester and they stayed with us throughout the transplant. I did not want to send them away being very aware that the transplant might not go to plan. I had convinced myself that Aiden would die, so I had to have every angle covered. His brother and sister had to be there. It might be the last time we would all be a family.

Aiden of couse proved me very wrong. The chemotherapy was very bad and made him feel terrible but he still managed to get into mischief. His best trick was to disconnect the drips from his Hickman line and watch the blood pour out. He found this hilarious. By about day five Aiden was sleeping a lot of the time and had very bad diarrhoea. The transplant day came very quickly.

Aaron came into hospital that morning he was very excited. I had done my ground work, he had been taken to theatre and we had discussed the BMT every day. He new everything that was going to happen to him. We sat with him waiting for his theatre slot. I asked him what he wanted for being such a good boy, sharing his special blood with his wee brother. He asked for a drumstick lolly pop from the shop. We were gob smacked. If that had been his sister, Ashleigh, we would have been bankrupt. Aaron was only away about an hour and a half. He was wide awake when he came back from theatre and all he wanted was chicken and chips, he was starving. He was chuffed to bits because he had a drip bag up was just the same as Aiden.

Aiden got the bone marrow the next day. I felt sick, I kept thinking he would suddenly reject and we would have to do the whole thing again. They managed to get just over a pint and a half of bone marrow which was brilliant. The actual transplant was over in half an hour. Aiden drank his bottle and fell asleep totally oblivious to all excitement.

'A Parents Perspective'

BMT CONFERENCE

After transplant it was all down to Aiden. The hard work had started. We were playing the waiting game. Everyday we were waiting patiently on the blood results to come back. It was a bit like death row. Aiden's bloods were slow to start with but when the blips started to appear on the print outs he was no time at all picking up. We only had one hairy moment. Well maybe not hairy his hair was long gone, but his potassium levels fell very low about day 5. He was put on a heart monitor but he came through that fine.

Aiden was amazing through out his transplant and we got to take him home on day 21. Our health authority had rented us a house just up from the hospital so we stayed there just to be close by. Aiden did not need a blood transfusion, platelets or any TPN feed which was brilliant. He just slept through the worst

of it.

Twenty eight days after transplant we were back in Scotland and starting all over again. It felt like we had a new baby. Aiden's clouding of the cornea had cleared, his skin was as soft as velvet, he had new nails, he could hear and was starting to make sounds. We were delighted. The frightening thing was being so far away from Manchester if he was taken ill. Thankfully that did not happen. Aiden thrived in transplant and I have not regretted it.

I feel we got our lottery win getting a perfect match for Aiden. We have been so lucky. I am glad I have my cheeky wee boy to keep me on my toes.

Angela Brown



Dr Charles Peters from the University of Minnesota USA speaking on, 'A Historical View of BMT'.



Dr Uma Ramaswami & Dr Fiona Stewart



Professor Anthony Bron, 'Corneal Transplants for MPS I'.



Mr Esmond Fogarty and Professor Bryan Winchester

Conference Speakers

BMT CONFERENCE

BMT Conference, Children's Programme

Friday 8th June was a busy day for all the MPS Society staff because of the Bone Marrow Transplant Conference at Northampton. After an early arrival to do desk duty and welcome families and professionals at the door, Alison Britton and I changed quickly from smart suits to casual jeans. We had been asked to accompany other volunteers by taking the children out for the day whilst their parents attended the conference. Although being slightly unsure of what would be expected of us, Alison and I found ourselves thoroughly enjoying the day and ready and willing to be volunteered any time in the future!

We spent the day at Drayton Manor Park, a nearby theme park. The MPS Society had never organised a day out at this park before and it turned out to be a very good choice indeed. There were plenty of rides catering for all levels of ability and for all age groups

so that no one felt left out. Some of the rides were more popular than others and after the twentieth trip on one ride we felt it was time to move on and explore the park further! However, if the kids were happy, that is all that counted.

The weather was very good to us and the park not too crowded. Bryn Edwards made an assessment of the park in terms of disabled facilities and we all agreed that a revisit to Drayton Manor Park would be a good option for the future. The children on the return coach trip all seemed very happy and talkative, and not in the slightest bit tired, unlike us adults! It was a very enjoyable day and I hope the kids returned to their parents with plenty of good stories to tell.

*Antonia Crofts
Assistant Development Officer*

MPS Children & Siblings at the Conference



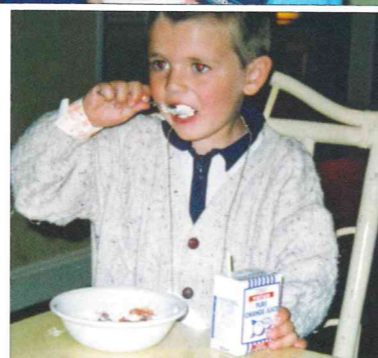
Chloe



Jordan Mount - Hurler



All on board for Drayton Manor Park



IN REMEMBRANCE

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

Annette Puddy who suffered from Sanfilippo Disease 03.10.83 - 14.04.01

Thomas Thacker who suffered from Sanfilippo Disease 05.06.88 - 03.05.01

Safiyah Hussain who suffered from ML II 28.02.1998 - 23.05.01

Angeline Chaudry who suffered from ML II 16.05.01 - 18.06.01

Hardev Rehal, 9 November 1986 - 2 May 2000

Hi, I'm Hardev Rehal's mum. Hardev had Sanfilippo and died May 2nd 2000 - just a year ago. He brought us so much joy and laughter into our lives, he taught me so much, e.g. tube feeding and giving medication. His smile and his funny look along with his beautiful big brown eyes followed you everywhere. He was 'MY' special son, Hardev was 13 and a half when he sadly passed away. He never gave me time to prepare for the biggest hole in my life. All those things I wanted to say, as Hardev slipped away. I told him to be happy so did his loving dad and brother Hardeep. Most of all we told him to carry our love with him.

I now work in the Ear Nose and Throat department at the hospital, which I enjoy so much. I get so much pleasure especially when I see special children come and be so brave. It's like Hardev is in front of me.

Hardev's brother and Dad were so close to him, his brother misses him so so much, and never goes a day when we are not thinking of him, or we will be having a chat and Hardev's name will come up in a funny way.

To us he was so gifted, he taught me so much and I know he's still looking after me

and looking down on me and saying 'good on ya Mum'.

I wrote a poem for Hardev,

'My beautiful boy' by Mum

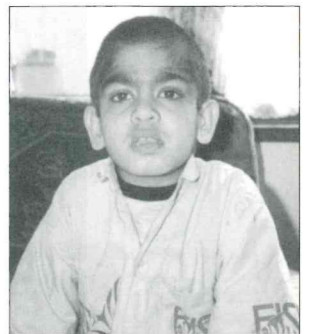
I miss you everyday
how much you'll never know.
Each day is a heartache
I can never let you go.

Waking up without you near me
is a pain I cannot bare.
But i hope you know
I will always care.

Soon I'll join you
where ever you are.
Pain free at last
but yet so far.

You were taken away from us
quick as a flash.
But our love for you
will always last.

Hardev, you'll always be in our hearts love
Mum brother and Dad



*Schmetterlingskind - German for Butterfly children
Named after the Television film that Emily appeared in*

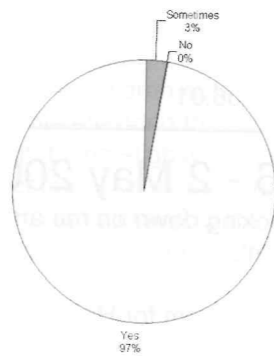


Emily Claire Volk
* 30. März 1983 † 3. Mai 2001
Schmetterlingskind

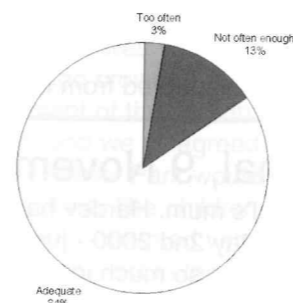
EVALUATION

As promised, here are the results of the readership survey that was included in the Spring mailing.

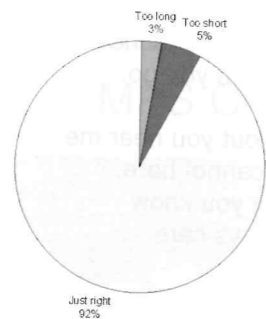
Do you consider the Newsletter to be an important source of news and information about MPS and related diseases?



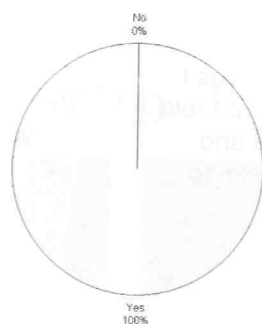
The Newsletter is published four times a year. Do you think this is?



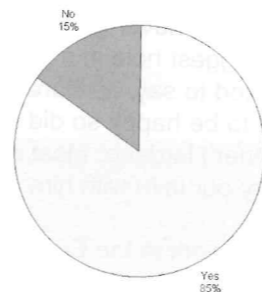
The Newsletter is normally about 40 pages long. Do you think that this is?



Is the format of the Newsletter easy to read?



To help fund the Newsletter, do you think it should contain advertisements?



Do articles in the Newsletter offer sufficient information? If not, what kind of information would you like to see included in future issues?

"I particularly like 'family' stories and medical information. More items on equipment and strategies for dealing with various problems eg. Puberty, sleepiness, types of beds etc. More exchange of info is very helpful. This is what happens at the conference, so updates etc are useful."

"The articles in the Newsletter always offer critical information for us, as we find it's a way of keeping in touch with what going on as we're unable to make it to many of the MPS functions."

Do you have any comments on the design and layout of the Newsletter?

"I think its great at the moment. Colour photos particularly enhance the Newsletter."

"Latest - Spring 2001 is an enormous improvement, and we offer our congratulations to all concerned"

"It is attractive and easy to read. I particularly like the shiny paper."

Please include any additional comments about the Newsletter.

"Poem, Spring Newsletter 2001. Please thank Mr R. Johnston of Notts. For making time to write such a very moving Poem on the MPS childhood Wood of Remembrance. My Brothers tree was in the very first batch planted, and was then 30 years too late. Such a touching and simply sincere set of verses, more than compensates for these painful memories and quite remote access."

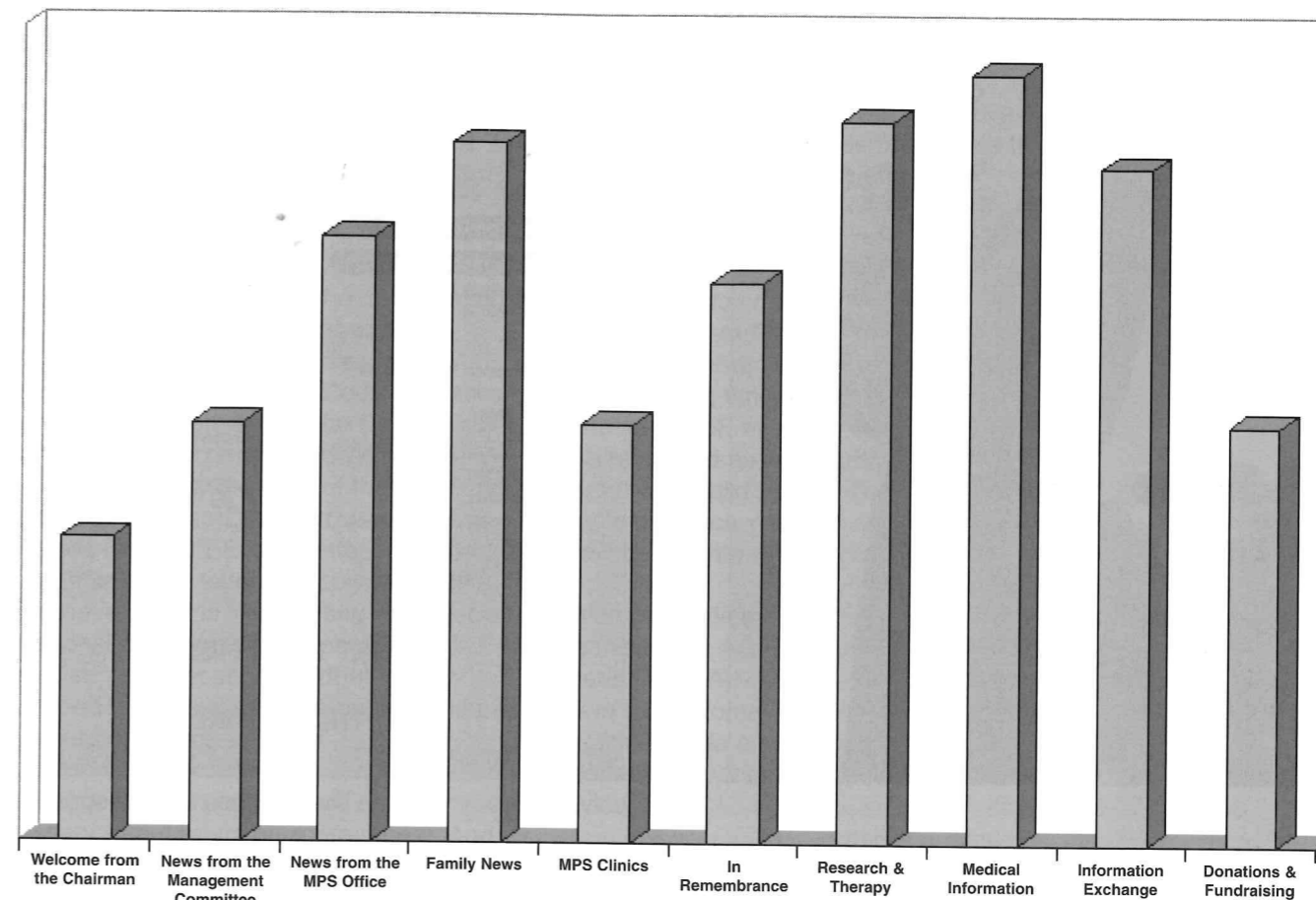
"When the Newsletter arrives, I get nothing done until I've read it from cover to cover. I then go back to it for reference from time to time. It is a valuable source of information not only to MPS families but professionals and friends who wish to know more."

"Advertising, this should be sympathetic with the Newsletter, it would also need to be increased in size if adverts were to be added."

Newsletter Survey 2001

EVALUATION

Which sections of the Newsletter are most helpful to you?



MPS Regional Clinic Evaluation

In the last newsletter we ran a feature on the role of the MPS regional clinics in providing a chance for families to meet specialist MPS consultants, members of the MPS Society staff and exchange news with other families.

When talking to families at the clinic we receive very positive feedback on all the clinics. We understand that many of you value these opportunities to meet the medical team and that, being regional, travel distances are often shorter and therefore easier and more convenient.

As you may be aware, the MPS Society is very keen to receive your comments and suggestions on the clinics and we aim to send out evaluation forms shortly after each clinic. From the clinics run since the beginning of the year we have collated all the forms we have received back in the office to produce the following summary:-

83 families in total were invited to the East Anglia, Birmingham, Newcastle, Bristol and Cardiff clinics 54 families attended.

54 evaluation forms were sent to those families that attended
33 evaluation forms were received back to the MPS office

Families were asked to grade several factors on a scale of 1 to 5 where 1 is poor and 5 is excellent. Scores for the five clinics held so far averaged out as follows:-

| | |
|---------------------------|---|
| Parking | 2 |
| Clinic Environment | 4 |
| MPS Support | 4 |
| Consultation with Doctors | 5 |

From the comments received it seems that parking was a real problem for many families and we will take this into account when making arrangements for future clinics. Unfortunately, parking facilities are not always under our control, but where opportunities do arise we shall do our best to make things as easy as possible.

INFORMATION EXCHANGE

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 13 Drottwich Road
 Worcester WR3 7LG

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Phone free on 0800 592 436
 or visit www.mobilityhire.com

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 or
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Phone free on 0800 592 436
 or visit www.mobilityhire.com

Disability Rights Commission

We are **RECRUITING** from the **28th February 2001** visit their website for more information

.....

► www.drc-gb.org
 or the Disability Now website at
 ► www.disabilitynow.org.uk

Linking Education and Disability

Looking for a new direction? Don't know where to start?

Call Freephone 0800 100 900 – and ask for Lead Scotland
 Helpline hours: 9 am–9 pm, Monday to Friday, 9 am–12 pm, Saturday

If you are:

- a disabled person aged 16 or over
- a carer phoning on behalf of a disabled adult
- a statutory or voluntary organisation working with disabled adults
- a provider of education or training
- a student or researcher looking for relevant information and you live anywhere in Scotland

Call us free for:

- information about education, training and lifelong learning opportunities
- guidance towards making the right choice for you
- continuing support

Lead Scotland provides this service for disabled people, through Learning Direct Scotland, the national guidance freephone helpline.



If you need information sent in an alternative format, please let us know.
 If you are a textphone user, please call: 0141 423 5710

INFORMATION EXCHANGE

Action on Entitlement

It is almost certain that this act will have passed through Parliament as the elections have been postponed due to the Foot and Mouth Crisis. In many ways this act is very positive, though Action on Entitlement (AoE), which represents a wide range of parent groups concerned with the education of children with special educational needs, is still somewhat concerned.

It is partly with thanks to AoE that the government has announced that the wording in Statements will not be changed from 'specify' to 'set out', and that they would enhance the guidance in the Code of Practice in relation to the contents of the Statements. Nevertheless, AoE remain concerned as they have not been shown a draft of the additional new guidance in the Code. From what they have read in the DfEE newsletter, in Parliament, and in letters in reply to parents, they believe that it is likely to say that special educational provision should only be 'quantified as necessary, and that it 'may often need to be expressed in terms of hours, equipment or personnel'.

That suggests that usually it will not be necessary to put hours on a Statement. And yet the reverse is more likely: generally the only way to write a Statement is to include the hours of additional teaching of classroom assistant time, although there may be

Waving Not Drowning

Waving Not Drowning: A Guide for Parents trying to combine paid work and caring for disabled children Published by Parents at Work, 45 Beech Street, London EC2Y 4AD Tel. (020) 76283565. Price £2.99 (single copy for any parent who has a child with a disability or special need), £3.95 for members of Parents at Work, otherwise £11.95

The Waving Not Drowning project is an established network for parents of disabled children who work or wish to work or study. The project is part of Parents at Work, a voluntary organisation that addresses the many issues facing working parents. This publication is a comprehensive guide for parents who may be already in work and want to know what options may be available to them such as flexible working hours, part-time or job-sharing. It also gives advice and guidance to those parents who may be returning to work or study after a break. As

exceptional cases where the Statement can specify in other ways. This is what the relevant case law says and just what the current Code of Practice explains.

AoE feels sure that LEAs will take advantage of any wording in the Code that allows them to avoid quantifying provision.

AoE are asking parents to write to their MPs to explain how important it is that the Code of Practice should continue to reflect the true legal position and should say, as now, that normally hours of the classroom assistant or extra teaching time should be specified in the Statement. AoE have a model letter that can be altered and as appropriate include details of your own child's case. The most effective way to convince your MP how important this is will be to write about your own experience.

Action on Entitlement include the following organisations: Action Support for the Special Needs Child, Advisory Centre for Education, Down's Syndrome Association, Association for Spina Bifida and Hydrocephalus, Independent panel for Special Education Advice...

For a model letter to write to MPs contact: Action on Entitlement, 17 Cumberland Road, Kew, TW9 3HJ. Tel: 0208948 1746 or visit the website www.aoe.org.uk

well as giving practical -guidance there are plenty of quotes and examples of parents experiences throughout the book, including a chapter of in-depth case studies. The guide includes a chapter on "Childcare" which discusses the various childcare options that may be available such as childminders, nurseries, nannies and so on ~~ includes useful checklists when interviewing potential care providers. The "You and Your Finance" chapter advises on benefits that parents may be entitled to, whether in full-time work, part-time work or perhaps being involved in voluntary, unpaid work. Another chapter considers the role of the statutory agencies such as Social Services, Health and Education while another looks at other sources of support such as that offered by Parents at Work, other voluntary organisations and support groups, including a listing of addresses and telephone numbers.



INFORMATION EXCHANGE

Carers



Carers and Disabled Children Act 2000

This Act will have come into force on April 2001 and will give local councils the power to supply certain services direct to carers following assessment. Persons with parental responsibility for a disabled child (parents or other carers) also have a right to ask for an assessment of their own needs.

Local councils may also make direct payments to parents of a disabled child to purchase services to meet the assessment needs of the disabled child and the family. Councils may also make direct payments to young disabled people aged 16 and 17.

Direct Payments

Local councils may offer direct payments to enable parents of a disabled child to arrange for the provision of services rather than rely on the services provided by the local council. Parents may want a direct payment because they think existing services are not suitable for their child and believe they can make arrangements for themselves which will be more beneficial.

A person who is considering using a direct payment to employ an individual to care for their child may ask the local council to carry out a check of that person under the Protection of Children Act. When a parent requests such a check the local council has a

duty to comply.

Direct payments are not an extra source of family income. They are paid instead of services after the family has been assessed to be in need of this service.

Short-term Break Vouchers

Councils will be able to offer parent carers vouchers to be redeemed against short-term breaks. Such breaks usually serve two purposes, both allowing the parents more time for other activities, e.g with non-disabled children, and affording the disabled child the opportunity to make new friends and participate in new activities.

Vouchers will be introduced from 1st October 2001, to allow for full consultation on the framework.

For further information...

The Department of Health has produced a Guide to receiving direct payments which is available in a variety of formats and an Easy Guide which comes as a tape, a CD and a booklet. For copies of both the Guide and the Easy Guide telephone 0541 555455.

The above information has been taken from the Department of Health Carers website at www.carers.gov.uk

Disability Living Allowance

From April 2001 children aged between three and four may claim the DLA Mobility Component for the first time (previously children did not qualify until they were five). This will only be for those children who meet the conditions of entitlement to the highest rate of the Mobility Component, having had difficulty making the transition to independent walking. The rate is £38.65 per week.

If your child is not walking by the age of two and a half, or is having difficulty making the transition, you should apply. The benefit cannot be paid until your child is three years old and there is a qualifying period before the Mobility Component can be paid, but this can start before your child is three years old.

If your child is aged two and a half to five years old and you think you might qualify for the Mobility Component you should claim

now. The Department of Social Security has said that parents of children already receiving the Care Component of DLA and aged between two and a half and five will be contacted and invited to claim Mobility Component. If you have not been contacted yet you can telephone the DLA on 08457 123456 or contact your local Disability Benefit Centre.

Also, if your intentions are to lease hire or purchase a car on the Motability Scheme your child needs to have been awarded at least three years of the higher rate of the DLA Mobility Component. A car cannot be leased/hire purchased unless you have a full three years. Therefore it is crucial that an application is made far in advance and that the car has been chosen and paperwork with the garage finalised eight weeks prior to entitlement beginning, in most cases this will be eight weeks before your child's birthday.



INFORMATION EXCHANGE

Airline Code for Disabled People

Disability organisations are being consulted over a UK code of practice for air travel for disabled people.

The draft code from the Department of the Environment, Transport and the Regions aims to improve the accessibility of air travel to disabled people and is aimed at everyone involved including travel agents, tour operators, UK airlines and airports. The Disabled Persons Transport Advisory Committee is producing a separate guide aimed at disabled air passengers.

Although the Disability Discrimination Act 1995 covers air travel in relation to booking services and airport facilities, aircraft are exempt from its provisions. The code will cover all aspects of air travel, from accessing

information through to arriving at the final destination. It will also cover the design of the airport and aircraft.

Organisations being consulted include British Council of Organisations of Disabled People, Mencap, Scope, Deafblind UK and Radar.

The Disability Rights Commission is also being consulted. Last year, its chairperson Bert Massie was turned away from a flight by Scot Airways after his disabilities were labelled a "safety risk". Massie was told on reaching the departure lounge that the airline's policy was that all passengers had to be able to walk onto the plane, although he had informed airline staff that he was a wheelchair user. For details go to www.mobility-unit.detr.gov.uk



Travel Insurance for Disabled People

Listed below are companies who offer travel insurance to disabled people. Whilst these companies cannot issue policies more than 3 months prior to travel, they will issue quotes.

Travelcare Ltd

69 High Street
Chislehurst BR7 5AQ
Tel. 0800 181532

Travelbilty

J&M Insurance Services (UK) plc
14-16 Guilford Street

London WC1N 1DX
Tel 020 7446 7626

Roger Chant Associates

47-49 College Road
Bromley BR1 3PU
Tel. 020 8290 2056

Hamilton Barr Insurance Brokers Ltd

Bridge Mews
Bridge Street
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Tel. 01483 255888



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New National Website

New National SIBS Website The aim of Sibs Encounter is to help support brothers and sisters of people with disabilities. The group offers the opportunity for siblings to communicate with their peers and share their feelings and fears. It is a modern and exciting group where the members take an active role in the organisation and decision-making. Thanks to the support of the Tuberous

Sclerosis Association people can communicate using the website, participate in social activities and take the time to "feel special". The site contains various rooms including a reception room, library and official news room. So if you know a sibling who would benefit from this support who has access to the internet, let them know about this site. Web Site: <http://www.sibspace.org>



INFORMATION EXCHANGE



Parental Leave

The Government has said that later this year, probably in the autumn, it will extend entitlement to parental leave (which is unpaid) to all parents with children under five not just those with children or adopted after 15th December 1999 which will be backdated.

Raincover for McLaren Major 2000

The MPS Society has recently heard from a family who have a sunshade and raincover suitable for a McLaren Major 2000 buggy

Listening Books

Listening Books Sam Fletcher writes: Many people with disabilities are tired of being 'special' and seek only to overcome barriers to leading reasonably ordinary lives. The Listening Books service removes one such barrier by providing access to the wonderful world of books through its audio book library. The Listening Books service is available to anyone who has difficulty reading in the usual way because they cannot see the book, read the book, hold the book or turn the pages, as well as to many others with specific learning difficulties, including Dyslexia, for whom reading print is frustrating or even impossible. This service can be provided to individuals, schools, nursing homes and other organisations. The library holds over 2,500 titles and this stock is being continuously expanded through our two digital recording studios. We have tapes for leisure and

Parental Leave

Parents of disabled children are to benefit from new measures introduced by the Department of Trade and Industry. Their parental leave entitlement is to be increased from thirteen to eighteen weeks and can be taken up until their child's eighteenth Birthday. This will give parents of children with disabilities greater flexibility to strike the balance between working and caring for their child's additional needs. Philippa Russell, Director of the Council for Disabled Children said: "Working parents with disabled children frequently experience barriers to employment and the ability to take additional days off for assessments, hospital visits or to care for a sick child will be greatly valued. When a child

in addition parents of disabled children will get an increase from of leave 13 -18 weeks. The provisions that leave dates must be agreed in advance and not be taken in chunks of more than four weeks remain.

which they no longer require. If you would be interested in receiving this please contact the MPS office for further details.

learning, adults and children. We offer a selection of tapes supporting the national curriculum at key stages two, three and four to help children in education and we have the four Harry Potter stories on tape. Listening to books is a great way to focus and develop your imagination and creative skills. One of our younger members has his own Web Site <http://www.iamdyslexic.com> where he describes Listening Books as a solution to his problem with a books. The membership fee costs £50 a year for an individual and starts at £100 for organisations. Postage and catalogue costs are all included in the Listening books "A lifeline for people with special needs membership". Contact: Listening Books, 12 Lant Street, London, SE1 1QH Tel. (020) 7407 9417 e-mail: info@listening-books.org.uk

is disabled, parents acquire many additional roles. They are likely to be providing transport, additional support and active participation in assessments, therapy and reviews. The extension of parental leave will be greatly welcomed and will help equalise opportunities for those parents who have additional caring responsibilities." This leave is available to parents of children who receive Disability Living Allowance and can be taken flexibly in blocks of a day or more at a time, up to four weeks in total, each year. The timing of the leave has to be agreed with the employer and notice has to be given. These changes will come into force later this year.



INFORMATION EXCHANGE

Caring Today

Mrs Fayyaz is the mother of Sannah aged 6 years who suffers from Sanfilippo Disease, unable to find suitable local support groups for her daughter, she has joined with other parents in Lancashire to establish Caring Today. - Ellie Gunary

Caring Today caters for children with rare conditions and are incurable. These children are hyperactive with severe Learning/behaviour problems and short lives due to their condition.

The need we are meeting is particularly important because no other disability group within our area is actually providing for these children's need. Caring Today is targeting children from the age range from 2yrs upwards. The majority of provision for this age group will be operated around recreational structure. We believe as parents/carers that children in this age group, given the right support are capable of learning as other children but at a slower pace. We want to harness and develop these skills in a positive way.

We believe that special needs children can be very creative and open in their acceptance of differences, but often find their tolerance blunted and impaired by the environments in which they live. We want to support children in developing and enhancing their tolerance levels. In accordance with the above we have implemented integration with their peer group,

Payments Drive

The Scottish Executive has allocated £530,000 to promote schemes allowing disabled people money to pay for their own care. The move follows a bill currently before the Scottish Parliament which will extend these direct payments to 16 and 17 year-olds. The Executive is also beginning a consultation with clients, families and carers aimed at seeing how the payments could be

Carers and the 2001 Census

For the first time the census included a question about caring responsibilities. The question asked "Do you look after, or give any help or support to family members, friends, neighbours or others because of: long-term physical or mental ill-health or disability, or problems related to old age?" Answers to this question should enable a

so that these children with special needs/disabilities do not feel isolated or abnormal.

We will actively monitor and evaluate the project by weekly meetings and evaluation sheets each session.

We know the project will be effective because we as carers/parents understand our children's need. We also will be recruiting special needs staff and administrative staff. As well as working with children, the group will also work directly with parents, individually involving them in establishing realistic objectives for the development of their own child's progress. Counselling will also be available for parents when necessary. We intend to attract children to the project from varying backgrounds. We are the best people to do the work because as parents/carers we are aware and have identified the needs of our children. We also have volunteers whom are prepared to give up their precious time to help with our group

The management committee who are totally committed to help these children with their special needs. They are available day/night if the situation arises.

The project will cost £30,000. We intend to raise the money by sponsorship, having stalls and receiving funding from Social Services/Education Department.

extended to all disabled people who want them, and how parents could use direct payments to buy services for their disabled children. An employers' code of practice' written by disabled people has been published to help those in Scotland receiving direct payments to understand their legal responsibilities as employers. £16.25, tel: 01415504455.



FUNDRAISING



Barbara Wilson runs the London Marathon

The 21st London Marathon took place on Sunday 22nd April, 2001. Probably for the first time in my life, I can begin to understand how the will and spirit of friends, family, colleagues and indeed, well intentioned strangers, can inspire you to achieve!

We arrived in Greenwich Park at approx. 8.15 am. It was difficult to imagine 32,000 Marathon runners. I was one of them! Nerves and anticipation meant that an immediate trip to the loo would be in order. I queued along with hundreds of others! Some three-quarters of an hour later, relief at last! Calmly making my way to row 9, somewhere towards the back of the Red Start, to take my place amongst the thousands of starters. After a great send off from Barry, Joanna and her best friend Kim, I inched my way towards the start line and set off on my great adventure.

It actually took over 20 minutes before I reached the official start point. However, that's the price you pay for being honest in your expected completion time! Thousands of well-wishers, music, horns, and whistles filled the air, urging us all to succeed. I felt excited and hopeful, steadily running along, being overtaken by a rhino and giraffe, perhaps not a good omen!

I was to see my family again at Bermondsey very near to the Rotherhithe tube station, about 11 miles from the start. Meanwhile I settled into the warmth, companionship and camaraderie of the other runners and to the kindness and enthusiasm of all the spectators that lined the route. What a fantastic atmosphere! I felt great and optimistic that I could reach the finish line in half my expected time! It was towards the end of this period



that I began to have serious problems with first my right foot and later my left. Not blisters or cramp like others were experiencing, but sharp, shooting pains from the toes upwards towards the ankle, then knee. Rather like a thick needle prodding into the base of my foot! Agony with every step! I had to stop running and considered withdrawing! It was then that the family came into view and with their help I stopped to make emergency repairs to my feet. I tried some anaesthetising/paracetamol plasters, freebies given out at the registration point in the London Arena. I then recalled all the kind thoughts and good wishes sent by friends, family and colleagues. This spurred me on! I continued and minutes later was joined by an elderly man! Yes, very strange! He was wearing a suit, raincoat, brown brogue shoes and held an umbrella. "Are you in much pain?", he asked, followed by, "No one will think any the less of you, if you drop out. You don't have to do this". The more he went on about this, the more determined I became to succeed! He told me he did this walk every year and no one had ever asked him to leave. He wasn't officially in the Marathon? He stayed with me and talked continuously while I listened, until the 16 mile marker approached. I looked around for my family and saw them ahead by which time this lovely but strange gentleman, disappeared out of sight!?

It was here at Canary Wharf (16 mile marker) that I had to stop for further emergency repairs, this time to both feet! I was in agony but realisation struck that I had actually managed to get through the last 5 miles and was now beyond the half-way point. A friendly policeman suggested I simply swapped over

FUNDRAISING

socks. It always worked for him, "seriously" he said! Well, what have I got to lose! By now, the crowds were thinning in places but small clusters of faithful supporters were continuing to party and there were many individuals who would shout out such inspiring words that I felt encouraged to carry on. Offers of free gin and tonics, cups of tea and the odd pint, were tempting along the way. Some of my fellow walking wounded could not resist, and fell by the wayside! I was struggling on slowly by now, wondering what the hell I was doing to myself and would there still be a finish line left even if I made it there! I have to admit that by now tears were coming very easily and frustration set in because any respectable finish time had gone out of the window, so to speak! I was feeling very cold and in much pain but I wasn't alone. The camaraderie, support and friendship offered by fellow walkers/runners was inspiring and comforting. Before I knew it the 20 mile marker was in sight. Well intentioned comments like, "Jesus is with you, you'll succeed" and "Believe in yourself, keep going, you'll get there" were shouted by complete strangers. I thought to myself, "I haven't come this far to give up" but it wasn't going to be easy. I kind of day dreamed through the next two miles because I honestly don't remember a thing about this stretch!

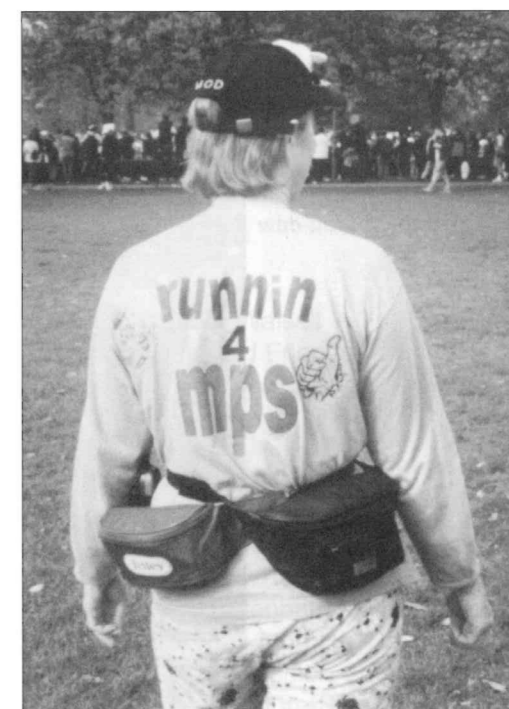
With just four miles left to the finish and the way I felt at that point, it might just as well have been 400 miles! I was tired. No, exhausted beyond belief. The field of competitors had thinned out to just a few stragglers and just the occasional word of comfort from a passer-by. The shooting pains I felt in my legs prevented any chance of more than a very slow pained walk. Was I going to make it? I then saw Barry walking towards me and after a few tears and a very large cuddle, I decided to carry on. It was then, that a saying one of our friends Steve, often says jokingly (in a slightly, slang, - Bromsgrovian or possibly Brummie accent!), came into my head, "you loose yourself down, you loose your friends down, you loose everybody down" ! I repeated this aloud, which caused Barry and I to laugh enough to carry me through for at least the next mile!

Big Ben was now in sight! The enormous crowds that I had been part of in previous years as a spectator, had almost disappeared and London was returning back to normal. "What happened to the Marathon? I pressed on round the corner into Birdcage Walk with just about a mile to go. Three competitors,

with whom I had changed positions a few times over the course, slowly and obviously painfully passed me by, another blow to my morale.

At last I turned the corner into the Mall with the end in sight. Had I really got there? Was there anybody left to see my moment of triumph? I needn't have worried. A round of applause and shouting of well done etc. greeted me as I crossed the finishing line. A race marshal hung my medal around my neck followed immediately by a very bright flash, my moment of triumph had been captured for posterity. Generations of Wilson's to come, will see this proud moment in my life. Immense relief and disbelief, all rolled into one! Not quite sure how I did it but I do know that I didn't do it alone! I ran for my daughter Joanna. I ran for M.P.S.

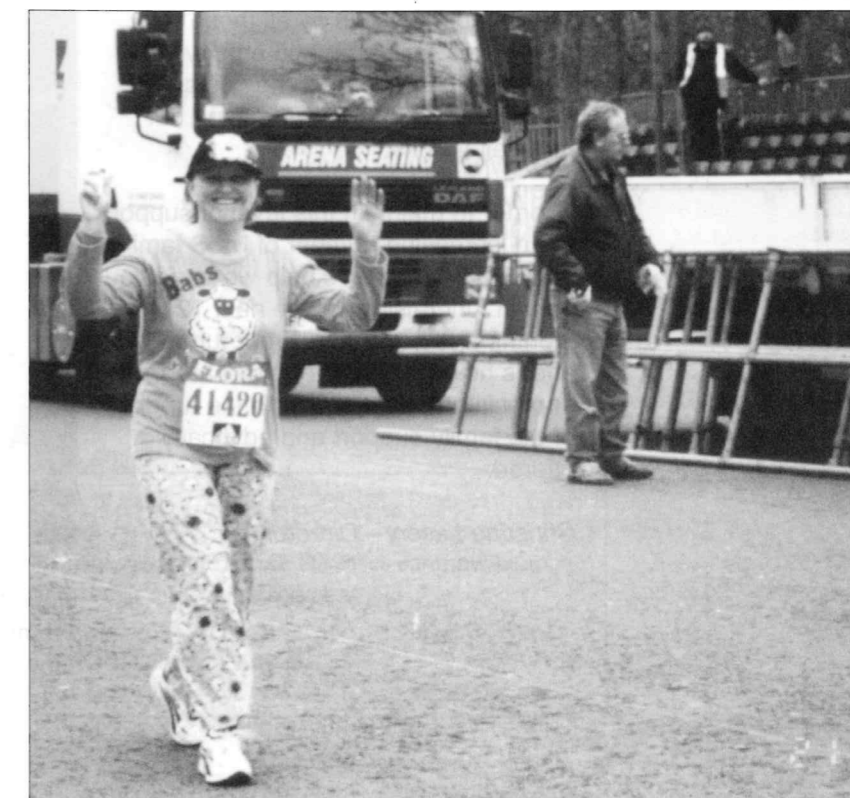
All I can say is that my pain is temporary and will be gone soon but children suffering with MPS have no choice, they live with pain! They're the ones I would have let down if I hadn't completed this Marathon!



Barbara heading towards the start.

"Thank you to all my family, friends and colleagues in the Careers Service, for supporting and sponsoring me. You reponce has been overwhelming."

'Made it' 26.2 miles!



FUNDRAISING



Amanda Natrella and Clare Bennett who ran the Bath half marathon, they raised £820.10.

The Society Needs You

Over the last 12 months the MPS Society has responded to more family support and advocacy requests than ever. The Management Committee have responded by increasing the staff team to meet these needs. The Society's ability to respond in this way has only been made possible by all those who have given their time and energy to raise the funds to meet the costs involved. To all of you we extend our heartfelt thanks.

In order to maintain this level of support to an increasing number of MPS families seeking the Society's help we need your help in fundraising for the next 12 months and beyond. All your fundraising efforts however great or small are most appreciated and are vital to the funding of the family support and advocacy offered.

Christine Lavery - Director



This is a picture of Roma Katherine Drayne, she has sent a £10 donation from her first Holy Communion, that took place on the 2nd June at St. Colmans Church

DONATIONS & FUNDRAISING

Donations

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

B.C.S Ltd – Leeds
 Cadogan – London
 DLM Charitable Trust
 The Really Useful Group
 Mrs Taylor – Wales
 John & Jane Heritage
 Reading Football Club
 The Berkley Group PLC
 Andreas Charalambous – Cyprus
 The Rainford Trust
 Joseph Strong Frazer Trust
 The Clover Trust
 John Huntley – Reading
 Angela Seymour
 The John and Lucille Van Geest Foundation
 John Lewis – Wycombe
 Waitrose – Chesham
 Argos – Milton Keynes
 Charity Flowers – Guernsey
 Eli Lilly & Company
 Lodge Greenock St Johns No 175
 Courtyard Designs – West Sussex
 Susan Lowry
 BHP Ltd
 Eurocamp
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 Babcock International
 Duni Ltd
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 Mr S Blanch
 NNC – Cheshire
 Viking Direct
 Nestle UK Limited – Aylesbury
 Sainsburys – Beaconsfield
 Buckinghamshire Building Society – Chalfont St Giles
 Electrolux – Luton
 Parfett's – Manchester

Fundraising

The Society is grateful to the following who held fundraising events

C M H T Solicitors – Walsall
 The Andrew's Family and Friends
 The Wellington Inn – Knaresborough
 Mrs M Lord – Lunch
 Bingo – Holt
 Peal O'Bells – Holt
 Wintershall Tennis Tournament
 Amanda Natrella & friends – Bath 1/2 Marathon
 The OPM Club – Draw/Raffle
 Caradon Plumbing Solutions – Cheltenham – Prize Draw
 Brenda Weston – Coffee Morning
 Anne Cotton and Heather – Fashion Show
 Northgate – Obsolete Furniture Sale
 Mrs M Stimpson – Webb Ivory Sales
 Gate Hangs High Crown Green Bowling Club-Wrexham
 Barbara Wilson – 2001 London Marathon
 East Down Institute – N/Ireland – Weakest Link Competition
 Harry & Rosemary Nurse – Car Boot Sales
 Marina and Dave Neverland – Car Boot Sales
 Jean Cockman – Bags & Cans
 Neil & Ian Coney – General fundraising
 Tom Holroyd – 2001 London Marathon

KINDLY DONATED TO THE MPS SOCIETY IN MEMORY OF

JAMES EDWARDS
 NEIL PALMER
 EDWARD NOWELL
 JULIAN WARREN
 ANNETTE JANE PUDDY
 WILLIAM HOLROYD

Donate Online

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

www.mpsociety.co.uk

MANAGEMENT COMMITTEE

NEWSLETTER DEADLINES

AUTUMN

31 September 2001

WINTER

17 December 2001

SPRING

31 March 2002

SUMMER

30 June 2002

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Mark Beniston
17 Hameldon Close
Hapton
Burnley BB11 5QY

Vice-Chair

Angela Brown
Steve Butler

Treasurer

Vince Hayward

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Rob Devine
Judith Evans
Sarah Long
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| Ellie Gunary | Assistant Director |
| Angela Ratcliffe | Development Officer |
| Alex Roberts | Project & Information Officer |
| Sarah Pritchett | Development Officer |
| Andy Hardy | Development Officer |
| Antonia Crofts | Assistant Development Officer |
| Alison Britton | Assistant Development Officer |
| Gina Page | Finance Officer |

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

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

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

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

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THE SOCIETY FOR MUCOPOLYSACCHARIDE DISEASES



19th National Conference on Mucopolysaccharide and Related Diseases

Hilton Hotel
Northampton

14th - 16th September 2001



Saturday 15th September 2.00pm - 12.45pm

Satellite Seminars

| | | |
|-------------------------|------------------|--------------------------------------|
| Home Adaptations | | |
| Overview | Dr Fiona Stewart | Belfast City Hospital |
| Homes fit for children | Ginnie Shaw | HODIS |
| Alex's Story | Ellie Gunary | MPS Society |
| Other Therapies | | |
| Aromatherapy | Sarah Horswell | Naomi House Children's Hospice |
| Play Therapy | Ailsa Foster | Rainbow Trust Children's Charity |
| Physiotherapy | Pauline Hensman | Royal Manchester Children's Hospital |
| Palliative Care | | |
| Overview | Dr Ed Wraith | Royal Manchester Children's Hospital |
| Facing Loss | Jenni Thomas | Child Bereavement Trust |
| Developing a Care Plan | tbc | MPS Society |
| Looking after me | | |
| Child Bereavement Trust | tbc | Child Bereavement Trust |
| | Angela Ratcliffe | MPS Society |

Saturday 15th September 9.15am - 12.45pm

| | | |
|--|--|---|
| Jeans for Genes Research | Dr Guy Besley Dr Maureen Cleary Dr Robert Coffin | Royal Manchester Children's Hospital Royal Manchester Children's Hospital University College London |
| Preimplantation Diagnosis | Professor Joy Delhanty | University College London |
| Personal Experience of Enzyme Replacement Therapy | Steve & Amy Holland | MPS Society USA |
| Enzyme Replacement Therapy Update | Dr Ed Wraith Dr Ashok Vellodi | Royal Manchester Children's Hospital Great Ormond Street Children's Hosp. |

HOPE TOMORROW

CARE TODAY

Friday 14th September 12 noon onwards

| | | | |
|-------------------------------|----------------|---------------------------------|----------------|
| Registration | noon onwards | Welcome and mini-market | noon onwards |
| Sibling workshop | 3.00 to 5.00pm | Children's entertainment | 7.00 to 8.30pm |
| Annual General Meeting | 7.30 to 8.30pm | Volunteers' Meeting | 9.15pm |

Saturday 15th September 9.15am - 12.45pm

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

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

Aims of the Conference

The MPS Conference offers parents and professionals the opportunity to learn about the management of MPS children and young adults as well as the latest therapies and research developments. The Conference programme offers ample time to meet and talk informally with other professionals and for parents to share experiences. Those suffering from MPS and their brothers and sisters have their own special programme.

What the Hotel Offers

The hotel is open plan and spacious. Bedrooms contain **one double bed or two double beds or one double bed plus sofa bed** and all are en-suite. Ground floor bedrooms will be allocated on a first come first served basis with priority being given to the more severely disabled. There is no lift to the first floor bedrooms. The Hotel has its own swimming pool and leisure club. There is ample level parking in the grounds. Conference delegates are welcome to arrive early on Friday and use the Hotel facilities. However we cannot guarantee that their rooms will be available until 3pm.

Children attending the conference weekend

No children under the age of 18 years will be admitted to the conference. Creche facilities will be provided for children under 3 years. MPS sufferers over the age of 3 years may be cared for in the creche by prior agreement with the Society. A full day outing on the Saturday and half day outing on the Sunday will be arranged for all children aged 4 to 17 years. A teenage outing will be organised on Saturday evening for brothers and sisters and those physically disabled by MPS. Babysitting and patrol facilities will also be available on Saturday evening to enable parent carers to attend the Gala Dinner. MPS children and their brothers and sisters will be cared for on the outings by trained and experienced volunteers resident in the hotel throughout the weekend. These volunteers will also undertake babysitting and patrol duties.

Sibling Workshops

A sibling workshop for brothers and sisters, aged 5 years to 11 years, will be held on Friday 14th September 3 to 5pm. If you wish for any of your children to attend this workshop please complete the sibling workshop booking form. For the first time we will be offering this sibling workshop to MPS families not attending the weekend conference and places for resident and non-resident families will be offered on a first come first served basis. The maximum number of places in this workshop is 20.

How to book

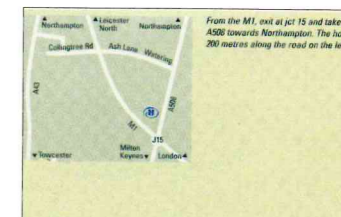
Enclosed you will find a registration form for the conference. This form can be completed by both professionals and MPS affected families wishing to reserve places for the weekend conference. If a registration form is not included with this programme, please telephone the MPS Office on 01494 434156.

Professional Delegates

The Society welcomes professional delegates who may attend for the weekend, part weekend or the Saturday only

Paying for the conference

There are three ways of paying for your conference reservation:
Cheque
Credit Card
Invoice to a hospital or institution



MPS Society, 46 Woodside Road, Amersham, Bucks HP6 6AJ
Telephone: (01494) 434156
Email: mps@mpssociety.co.uk
Registered Charity Number: 287034

Fax: (01494) 434252
Web Site: www.mpssociety.co.uk
CME Accreditation Applied For

Jeans for Genes Day

5th October 2001



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

I'm ready...

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

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

...are you?

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

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

Call freephone

0800 980 4800

or visit

www.jeansforgenes.com

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



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