Society for Mucopolysaccharide and Related Diseases

National Registered Charity No. 287034

Autumn Newsletter 2004

Care Today, Hope Tomorrow

What are Mucopolysaccharide Diseases & Related Diseases?

Mucopolysaccharide & Related Diseases are individually rare; cumulatively affecting 1:25,000 live births. One baby born every eight days will be diagnosed with an MPS or Related Disease. These multi-organ storage diseases cause progressive physical disability and, in many cases, severe degenerative mental deterioration resulting in death in childhood.

What is the Society for Mucopolysaccharide Diseases?

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

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

How does the Society meet these Aims?

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

Telephone Helpline
Includes out of hours listening service

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

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

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

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

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

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

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

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

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

Front cover photograph: Sibling Activity Weekend

Newsletter

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Newsletter Deadlines

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

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

Contents

Contents

Editorial

Welcome to the Autumn 2004 edition of the MPS Newsletter. The advocacy support team has worked particularly hard over the last few months compiling articles for the following pages. I hope you find them of interest.

Amongst others there is a big story on our recent Sibling Activity Weekend. This was a highly successful and fun event as you will read, but it is just a shame that we received so few bookings.

To boost the Society's fundraising and by way of thanking all our supporters, the fundraising section has been removed from the main newsletter and is now an individual publication in its own right. The fundraising bulletin will be a quarterly edition packed with stories and ideas about how you too can help the Society. Contact us now if you want more information or to receive a fundraising pack.

Antonia Crofts, Editor

This edition of the MPS Newsletter has been funded with the kind support of a grant from the Roald Dahl Foundation.

Chief Executive's Report

No one, least of all members of the MPS Society could remain oblivious to the sad news of Jacob's death from Hunter Disease and subsequent arrest of his parents Andy and Mary Wragg. Whilst on leave, I was painfully reminded daily of the tragedy unfolding through the British papers on sale in Swiss newsagents. This event inevitably put MPS in the spotlight and all of you who saw the Society's Assistant Director, Ellie Gunary, on television will know how well she did representing the way the whole spectrum of Hunter Disease affects individuals. Not surprisingly, the views of our members expressed to the Society are wide ranging. Over the coming months these tragic events will be played out in court. The MPS Society remains committed to supporting and upholding the rights of MPS families no matter where their sympathies lie.

Enclosed with this newsletter is your registration form for the MPS National Conference to be held on 1 - 3 July 2005 at the Northampton Hilton Hotel. By putting together three separate conference programmes on the Saturday which will run simultaneously we truly hope this will encourage many members and their families to attend for the weekend. One of the key aims of the weekend is to provide up to date information on all aspects of MPS and Related Diseases. Whilst this conference will be family-orientated with a full children's programme, professionals will be equally at home becoming knowledgeable on all aspects of clinical and social aspects of MPS.

Over 80 families entered the draw to be selected for a place to the American MPS Disney Conference to be held in Orlando in December. Fifteen lucky families were drawn by the children of Chenies Primary School.

Finally, as you will see later on in the newsletter, the Sibling Weekend at the Pioneer Centre was most successful and those brothers and sisters who took part had a brilliant time. As highlighted in Clare Cogan's article, only 10 siblings went on the weekend when we could have taken 50. It is now up to you to let Clare know whether you would like another sibling weekend organised for your children.

Christine Lavery
Chief Executive

Introducing... Sarah Cornwell

My name is Sarah. I am a new recruit to the MPS Society, where I will be working alongside Clare Cogan and Sophie Denham as Advocacy Support Worker.

Before starting here I was at university. I graduated in July 2004 with a 2.1 Law degree from Southampton University. Having done this degree I decided it was time to put my skills and experience to some good use and find a job!

As yet I am still settling into the 9-5 routine, something of a mystery to me as a student. University appeared to be scheduled around late mornings and late nights. These were either due to a hectic social life or at other times due to deadlines for essays and dissertations. University was great fun, from which I have learnt so much. But as they say, all good things come to an end, and it is now time to start repaying student loans and overdrafts incurred along the way.

The job here at the MPS Society itself is very rewarding and I only hope I can be of help to you all in supporting you through your experiences. I look forward to meeting you at clinics and other events up and down the country.

What have I been doing for the past 18 months?

Sophie Denham returns...

Hi! It's Sophie here. As some of you may know, I am currently in my last year of training to become a qualified Social Worker.

The course I am undertaking is a Diploma in Social Work (DipSW) through the Open University. It is a part time course with most of the learning being home-based, although I did need to do work other than in my usual workplace. I have been sponsored by the Society to do the course and I opted to complete it in 2 years.

In my first year I have done two courses. One, which looked generally at health and social care, was very much home-based with optional tutorials. The other was stage one of the DipSW, which as well as home learning had a compulsory 55 day placement. This could have been within your own workplace or in another agency. Although we tried to get a placement outside of the MPS Society, we were unsuccessful, as few places in the area accepted Open University students. Thus my first placement had to take place within the MPS Society.

During my time on placement, Ellie volunteered to be my practice teacher. This meant that not only was Ellie my manager but, during my time on placement, she would have to undertake extra duties as my practice teacher. This was a whole new experience for both of us, but I feel we adjusted well to the new situation and I personally developed and learnt a considerable amount during this time. I would at this stage like to extend my thanks once again for all the time and hard work Ellie put in as well for passing me! I would also like to thank all the families who helped me meet my competences and who allowed my practice to be observed (I did give everyone the chocolates I promised didn't I?).

In passing the practice placement, all that was left for me to do was to pass my exam. This was the most nerve-wracking part of all. I should, however, have had more faith in my own abilities as I passed with flying colours. This meant that I was able to proceed to year two, where the heat was really on with three courses to take.

Year two started in February 2004 and my three courses were in law, children and families, and stage 2 of the DipSW.

As well as doing these three courses I had to complete an 85 day placement in another agency. As with the year before, this proved to be an uphill struggle as the Open University still was not widely recognised within the area. The panic was over when I was offered a place, working in a Social Services Referral and Assessment Team (aka The RAT team) for children in the local area. My placement with them started in late February and finished in late July 2004.

Working within a statutory service was very different from working within the voluntary sector, so it took me a little while to settle in but I soon felt part of the team and was very well supported by all members of the team as well as by my practice teacher who was also the team manager. One of the main aims for me on this placement was to experience other client groups to ensure that I had a broad knowledge of children and families not just in the field of disability.

I found my time on placement, exciting and uplifting and felt that my confidence increased two-fold as I was working with children and families where, in many cases, we did not know what the concerns were or on which we had very little information. The placement also allowed me to better understand and develop a good working knowledge of the roles and responsibilities that a statutory service has. I also developed my knowledge of the Children's Act and other legislation together with the operational guidance used such as the Department of Health Assessment Framework. The placement also afforded me the opportunity to work in the field of Child Protection, which was an area that I had had little involvement with previously.

In terms of course work I am still currently busy working through my many assignments, although have submitted my 10,000 word report (which took me months of planning, stress and more grey hairs!). I also had confirmation this week that my practice teacher has recommended that I pass stage 2 of my Social Work training (Yeah!).

This only leaves me with two exams left to do which are taking place in October. I will then have to wait until late December to find out if I have passed.

News from the Management Committee

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed at the Management Committee Meeting on 10 July 2004 at the East Midlands Airport Hilton Hotel.

Election of Officers

The following Trustees were elected as officers: Chairman - Barry Wilson; Vice Chairman - Judith Holroyd; Vice Chairman - Bob Devine; Treasurer - Judith Evans. This was proposed by Ann Green and seconded by Sue Peach.

The Chief Executive and Assistant Director thanked all the Trustees for their commitment to the MPS Society.

Advocacy Events

The feedback received from members who attended the Alton Towers Family Weekend and Annual General Meeting was excellent. It was agreed to repeat this event in 2006.

A draft programme for the National Weekend MPS Conference 1-3 July 2004 was tabled and approved. A proposal to incorporate the Remembrance Day in the Childhood Wood into the conference programme for bereaved members was agreed.

Advocacy Support

The Assistant Director spoke to the regional clinic programme and gave the Trustees an update on the progress made evaluating the Birmingham and other clinics throughout the UK.

Trustees were informed of the process involved in selecting 15 MPS families to attend the MPS Disney Conference in Orlando in December.

Policies

Terms and Conditions of the Medical Research Grants was agreed subject to the following amendments

a) the lead investigator must either be employed by the UK institution or form part of a formal collaboration with a UK institution

b) that acknowledgements in published papers, medical journals etc must recognise the funding sources as the MPS Society and Jeans for Genes

Research Grant Update

Judith Holroyd reported on the meeting she and the Chief Executive had with Professor Bryan Winchester and Dr Claire Beesley from the Institute of Child Health, London to discuss the research project on Biomarkers for MPS funded by the MPS Society.

Jeans for Genes

The Chief Executive, Christine Lavery, reported on the excellent job MPS member Chris Holroyd is undertaking on the Jeans for Genes Charity Board along with MPS Trustee and member Bob Devine.

Publications

The Chief Executive informed Trustees that the ML II and ML III booklets have gone to print and that the MPS VI booklet will be printed in August 2004.

Professor John Joseph Hopwood - Member of the Order of Australia

Christine Lavery

The MPS Society would like to congratulate Professor John Hopwood of Adelaide Women and Children's Hospital on being made a Member of the Order of Australia.

This award is for service to biomedical research in the field of lysosomal storage diseases and to the community, through support for people affected by these diseases.

Professor Hopwood has a long association with the MPS Society dating back to the mid 1980s.

In 1987 in collaboration with Dr Ed Wraith at the Royal Manchester Children's Hospital over 100 MPS II boys and their mothers gave blood samples at the MPS Annual Conference to facilitate the finding of the MPS II gene. Amazingly, a complete deletion of the MPS II (Hunter) gene was found in two members. We all hoped but no one could have known then that due to that important discovery, 15 years later there is now a worldwide clinical trial of Enzyme Replacement Therapy for MPS II sufferers.

Advocacy Support Bulletin Board

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

Regional Clinics and Events 2004

October

Fri 22	Childhood Wood Tree Planting

November

Wed 3	BMT Clinic
Fri 5	BMT Clinic
Thu 18	Northern Ireland Clinic

Wedding Bells for Kirsten and Justin

Julie and Ian Macintyre

Julie and Ian Macintyre's daughter, Natasha, had Sanfilippo Disease, MPS III. The Macintyre's recently wrote to the Society to let us know about the marriage of their other daughter, Kirsten. Here is an extract from their letter...

We thought you might be interested by the enclosed photo after all the years we have been in touch with you. I think it shows that many positive blessings can come out of having MPS in the family.

As you are aware, when Natasha became 19, she had to leave school. She went to a Day Centre, run by Parity, for three days a week. There, amongst others, she was cared for by an extremely caring young man.

Natasha weaved her magic and made sure he and her sister, Kirsten, met! The outcome of Natasha's help was that on 29 May 2004 Kirsten and Justin were married!

It was a beautiful service. During the prayers the Vicar included a prayer for Natasha without whom their paths may never have crossed.

The Bridesmaid's dresses and the church and reception flowers were in Helen House's 'Forget me Not' blue. Justin's cravat and waistcoat embroidery also carried on the theme. A wonderful reception was held at Camberley Heath Golf Club. The day was totally vegetarian and full of friends and family, love and dancing.

We are sure Natasha must have looked down with happiness, pride and joy to see her match-making had worked for her dear sister and new brother-in-law.

Wedding Bells for Colin Arrowsmith's sister, Claire

Clare Cogan

I met Claire for the first time at the Sibling's Weekend in July and can only describe her as the calmest bride-to-be I have ever met!

With only two weeks to go until her wedding she was cool, calm and collected. With recent memories of my own wedding in February this year, I don't remember a moment when I wasn't completely overwhelmed!

I was very glad to hear that the day went well and wish Claire and Dan every happiness for the future.

Do you have any stories or good news that you would like to share with us?
If so, contact the Society now on 01494 434156

New Members

Jane and Pete's daughter, Melissa, has recently been diagnosed with MPS I Hurler Disease. Melissa is 7 months old. The family live in North West England.

Lisa and Paul Martin's son, Toby, has recently been diagnosed with Hunter Disease. Toby is nine years old. The family lives in South West England.

Jennifer and Barry Quant's son, Lewis, has recently been diagnosed with Sanfilippo Disease. Lewis is nine years old. The family live in South West England.

Elizabeth has recently contacted the Society and has a diagnosis of Fabry Disease. Elizabeth is 65 years old and lives in Scotland.

Naveed and Saheera's son, Usman, who is 17 months old and daughter, Ayesha, who is two months old have recently been diagnosed with Sanfilippo Disease. The family live in the North West.

Caroline and Craig have recently contacted us about their daughter, Paysley, who has Hurler Scheie Disease. Paysley is six years old. The family live in Scotland.

Paul and Judith Black have recently become members of the MPS Society. Paul has been diagnosed with Fabry Disease. He is 40 years old and lives with his family in Nothern Ireland.

Diyal and Ann Nonis' daughter, Roshani, has recently been diagnosed with Sanfilippo Disease. Roshani is 7 years old. The family live in the South East.

Tim Hooper's son, Jamie Macfarlane, has recently been diagnosed with Sanfilippo Disease. Jamie is five years old and lives in the South of England.

Karen and John Coombs' daughter, Alice, has recently been diagnosed with Sanfilippo Disease. Alice is 2 years old. The family live in the South West.

Antony Selwood has recently contacted the Society. He is 44 years old and has been diagnosed with MPS I Scheie Disease. Antony and his wife, Pauline, live in Wales.

Deaths

Our thoughts are with the family and friends of:
Jacob Wragg 23 November 1993 – 25 July 2004

Disney Draw Winners

Congratulations to the 15 lucky families who were drawn out of a hat by the children of Chenies Primary School.

We wish you a wonderful time at the National MPS Society Disney Conference in December and look forward to hearing all about it!

Ingram Family (MPS I BMT); Fuzzard Family (MPS II); Logan Family (MPS II); Coney Family (MPS VI); Heath Family (MPS II); Rudham Family (MPS II); Oliver Family (ML II); Cooke Family (MPS I BMT); Lewis Family (MPS III); Mooney Family (MPS III); Pain Family (MPS I BMT); Longley Family (MPS IV); Preece Family (MPS III); McKnight Family (MPS I BMT); Hall Family (MPS II).

Births

Helen and Anthony Corcoran are delighted to announce the arrival of a healthy baby boy Archie Jake Corcoran on 13 August 2004.

Katie Elizabeth Heath was born on 23 July 2004. She is a sister to Jack who has Hunter Disease. Katie will be a carrier of MPS II but will not be a sufferer.

Jack Heath MPS II, and sister Katie

Congratulations to David Oulton, MPS II, for passing his driving theory test.
Well done!

Announcements

It's just a pity it's only once a year!

The Best Ever AGM!

Judith Evans

We have just arrived home from the AGM and fun weekend and I honestly think it was the best yet!

Following the great time we had at Alton Towers Hotel last year, we were looking forward to going again and to checking out the new Splash Landings Hotel. We were not disappointed! As soon as we walked into the Reception Area it really did feel as though we had landed in the Caribbean (although since we've never been fortunate enough to go to the real place, we can only imagine!).

Making our way to the room, the fantasy continued as we walked along the 'sea-shore' carpet and into our 'beach-side shack'. Joanne was thrilled to find she had a bunk bed to sleep in and immediately announced she would be sleeping in the top bunk. Well, what's the use of a powered wheelchair with rising seat if you don't use it to propel yourself onto the top bunk (please, nobody tell Dr Wraith!). Joanne soon unpacked by transforming the bedside cupboard into her own mini-bar with the bottles sneaked from home. Guess whose room was the venue for the late-night party!

With views over Cariba Creek Water Park and Ma Garita's cocktail bar 'just round the corner' we really did feel that we were on holiday and soon made our way down to the watery paradise. I don't think I can adequately describe the fun we had – you just had to be there! It really is the most fantastic place and yes, even I couldn't resist the tube slide and master blaster ride!

It was great to see so many families at the AGM and I am sure the background soundtrack of 'Finding Nemo' from the children's party next door made all the official business much more interesting for everyone concerned. This was followed by an excellent dinner and time for socialising with old and new friends.

We woke to a glorious sunny morning on Sunday and, having survived the breakfast scramble, made our way to the theme park. Joanne and her friends were way ahead of us and had probably been on ten rides before we even got there; not for us the white-knuckle rides like Submission, Nemesis and Oblivion. My adrenalin doesn't need to be pumped anywhere!

However, we enjoyed a gentle stroll through the gardens and a ride on the Sky Train, but later I was persuaded to join Joanne and her gang on the Congo River Rapids and the Flume where I got suitably soaked (they wouldn't let me wear my plastic mac). All too soon it was time to hit the road back to Scotland but we'd had a great weekend.

Thanks are due to Sam and all the staff at the office for their hard work in arranging such a successful and enjoyable weekend.

The AGM Evaluation

Sam Vaughan

The votes are in, and it's now official… the Alton Towers Fun Weekend and AGM was a raving success!

The MPS Society is constantly striving to provide a good service to its members, and the only way to know if we're on the right track is to do an evaluation. The questionnaire was sent out to 30 families, and 19 questionnaires were returned. The questionnaire concerned each family's experience of a number of aspects of the weekend, including the accommodation, entertainment, childcare and AGM venue.

The AGM Evaluation

95% of people thought that the Alton Towers was a fantastic venue for the AGM and 'couldn't fault the hotel'. The hotel provided fantastic facilities such as the Cariba Creek Water Park and the Alton Towers Theme Park, which kept all the families entertained! Some people did point out that our brochure stated that people could access Cariba Creek at any time over the weekend, but only Alton Towers on the Sunday; this was confused by the fact that people were receiving their tickets to Alton Towers when checking in, and they were the wrong ones! The hotel reception weren't supposed to be issuing tickets at all, so we do apologise for the confusion, and hope that next time the reception will be briefed more effectively!

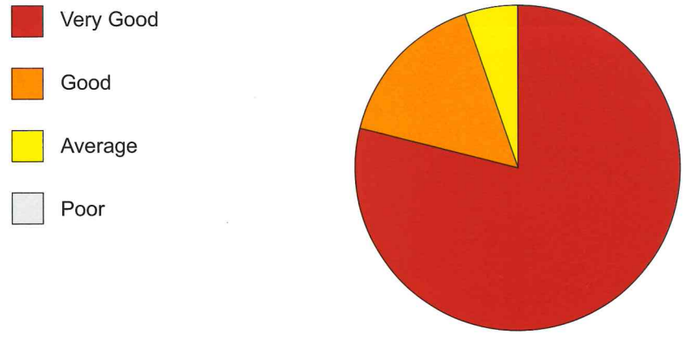
The AGM was also successful, but was subject to slightly more criticism. This was due to unforeseen noise levels from the children's party next door. At the time we did try to rectify the problem but there was a fault with the sound controls, and so therefore the music could not be turned down. We will try to avoid this situation altogether next time, by not putting the AGM next to the children's party!

A massive 79% of people thought that the adult's Gala Dinner was a very good event and 'one of the high spots of the year so far'! It provided parents of sufferers the opportunity to meet up with familiar faces, and meet new faces too. From the feedback we received, it was noted that Fabry sufferers who attended the AGM would have liked to have met and shared experiences with more Fabry sufferers.

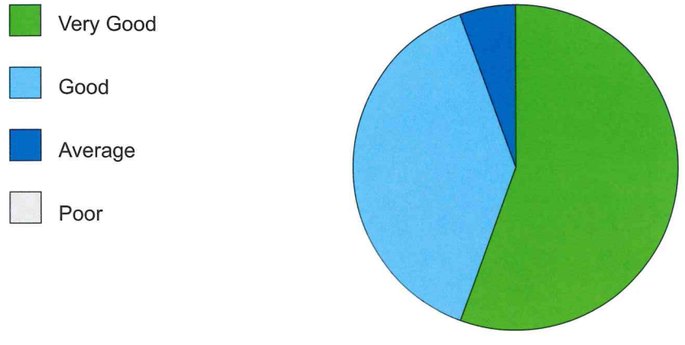
The children's gala tea was just as successful with just under 90% of people saying it was very good. Although, it was noted that the 'entertainment was more for younger children', and we will try to combat this in the future by providing more for the teenagers who attend.

I am pleased to say that everyone was unanimous in saying that they would definitely attend the next AGM Fun Weekend. We had such a great response from everyone which was very encouraging, and the feedback indicated that it was a fantastic weekend, and as one person put very aptly, 'it's just a pity it's only once a year!' ■

Gala Dinner



Hotel Accommodation



Events

The Sibling Activity Weekend by Clare Cogan

As you can see from the articles on the following pages, the Sibling Weekend was fun-filled with a variety of activities, the most enjoyable ones seeming to be where water was involved!

It was disappointing that only ten children and young people ended up attending as the event was planned to cater for many more. I hope, in light of the positive feedback which we received from the children and information from the families that having 'something for the siblings' is important, that any future event will be more popular.

Unfortunately, we are unable to run any future sibling events on this small scale due to the time it requires and restrictions regarding the funding which we receive. The MPS Society would be very happy to run a siblings weekend in Spring 2005. However, it is unlikely that we would be able to secure a venue to fit in with school holidays at this late stage, so parents need to be mindful that any weekend arranged would involve their children taking the Friday off school if they wanted to participate.

If we don't hear back from you about whether you want this event to go ahead then we will have to return the funds received from BBC Children in Need which were awarded for this specific purpose. It is also unlikely that we will revisit this area of support in the future. **Therefore, if families, children or young people want something to happen in the future I need to know, and soon!**

Nikki McAuliffe gives us a Volunteer's Perspective

Well, what an amazing weekend. I'm usually quite an energetic person, but I must admit by the Sunday I was ready to go into hibernation for a month! Let me give you a sneaky insight into our sibling activity weekend.

Friday, day one. I set off from my home in Reading at 7am. Seven long hours later, via Ely, I arrived at the Pioneer Centre. The afternoon began with an ice cream challenge, which meant that we could preserve our energy for the next day as we were anticipating quite a physically challenging day of activities. After dinner we headed off for a very entertaining evening around the campfire, which involved a lot of singing, shouting and banana-munching!

So into Saturday, bright-eyed and bushy-tailed we headed off for our first team activity, bridge-building. I think I'm safe in saying that there was a unanimous sense of achievement amongst the group once our pulley bridge was erected, especially when we all took a turn in being pulled across the rope!

Saturday afternoon was spent being slightly more competitive in as much that we were attempting to make possibly the best raft ever. Well, the raft was indeed a structural masterpiece, on land! When it came to hauling it into the chocolate coloured water, it quickly became apparent that the raft was not as sturdy as it looked. Nonetheless, with a little perseverance coupled with a lot of giggling and teamwork, we all managed to pile onto the raft.

But, there's always one isn't there! The one cheeky chappy who can't wait to tip the raft and get absolutely soaked, mentioning no names (Ian!). So, what with Siobhan's threats to sue if she got wet, the smell of icky pond water and the continual shrieks of laughter, it turned out to be a very entertaining and memorable day.

This takes us now into our final day and the activity I was most dreading, the zip wire. I will take this opportunity to once again give all credit to those of the children that did (and attempted to) brave the 30-foot climb and launch themselves down the wire!

So there we have it, a potted view of the siblings weekend. I can't say it enough, it really was a fantastic weekend and we even had the weather on our side. I feel privileged that I have met such a fantastic group of siblings and I definitely wouldn't turn down an opportunity to be part of an activity weekend in the future.

Kirsten Hill wrote to let us know what she thought...

The Sibling Weekend was great. All the activities were different and great fun for everyone. It was also good to meet new people who also have brothers and sisters with MPS, and also old friends, which I had made at the conferences a few years ago.

My favourite activity was raft building because we all got really wet and laughed so much when people fell in! We had a few wooden poles, big blue barrels and loads and loads of string and we had to think up a design and a raft which would carry our whole team across a big, muddy pond.

When we had to put the raft in the water, all the barrels popped out of the raft and floated off so people dived in to rescue them to stop the raft from sinking! Once the raft was floating we all jumped on but eventually everyone got pushed in, and our raft collapsed so no one was dry at the end! It was so much fun.

My Sibling Activity Weekend by Joseph Flaig

When I arrived on Friday, we had an hour's free time to explore the huge grounds in the Pioneer Centre. It was brilliant!

There was an excellent shop which sold souvenirs of all kinds, from karabiners to juggling balls, torches to key rings. There was a basketball court that I used frequently, a football pitch, swings, and in our main building, a lounge with a TV, sports equipment, pool table, refreshments, football table and loads of chairs.

Joseph Flaig wrote in to say how much he enjoyed the sibling weekend. Here he tells us a little bit more about what happened.

At 3pm, we started on our first activity, the ice cream challenge! This involved a treasure hunt, a scavenger hunt, a quiz and collecting staff signatures in teams to win different ingredients and equipment for a bowl of ice cream. I really enjoyed this activity, especially the ice cream bit! After an hour of playing basketball, it was time for dinner, which was my favourite, fish and chips!

After that we had half an hour to get changed for the campfire. The campfire was in the middle of the wood, accessed by a winding path. There were nine benches in groups of threes set around the campfire. There were loads of songs, tricks, activities and games, and I think I took part in all of them. My favourite was the banana duel. After a cup of hot chocolate, it was back to the lounge and then to bed.

In the morning we walked over to the Red Deer for breakfast. There was an excellent choice of cereals, yoghurts, and jams. After this we washed and prepared for our group's first activity; raft building. We walked down the field and across the road to the lake to see our materials before going back to the hall next door to the Red Deer to start planning. I put forward my idea, but it was narrowly defeated by Gary's.

We went back down to the lake and started building our raft and I even learnt some new techniques. Eventually, we managed to finish the raft and launch it onto the lake. I went on it, rowing at the front. However, it started to sink and Ryan and Emma fell off. I didn't get wet though because I jumped onto the bank. After dismantling the raft, we went back up to the Red Deer for a lunch of jacket potatoes and baked beans.

We finished lunch, then went to look at the bridge building materials, and started our planning before actually building anything. We settled on two tripods with a piece of rope between them so that we could be pulled across.

We then went back to the Red Deer for dinner (chicken with vegetables) before swimming. I really enjoyed swimming in the heated outdoor pool, trying desperately to get on top of an inflatable dolphin, and then playing some kind of water volleyball. After that, we went back to the lodge to get showered before bed.

In the morning, we walked over to the Red Deer for breakfast, before starting our first activities. First my group had a go on the zip wire. Seeing as I am afraid of heights, I was absolutely petrified. What made it worse was climbing rung after rung up a huge telegraph pole and standing on a 1m square wooden platform with another person. However, I overcame my fears and I was the first to go on it! I still really envy Gary, who went upside down.

After a treasure hunt that my team won, it was time for the last meal, a roast dinner, before it was time to go. I really enjoyed the whole weekend. Thank you to Clare for organising it, and Nikki and Simon for helping, looking after me and generally being nice.

Page 12: Selection of photos from the Sibling Weekend. Page 13: Photos clockwise from top right: Nikki, Claire Conway, Hollie and Annie Stuart; Ian Grandidge. Page 14: Joseph Flaig and Gary Devine.

Remembrance Day at the Childhood Wood

Ellie Gunary

On 11 July 2004 eleven families met at the Childhood Wood to remember family members who had died from an MPS or Related Disease. Amongst these families were those who had planted their tree at the first planting eleven years ago and one family whose child had died very recently and will be planting a tree at the next planting in October this year. These members, Sharon and Linda, wrote after the event…

'Thank you for inviting us to the Remembrance Day. It was nice to see everyone again albeit under the circumstances. This was our first visit to the Childhood Wood. We found it very peaceful and moving and we will be honoured to plant a tree for Thomas in October'.

Whilst the wood is a peaceful place, giving opportunity for reflection, it was also pleasing to see Sherwood Pines where the Childhood Wood is situated being increasingly used by local families, cycling, walking and generally enjoying summer afternoons outdoors. Eleven years on the wood is growing, the older trees are taller and natural undergrowth pervades the area giving a much more established feeling.

After a slightly delayed start to the afternoon, caused by rather longer journeys than anticipated by some who became lost along the roads of Nottinghamshire, Barry Wilson, Chairman of Trustees, welcomed everyone to the Wood.

Photos clockwise from top right: Andrew & Vivienne Culley; Henry & Julia Duckmanton; Gordon & Alison Rowe

Wilma Robins read the poem 'Remember' by Christina Rossetti and I read the names of the children and adults being remembered by the families present.

The remainder of the afternoon was spent remembering together and catching up with each other over lunch, which was provided by the local café in a marquee nearby.

My thanks go to those members who pitched in and helped set up the tables and chairs and to Angela and Andy Seymour who fetched more hot water for teas and coffees when it was needed.

All too soon it was time to set off home which, for those of us returning to the London area, was quite a slow and arduous journey with our timing coinciding with the end of the Grand Prix at Silverstone.

REACT come to the Rescue

Robyn has Hurler Disease, MPS I. She was desperate to get out and play. But there were five big steps down to her small garden, which was very uneven, and Robyn was always falling over and hurting herself.

As her joints became stiffer and her hearing and sight were failing, going out into the garden became just too dangerous.

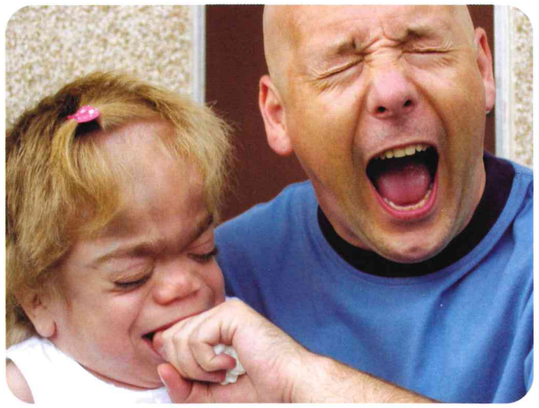
React came to the rescue by providing some simple decking, which gave Robyn a very safe area to play which was in full view of her mum and dad. Her dad Gilbert said 'We could put the paddling pool out there and let her splash around and enjoy the sun.'

Sadly, Robyn's condition is worsening all the time and she has now lost all mobility. Most children with Hurler Disease do not survive beyond their tenth birthday and Robyn is now nearly eight.

'We still get great use from the decking though' says Gilbert. 'Robyn loves to sit outside in her wheelchair and feel the sun and the breeze on her face'.

React has recently been able to help Robyn once again with a hydrotherapy bath. 'The warm water is the only form of physiotherapy available to Robyn because of the stiffness in her joints. They can be so painful but the water relaxes Robyn and gives her a chance of a good night's sleep.'

We went to so many people but only React could help. Gilbert told us that without React they could never have afforded the bath and Robyn uses it every day.



Editor's Note: This article appeared in the Summer '04 newsletter published by React. We have kindly been given permission to reproduce it here.

Want to learn more about React? Check out page 36 for more information

Roma Drayne gets a place at Grammar School

Bernie Drayne

Bernie Drayne wrote to the Society recently to update us about her daughter Roma (MPS IV).

Roma has successfully been admitted to Friends Grammar School this September. Bernie will do an article for the next newsletter as she felt it would be useful for others to read about what can happen when the MPS Society and the local MPs get involved and challenge the system.

Bernie writes 'I think the experience has made the education board really take notice of what is going on although they won't admit to their shortcomings.'

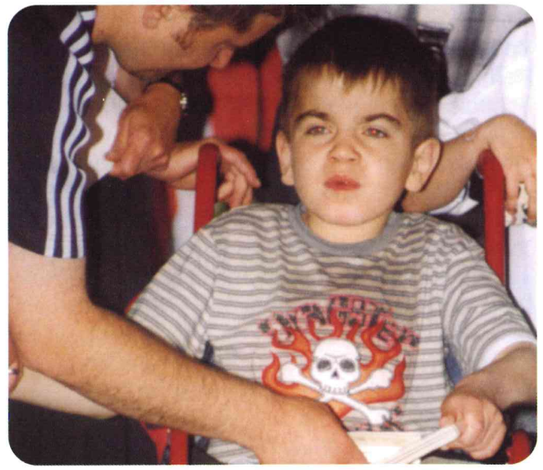
Aaron's Story by Michelle and Adrian Craig

Michelle and Adrian have three children. Aaron is 11, Ryan is 9 and Liam is 2. This is their story.

Aaron was born a healthy baby weighing 8lb 1oz on the 8 July 1993. We were so happy. Aidie wanted a boy, I would have liked a girl but after a long 14 hour labour and a ventouse delivery we were over the moon when Aaron was born healthy.

Aaron developed normally but at the age of 12 months he had glue ear and overgrown adenoids and was very poorly most of the time. This went on for a number of years. By the time he was 3 years old his behaviour was terrible.

'Aaron's health was a constant battle, he never seemed to be well.'

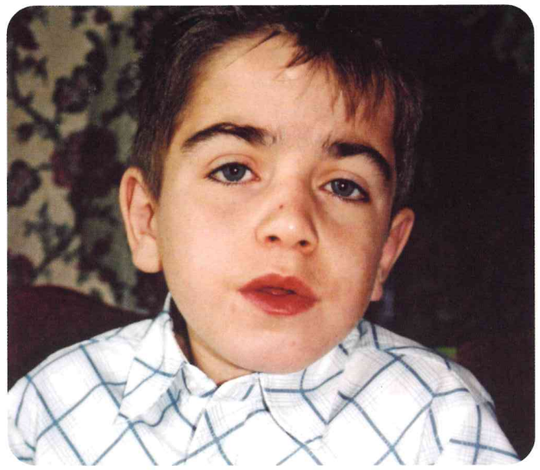


When he reached the age of 3 he was 'statemented' as having moderate learning difficulties, delayed speech and behavioural problems. Aaron went to a special school, which catered for children with these needs.

At the age of 6 he was then diagnosed as having Autistic Spectrum Disorder. Although we were not convinced that this was the right diagnosis, we settled for it.

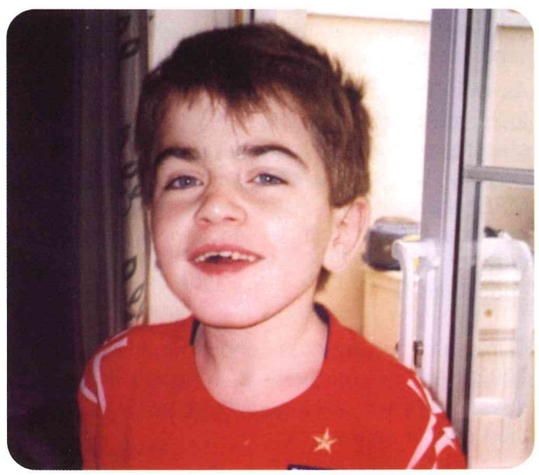
'It is very hard to live with a child who has special needs but no diagnosis.'

Aaron started to lose his limited speech and showed signs of mobility problems. Following this, he was diagnosed with a disease called Multiple Epiphyseal Dysplasia, which is a crippling joint disorder. In September 2003, Aaron underwent major hip surgery to help increase his mobility.



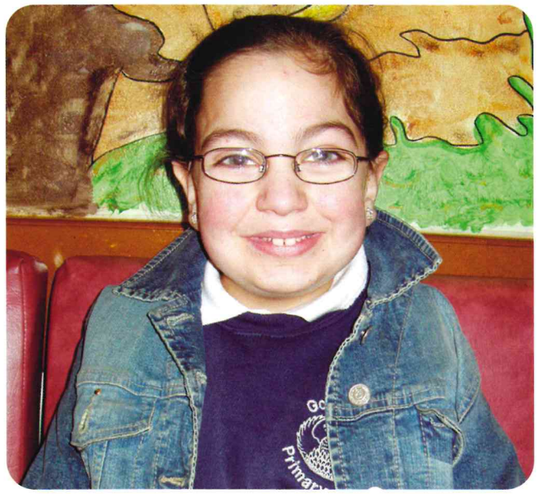
In February 2004, after a routine annual medical, the school doctor felt that Aaron had some characteristic facial features and also features of his developmental progress, which were striking to her. A urine sample was tested for mucopolysaccharidosis and in April 2004 it was discovered to our shock that Aaron now aged 10, had in fact got Sanfilippo Disease. This was such a shock. At first we didn't really understand but after lots of research and help from various sources we now have to come to terms with what lies ahead.

There are still a lot of uncertainties for Aaron, but with the love and support of his family we hope to make his quality of life comfortable and enjoyable. Thankfully, by pure luck, Ryan and Liam are not affected.



Emma's Transition to High School

John and Emily Slater



Emma was at Gorsefield Primary for about 6 years in the Resource Provision (RP) unit.The teachers and children knew about Emma and her problems and she was well liked by all, especially the RP unit children. Emma loved it there. This was one area we didn't have to worry about.

The main thing was to make sure she would be happy going to school every day and this school provided that.

We actually started looking for a high school two years before Emma was due to start, when her sister Samantha was about to change to a high school. At the parents open evenings, our main concern then was Samantha, but we checked out all the special needs RP units at all the local schools. This reduced the number of schools from 5 to 3.

As with any child going to high school, it's a big step for both the child and parents. But with a child with MPS there is so much more to consider. Not only is it the right school but also can they cope with Emma's needs physically and educationally.

Our search for the right school began in October 2003. We set out our own guide lines and questions which we would apply to each school then decide on the best.

The school Emma would feel most happy at was entirely her choice (but we would obviously advise her of the good and bad points). The school had to accommodate Emma i.e. not too many stairs, not too big, as Emma is currently having problems with her hips and knees and may in the future need wheelchair assistance and, if possible, a school with an RP unit as Emma has done so well in this small environment. A school that would take a genuine interest in Emma and her needs.

After looking at the initial three schools we soon narrowed it down to two, one of which was highly recommended by Emma's current school and it was taken for granted that she would go there. But Emma wasn't too keen on this school, and rule one, was a school where Emma felt happy.

This now only left one school - Tottington High. Emma liked it, it had a RP unit and the staff seemed helpful and interested in Emma, which hadn't been the case at some of the other high schools we'd looked at.

After selecting the school we now had to sort out the statement and get what was required for Emma. At this point we asked the MPS Society to give us the benefit of their experience. Ellie and Clare attended the review meeting with us (thank you both). Previously at the school open evening we had asked for a rep from the school to also attend. They sent 2SSA teachers who would be directly involved with Emma.

First impressions were good. All went well at the meeting, some good points were raised. Ellie and Clare wanted a more detailed explanation of MPS I and I'm pleased to say it is now being included in the statement.

Emma has a diagnosis of MPS I, Hurler Disease. This is a well documented, progressive, degenerative condition leading to progressive damage directly to the skeletal system, joints and major organs. In this rare metabolic disease, certain essential enzymes are absent. As a result, complex sugars remain stored in the skeleton and organs of the body causing progressive damage.

Symptoms of Hurler disease include inguinal or umbilical hernia, enlarged liver and spleen, severe skeletal deformities, short stature, joint stiffness, heart disease, recurring airways and ear infections, obstructive airway disease, deafness, corneal clouding and severe learning disabilities.

Emma has undergone a Bone Marrow Transplant (BMT), which has significantly reduced the progression of some aspects of her disease. The liver and spleen, heart disease and learning disabilities are positively affected. However, the bone disease continues to progress post BMT. Emma has the skeletal disease associated with MPS I, including scoliosis of the spine. It is widely recognised that children with MPS I who have had a bone marrow transplant may need significant levels of support to promote a healthy psychological and social development.

Also added was future support if needed: Ongoing opportunity for Emma to have access to psychological support to meet her psychosocial needs; Regular review of physiotherapy to ensure that these needs are being met; A system of support to ensure that Emma's needs are met during lunchtimes e.g. to carry her tray and during games/PE e.g. to help with dressing and tying her shoe laces.

Also discussed was seating for Emma both in class and science labs. Again the school were prepared to solve this problem by putting a chair in every department for Emma. Another of Emma's needs was a posture pack, a pack that has an angled seat cushion and an angled folder so Emma doesn't have to look down all the time, relieving neck pain. Again the school said they will put one in every department.

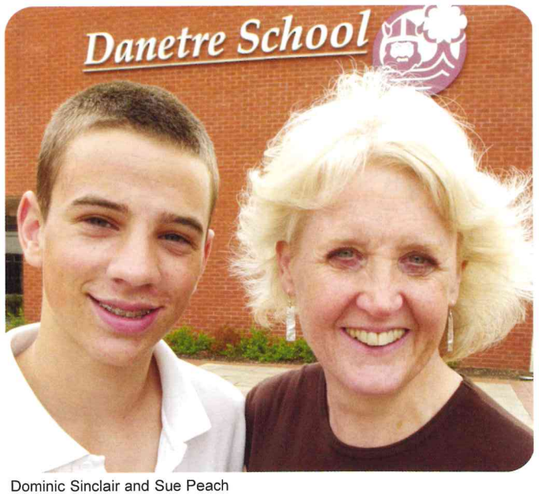
It seems too good to be true but time will tell. Due to all the input from Ellie and Clare, the two teachers decided they needed more information so asked the MPS Society to visit the school and give a presentation to all teachers so they would all be aware of Emma's needs. This will take place on the 2nd September, the day before school opens.

The two teachers invited us to go into the school and have a walk around each class Emma would be using to see if all was ok and discuss the seating plans, and any other concerns we had. By this point we knew this was the right school for Emma.

Everything has seemed to go ok up to now. The school is on one level apart from the art room, which Emma would use once a week, and Emma is looking forward to starting high school. Thanks for your help getting Emma into high school. ■

If you would like support from the MPS Society's advocacy support team please contact us now on 01494 434156

Raising funds for Jeans for Genes



Dominic Sinclair and Sue Peach

A Daventry schoolboy has become the apple of his teacher's eye after raising more than £1,000 for a charity close to her heart. Dominic Sinclair, 15, from Newnham, raised £1,250 in less than a year for the charity Jeans for Genes.

His Danetre School English and Drama teacher, Sue Peach, became a trustee of one of the four founding charities of Jeans for Genes following the loss of her daughter Elisabeth to Hunter Disease.

Mrs Peach, 54, said: 'Elisabeth died in 1984, when she was four years old and five years ago I became a Trustee of the Society for Mucopolysaccharide Diseases. As a parent of a child lost to a genetic disease and a Trustee, I have been helping to raise funds and the profile of Jeans for Genes and give regular talks at school about it.'

Caravanning enthusiast Dominic said: 'After I heard about Mrs Peach's daughter dying from a genetic disease I decided to donate the money I raised to Jeans for Genes.' Dominic, who is the junior and teenage representative of the national Elddis Caravan Owners Club, raised cash through events he organised for the club.

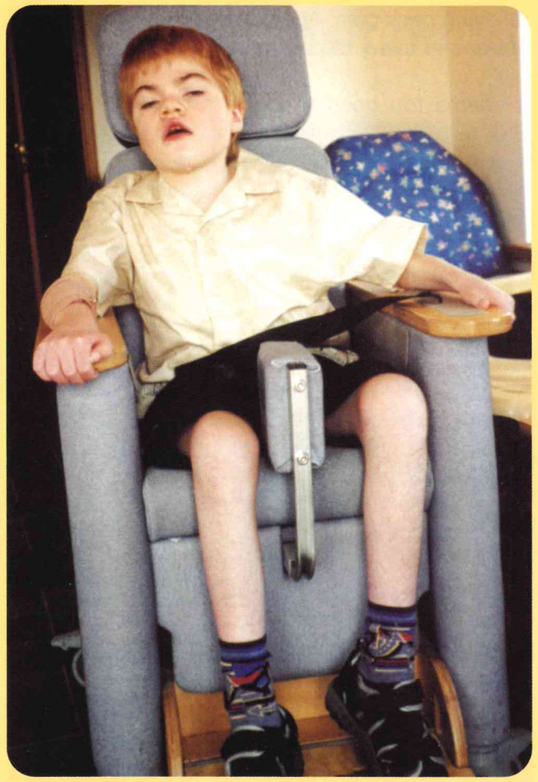
Mrs Peach said: 'I was amazed when Dominic told me what he had done. I am very proud he has taken this to heart and raised money for what is such a worthy cause.'

Jeans for Genes is a national appeal to raise money for a number of charities by inviting people to donate a pound in exchange for wearing jeans for a day.

To get involved with the Jeans for Genes campaign call free on 0800 980 4800

Editor's Note: The article & photo are reproduced with kind permission of the Daventry Express, July 2004

Members' Stories



Life with Kyle

by Alison Shields

Kyle will be twelve on 18 October 2004. He was diagnosed with Sanfilippo Disease, MPS III, on 31 October 1996, when he was just 4 years old, a day seared into our memories for life.

Things have changed a lot since then, Kyle is no longer the uncontrollable child he once was. It has not been easy, as we have watched him deteriorate over the years. We have watched him lose his speech, lose his ability to run and jump and generally cause havoc. He can no longer walk on his own and needs assistance, tiring easily.

Kyle started having seizures in April 2003 and since then I now sleep with him, in his room every night. His sleeping pattern is still very irregular. Thankfully my mum usually stays one night of the week so I can enjoy a full night's rest.

Kyle's swallowing has also deteriorated going from a normal diet to mashed foods and now a liquidised diet and thickened drinks, but he still has problems swallowing. We moved house in December 2001, staying in the same area, but moving from the shore to the foot of the mountains. We had no joy with our plans to adapt our old home, we just seem to hit one obstacle after another.

My mother-in-law lived next door to us on her own and we decided to sell both houses and build a property that would meet all our needs.

It has worked out brilliantly. Andrew's mum has her own living area; kitchen, bedroom and bathroom and we have been able to provide Kyle with a large bedroom with ensuite and overhead hoist. Kyle also has a multi sensory station provided by 'Make A Wish Foundation', a wonderful organisation dedicated to fulfilling the wishes of terminally ill children. They also provide what they class as comfort wishes for children who are unable to express what they would desire.

Kyle loves his equipment, which has a bubble tube, fibre optic strands which change colour and enclosed in the beanbag are two large speakers, which vibrate to music when plugged into a HiFi. His room also has a camera mounted in the corner so that when he is in bed we can watch him on any television in the house.

The new Children's Hospice opened in Belfast in October 2001. The facilities are brilliant and the staff are really caring and dedicated. Unfortunately it is a 1½ hour drive for us and, as it is the only one in Northern Ireland and there are so many families in need, respite is extremely limited.

Kyle did receive respite, two nights per month and during holiday periods, locally at Orana in Newry. He has been going since he was diagnosed almost 8 years ago now; but this respite has now been terminated.

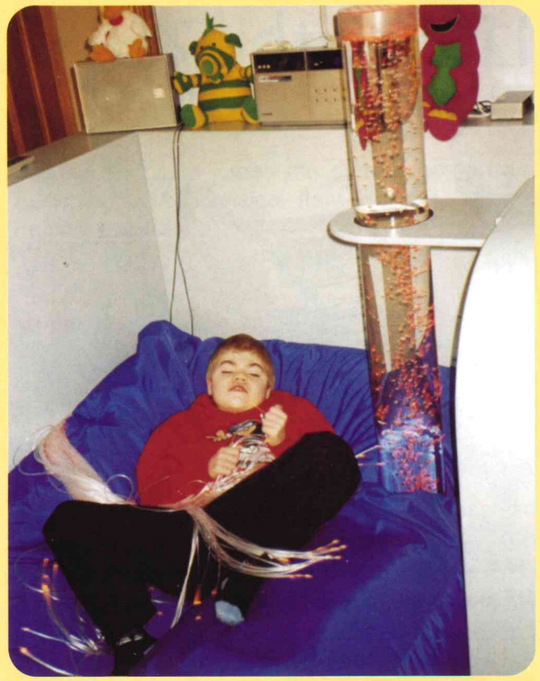
Kyle had to have a videofluoroscopy in April because of his swallowing problems and weight loss. This identified a high risk of aspirations and it was recommended that alternative feeding methods should be considered urgently.

Once Orana heard these results Kyle's respite was suspended and a risk assessment requested. Our social worker thought there would not be a problem, the assessment was completed, then on 15 June at Kyle's review we were told that Orana could no longer meet Kyle's needs as they had become too substantial and they had not got the nursing staff. We had not expected this as I had two weeks holiday planned and we were depending on Orana for respite.

Fortunately, the Children's Hospice was able to take Kyle for 5 nights and my sister took leave from work and flew over from her home in Derbyshire to keep Kyle for the remaining 9 days and nights. To her we will be eternally grateful, she would not hear of us cancelling, guessing she realised how much the family were all in need of a break.

Kyle is due to go into the Royal Belfast Hospital for sick children on 24 August in preparation for his gastrostomy the next day. I am still a bit apprehensive not only about the operation but about coping with administering his feeds afterwards and looking after the wound site. I know it will provide a long-term method to ensuring that Kyle gets all his nutrition, fluids and medications safely. I will fill you in afterwards with how it goes.

I know we face difficult days ahead; we all love Kyle very much and do not want to lose him. I do not think anyone is ever ready to lose someone they love.We grow up expecting to lose grandparents and perhaps parents on some far away day but not your little boy.



We treasure each moment we have with him. He is one very special little boy. ■

Lisa Nurse celebrates her 33rd birthday!

Hi everyone! Here we are again celebrating Lisa's 33rd birthday. It seems only yesterday that we celebrated her 21st. We are so proud of her.

As most of you know, Lisa is in a nursing home now between Monday and Thursday and comes home Friday til Sunday. At the home she gets spoilt by all the care staff and residents as you can see from the photos here.

Our thanks go to those who sent cards and good wishes to make Lisa's birthday a day to remember.

Also, we have just come back from Butlins Holiday Camp at Bognor Regis. The weather was great and Lisa enjoyed herself!

Lisa Nurse has Sanfilippo Disease, MPS III. Her parents kindly sent in some photos and a story about Lisa's 33rd birthday celebrations at Newington Court.

Members' Stories

Life with Ben and Jake

by Sheryl Lloyd

No one pretends that life is going to be easy when looking after a child with Sanfilippo Disease, but having two children is tiresome. Ben is nearly 7 years old now and Jake has recently celebrated his 4th birthday (8th August), ahh bless.

Jake had a lovely time at a small party organised in his honour, with those close to him, partying away. I have found it is difficult organising parties with children who are not affected by the disease. This is because the behavioural differences associated with Sanfilippo, the hyperactivity and tantrums, may not bode well with others. Ben and Jake often display such behaviour. That is why we had our friends Gail and Nick, who have a daughter Faye, who is also affected with Sanfilippo, at the party.

But Ben and Jake are typical brothers. One minute they get on, the next they cannot tolerate each other. This normal love/hate relationship is heightened by Sanfilippo, making life very interesting for us.

Their playroom (a conservatory built on to the house) is every child's dream, with a 42" plasma screen television with DVD and video, a ball pool, soft play area and much much more. Yet this is just inside! Outside they have a complete adventure playground. They have a big hut with a telescope (just so they can watch out for us parents). Also attached is a drawbridge-like ramp connecting a wavy slide and ladder. Hours of fun to be had, if only the weather were not so bad!

Come September and Jake will be starting full time school. Exciting stuff as he will join his older brother at Reynolds Cross, both catching the morning bus together! Time will only tell how the two brothers will get on. For me this will be some well-earned time out to relax a little and time for me to do things for myself.

But not for long… There are holidays to go on, and one very special trip has been planned for October. A trip to Florida for two weeks, with Nan and Granddad coming too. Not only are Ben and Jake going on a transatlantic trip, but Ben will also be swimming with dolphins. Unfortunately for Jake you need to be 6 years old, but Stuart, granddad and myself will be making the most of it, joining Ben in the water for a truly magnificent experience. The one thing that could be challenging is the nine hour flight there and back. However, I have been weighing up the options, to see how two such happy yet hyperactive children can be entertained, and I have Stuart's mum and dad to hand.

Living with Ben and Jake can quite often be chaos. There are good days and bad days, days when they will be calmer and days when they are constantly hyperactive.

Life is definitely different and I think you make the most of what you have, and do as much as you can.

Both brothers have travelled extensively this last year visiting Paris on a number of occasions and soon off to Cornwall for a week. The architects will also be round in the near future to sort out adaptations for having extensions to our two bedrooms, a bathroom and a lift up from the garage. There is no stopping in our household!

Joanne Evans wins Childnet Academy Competition

Judith Evans

As wheelchair users, Joanne and Zara felt that the resources available for young people like them could be patronising and stereotypical so when the charity Whizz Kidz announced their No Limits award scheme (funded through the Lottery Commission) they agreed this would provide a way to change things.

With the help of their grant they invested in web design software and went on a one day course at South Bank University in London to learn how to use it.

On their website (www.wheelg2life.info) they provide information on benefits, housing, education, social life etc. This is information that is important to disabled youngsters.

Last year, Joanne Evans (MPS IV) and her friend Zara Todd applied for a Whizz Kidz No Limits Millenium Award to create a website that would act as an information bureau for young disabled people.

Creating the website was a huge challenge particularly as both Joanne and Zara were studying for important exams at school at the same time. Joanne wrote a press release about the site and managed to get it mentioned in several disability publications as well as getting linked to other disability-themed websites. Joanne also did a radio interview which was broadcast throughout the day. So far, they have had over 1,300 hits on the site.

Joanne entered the site for the 2004 Childnet Academy competition and was thrilled when she heard that the site had been chosen as one of twelve winners worldwide.

Joanne and Zara joined the other winners from Australia, USA, Jamaica, Italy, Holland, Taiwan, Ireland and the UK at the Cable and Wireless Childnet Academy in London for a week in April.

The week was a great mixture of work and play with workshops covering all the different aspects of web design, internet use, PR etc. They were run by experts in each particular field, combined with visits to the BBC and Virgin Radio.

Joanne and Zara spent a fascinating day at Cable and Wireless and attended workshops covering everything from the history of the internet up to future developments of the web & communications in the 21st century.

This culminated in the Awards Ceremony and party in the evening with over 200 guests. There was plenty of time for socialising and sight-seeing trips around London, a river cruise on the Thames and yet another 'spin' on the London Eye.

Perhaps the most positive outcome of the week was all the new friends that Joanne made worldwide and with whom she is now in regular contact via the Internet.

Cable and Wireless created the Childnet Academy in 1995 with the aim to help make the Internet a great and safe place for children. They have sponsored the awards and Academy for over 6 years. (www.childnetacademy.org)

Left to right: Gareth Jones (MD of UK Corporate Markets Cable & Wireless), Joanne (MPS IV), Helen from Childnet Academy, Lizo from BBC Newsround

8th International Symposium on MPS and Related Diseases

Ellie Gunary

On Thursday 10 June 2004 five members of MPS staff, Clare Cogan, Sophie Denham, Christine Lavery, Cheryl Pitt and I, set off for Germany to attend the 8th International Symposium on Mucopolysaccharide and Related Diseases in Mainz.

The Willink Unit Team from Royal Manchester Children's Hospital

The family programme began on Thursday afternoon with a session 'From gene to therapy' which addressed the principles of genetics, genetic counselling, and the principles of therapies including Bone Marrow Transplantation, Enzyme Replacement Therapy and Gene Therapy.

After an evening of networking and meeting with other delegates who included all the eminent medical and support professionals involved in the field of MPS and Lysosomal Storage Diseases worldwide, Saturday began with a morning session chaired by Christine Lavery.

We heard a moving account from a mother whose daughter with MPS I Hurler Disease was receiving Enzyme Replacement Therapy and perspectives from a doctor and from a patient association, with Zsuzsanna Román speaking about the evolution and support offered by the MPS Society in Hungary.

Barbara Wedehase & Steve Holland from the US MPS Society

Cheryl's talk on the psychosocial outcomes of BMT for individuals affected by MPS I was well received and gave all present food for thought as she shared some of the findings from her pilot study.

After lunch on Saturday there was a choice of sessions from one looking at management of bone disease, neurological aspects, pulmonary function and attention deficit disorder, one looking at anaesthetic, cardiac involvement, ophthalmology and hearing and a third session looking at disease patterns in each of the MPS Diseases. Genzyme ran an interactive session which considered ERT treatment for different patients with MPS I and TKT ran a session on ERT for Fabry Disease.

Representatives from the Polish MPS Society

Between sessions each MPS Society had a stand with display materials and booklets for delegates to visit and from which to obtain information. We had many visits from international MPS families and provided support and information as requested.

This time was also used to meet with the pharmaceutical company representatives present to discuss support to ERT patients and achieving funded ERT, with the International MPS Society leaders and with clinicians and researchers from other parts of the world, all invaluable in informing the support we offer here in the UK to MPS Society members.

The highlight of the gala dinner on Saturday evening was the entertainment. Siblings throughout the conference had participated in a circus workshop and their opportunity to demonstrate the skills they had learnt came after the final course had been served and coffee was being drunk. We were all highly impressed by the skills demonstrated by the children on unicycles, juggling, balancing and as clowns. The loudest round of applause for the whole weekend certainly went to this group of children.

The International Symposia are held every two years and towards the end of the gala dinner the Italian MPS Society announced that the next International Symposium would be held in 2006 in Italy. ■

Dr Charles Peters, Barbara Lyons and Matt Patterson

A Parent's Perspective by Derya Gok

Dilaria was born on 9 February 1996. Prior to her birth in September 1995 I underwent prenatal diagnosis and it was indicated that the baby was OK. Very soon after Dilaria's birth, in fact when she was only four weeks old, she was diagnosed with cardiac problems and underwent heart valve surgery in June 1996. At this time Dilaria was diagnosed with a kyphosis and in August 1996 her cardiologist referred her to a paediatrician on suspicion of a Mucopolysaccharide disease.

On 5 December 1996, at ten months of age Dilaria, was diagnosed as MPS I, Hurler Disease, by Professor Michael Beck in Mainz. At the end of December Dilaria was tissue typed for Bone Marrow Transplant but no donor was found. This was our last hope.

In January 1997 we said to ourselves 'Alright we need to go on somehow'. Dilaria had her tonsils and adenoids removed and her umbilical cord hernia repaired. Life went on. Her facial features changed over time, her tongue got larger as did her liver and spleen.

In November 2001 it was suggested that Dilaria might benefit from Enzyme Replacement Therapy and on 7 May 2002 news was received from Professor Beck that there was a chance of Dilaria receiving ERT even though the drug was not, as yet, approved in Europe. What was there to lose? If Dilaria did not have ERT she was highly at risk of having a cardiac infarction and had already stopped walking.

On 22 August 2002 Dilaria had her first infusion of enzyme. Whilst waiting for ERT it was our permanent nightmare that Dilaria would die before treatment came. We sought the views of many people. Medical opinion was divided. Our paediatrician had always been there for us. He was very objective and always treated Dilaria with courtesy.

So where are we today? At the age of 8 years 4 months, Dilaria is only 102 cms in height. However she can now run, jump and skip. Her joints have improved and Dilaria has much better co-ordination. Her tongue is much smaller, her heart problems have receded and, although she still needs hearing aids, this is at a much lower level.

Unfortunately, Dilaria's intellectual development has not improved and indeed her progress as predicted for ERT has not opened up the brain barrier.

Before ERT was approved, the family travelled from Dortmund to Mainz, a round trip of 400 miles. Dilaria has a sister and this meant she too had to make that journey weekly. Now Dilaria has her ERT very much closer to home in Munster.

There was mental stress and feelings of disappointment but now the family feel that 'Dilaria is doing fine and if she is doing well, we are good as well'.

Derya expressed her thanks to everyone particularly the German MPS Society, Genzyme Corporation and Professor Beck and his team at the Kinderklinik, Mainz. Derya finished by saying that she would do everything she can to get assistance for Dilaria and she is happy to accept help.

Editor's Note: This article is taken from a presentation made at the International Symposium on MPS and Related Diseases, June 2004, Mainz, Germany.

International News

International MPS Societies Leadership Meeting &
International Perspective on ERT for MPS I

By Ellie Gunary

During the International Symposium for MPS and Related Diseases, held in Mainz, Germany, June 2004, the International Societies Leadership meeting was held. These meetings provide an opportunity for the Chief Executives, Presidents and Chairs of the international MPS Societies to meet, share ideas and discuss pertinent topics to the field of MPS.

Three items for the agenda had been agreed beforehand: managing the media, fundraising ideas and ERT update. Presentations were made by all the groups represented from those such as the recently formed Spanish MPS Society to much more established groups such as ours in the UK.

From the session on managing the media, training offered to one group by Genzyme was identified as having been particularly helpful and it was agreed to investigate whether a training session could be held at the next International MPS Societies meeting for all the groups. All the MPS Societies share common aims which include raising the profile of MPS and Related Diseases and the needs of members which makes media training of international interest.

Each group then shared ideas on fundraising, giving details of successful events. Christine Lavery spoke of the UK collaboration with other charities in the Jeans for Genes campaign and we learnt of sporting events such as 5K walks, mini marathons and sponsored events being held.

After hearing other countries experiences of achieving ERT for MPS I, I gave details of the UK's experience. It was interesting to learn of each country's different health care arrangements and the different challenges faced in achieving funding for ERT on an international level.

Ellie Gunary and Dr Uma Ramaswami

Representatives from the Italian & Spanish MPS Societies

In Norway ERT for MPS I has been approved but, similar to our situation in the UK, each region has a budget and applications are made to regional health authorities who must cover the cost of treatment within their allocated budget. The Norwegian MPS Society, recognising the inequalities in this system, is again like in the UK advocating for a national approach and specifically in Norway, for national guidelines and a national committee to evaluate each case.

In Sweden a decision has been made to pay for all MPS I patients. Patients in the USA are required to seek funding through their personal medical insurance or their state insurance. Whilst in Italy, for each patient their physician applies to the Ministry of Health for authorisation to import Aldurazyme for their patient.

Canada has a system similar to the UK. Aldurazyme was approved in Canada in May 2004. The actual implementation of healthcare though is done at a provincial level and it is up to each province to decide how to spend its health care budget and in particular which drug it will cover. Outcomes of provincial funding applications in Canada are currently pending.

There are other countries such as Australia from whom we learnt that Aldurazyme is not yet available so no patients are receiving treatment. At the end of the meeting it was agreed that keeping up to date on the status of ERT funding in each country would be invaluable and I have taken on the role of collecting information from each country in order to send out a regularly updated email information sheet for the international groups.

A New Children's Hospice for Germany

Christine Lavery

En route to my holiday in Switzerland this July, I had the privilege of making a personal visit to the newly opened Kinderhospiz Regenbogenland in Dusseldorf, Germany.

The Children's Hospice Movement in Germany was founded in February 1990 by a small group of MPS families that included our dear friends of twenty years, Richard and Elizabeth Volk whose two eldest children, twins Hendrick and Emily died in their late teens from Sanfilippo disease in 2001.

After much research that included visits to the UK to gain the inspiration, guidance and know how from Helen House in Oxford and Martin House in Yorkshire, the first German Kinderhospiz Balthasar came into being in Olpe in 1998. Since then two more Kinderhospiz in Bavaria and Bremen have been built. The fourth and new Kinderhospiz in Düsseldorf is called Regenbogenland which translates to 'Rainbow Land'. It is not difficult to see why as the whole frontage of the hospice is like a pastel shaded rainbow.

In the UK we have become acclimatised to children's hospices being new, built with room for not only visionary interior design but also exterior. Many children's hospices are in rural areas so I was interested to see how a Kinderhospiz in a major city would work.

As you would expect all four floors of the house are fully accessible and there are some interesting design aspects including an atrium in the middle of the children's floor. This means that the children can see all the seasons and even experience touching snow.

Farbkonzept Vorderansicht

One of the striking attractions of this city center Kinderhospiz was its accessibility to public transport and a nightlife for parents resident with their children. Dusseldorf's old town is known as the longest bar in the world!

Not everything can be judged by a building and I am particularly struck by Kinderhospiz Regenbogenland's emphasis on the involvement of siblings and how siblings are feeling. These quotes from brothers and sisters are particularly poignant.

'Mum and Dad don't see me anymore. They are only ever with Thomas. I wish I was dying and then Mum and Dad would be with me.' Stephen aged 9

'Mum and Dad cry a lot. They are sad because my little brother must die. I don't cry so that Mum and Dad won't get sadder. I comfort them.' Melanie aged 8

Achieving Enzyme Replacement Therapy for MPS I & Fabry Disease

Ellie Gunary

Members of the Advocacy Support Team remain committed to providing the individual advocacy support service to all MPS Society members. Achieving Enzyme Replacement Therapy for individuals with MPS I and Fabry Disease continues to be my high priority and one in which I continue to take a lead role with Christine, working with clinicians at the tertiary centres for MPS I and Fabry Disease as well as with Alison Millar at Leigh, Day and Co. solicitors.

As a result of the MPS Society's support to date, eight individuals with MPS I and eight individuals with Fabry Disease are now receiving funded Enzyme Replacement Therapy who were previously either not receiving a response from their Primary Care Trust (PCT) or had been refused Primary Care Trust funding. A further nine individuals are currently being actively supported.

The response to the report, commissioned by the West Midlands Specialist Services Agency, into ERT has now been sent. This report, before it was produced, was being cited as an excuse by many Primary Care Trusts for not yet making a decision in relation to funding ERT. Yet, when it was published, it was seriously flawed. The response has been acknowledged but as yet nothing further has been heard.

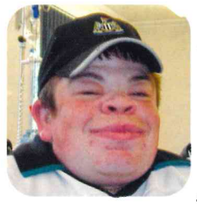
Following the meetings both with MPs in December 2003 which led to a meeting with John Hutton MP, Minister of Health in January 2004, MPS now understands that the Department of Health has agreed national designation and funding under the auspices of the National Specialist Commissioning Advisory Group (NSCAG) to provide a diagnosis and management advice service for Lysosomal Storage Diseases to include Fabry Disease and MPS I. All the details are not yet known and final confirmation is awaited. The MPS Society has been instrumental in achieving this very positive outcome.

NSCAG was established in 1996 to advise ministers of the identification and funding of services where central intervention into local commissioning of patient services was necessary for reasons of clinical effectiveness, equity of access and economic viability.

NSCAG aims to help patients by improving access to uncommon services whilst at the same time seeking to sustain high levels of expertise by designating specialist centres. It aims to help local commissioners by smoothing out risk and removing from them responsibility for the unplannable, and it aims to help providers (PCTs) by assuring a cash flow to support rare and expensive treatments. This arrangement was set up specifically for rare diseases where setting up regional services is not clinically or economically justified.

Whilst PCT's will still be required to fund the drug costs the service costs will be met centrally. This is a huge step in the right direction for lysosomal storage diseases and a very positive development in respect of the national response sought for funding for Enzyme Replacement Therapy for lysosomal storage diseases in England. However, there is much work ahead to persuade the Minister of Health to centrally fund the ERT drugs. Our thanks go to the doctors and MPs who supported MPS in this endeavour so far.

Colin Arrowsmith keeps us updated...



My name is Colin Arrowsmith. I am 24 years old, live in Newcastle and I have Hunter Disease. I started on the ERT trial in January this year.

On 4 January I had to visit Manchester Children's Hospital for the base line test week. There was a criteria on the lung capacity test for which you had to be under 80%. Even though I only got 27% in earlier tests I was nervous because I wanted to be on the trial so much.

My first infusion was on 14 January. We were really excited and relieved the day was finally here. However, the day flew by as we played football on the Play Station 2!

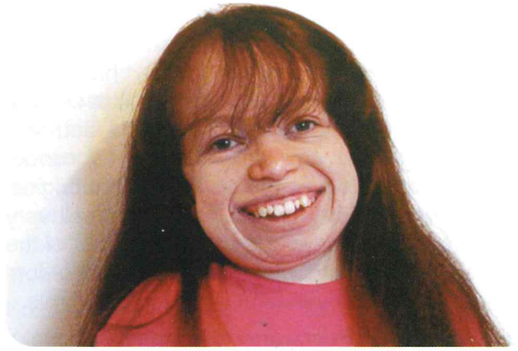
As the weeks went by I noticed my skin and hair felt softer. On week 8 I had a mild reaction with an itchy face and a rash. This carried on for about six weeks, getting milder and milder as the weeks went on. On week 17 we had another week of tests to see if there was any progress. My walking had improved and on the video test my balance and pulling a jumper on and off were better than in January. I also came off my strong painkillers which I had been on for 6 years. Now I just take one painkiller for bed.

Since week 17 I have started walking around the house and I can walk up the stairs. I hardly ever use my wheelchair in the house and I can now walk out to my car.

I can't believe how quick everything seems to be happening! I have to go for another week of tests on week 35 so I am hoping things have improved further. I will keep you updated.

Enzyme Replacement Therapy Jennifer's Story

Jenny Johnston



I was born in 1983 and diagnosed at three years of age with MPS I, Hurler Scheie Disease which is a severe, degenerating condition.

It was not until passing my Standard grades and going on to study three highers that my physical condition started to deteriorate rapidly. I was unable to continue with high school. With great difficulty, I only accomplished studying and passing Higher Biology, with the help of the outreach teaching service, at home. I was experiencing extreme suffering all the time.

My body, especially my back and joints started to affect me daily. My life took a dive for the worst. It was getting harder and harder for me to cope with the everyday tolls of life.

Increasingly painful joints, with their limited and restricted range of movement, made frequent tasks difficult and mostly impossible. These included washing, dressing and feeding myself. I was very uncomfortable sitting, standing, walking and even lying in my bed was painful, but it was all I could do – lie in pain.

I had a huge liver and spleen from the continuing build up of mucopolysaccharides. Breathing was heavy and shallow, my lung capacity was small and at night I snored really loudly. I suffered bad colds and infections, which made me very deaf. My growth velocity was consistently inadequate, resulting in me having a short stature.

Previously, I went to many hospital appointments but all they could do was monitor my worsening condition. I had given up on hospitals, as there was no treatment available for me at this time. It was just a matter of waiting until I surrendered to the misery of the disease. My life and health crumbled before my eyes. I hated and despised the state of my weakening body. What future was there for me? A circumstance I didn't want to think about.

It was 2001 when I was saved! Reinstated with the missing alpha-l-iduronidase enzyme, my body was given a breath of life. Improvements were noticeable very soon after I began enzyme replacement therapy (ERT).

The benefits of ERT were substantial, comparing myself now to how I was, the things I couldn't do then to the things I can do now (and enjoy doing) as I look back on how I once suffered. An everyday troublesome situation was, for example, eating my dinner at the table with my family. I could not sit because of the shooting pains through my joints and down my legs. Sitting anywhere, watching TV or at the cinema, or travelling in a car was something I never contemplated.

Today, the accumulated mucopolysaccharides, which were present in my liver and spleen, have now been eradicated and these organs have returned to a normal size. I can now travel about more comfortably in the car and can enjoy shopping. I attend college and am studying towards an HNC in Law and am also doing a course in IT. Coughs, colds and infections have been greatly reduced along with the associated deafness.

My lung capacity is still increasing, my loud snoring has totally stopped and joint movement, flexibility and energy levels have increased, allowing me to do more and by giving me greater independence. Laronidase treatment, for me, is a lifesaver. It reversed and halted the degenerating effects of my condition.

As of now, I feel I am still improving and I feel fantastic. With this treatment I have a future – without it, I don't.

I am thankful I was young enough to receive treatment, as with the severity of my condition, without it I would probably not have lived to write this. I am pleased for young MPS I sufferers who can receive ERT (Laronidase) now and stop them deteriorating to the stage I was at. Laronidase enzyme replacement therapy is an extraordinary treatment with eminent advantages for MPS I sufferers. Laronidase is now a licensed drug in the United Kingdom.

I would like to take this opportunity to thank Genzyme and BioMarin for producing Laronidase for the treatment of MPS I. I am truly grateful to everyone who has supported me through my progress on Laronidase treatment. This includes the MPS Society, Willink Biochemical Genetics Unit teams and professionals at Glasgow Gartnavel Hospital, where I continue my weekly treatment, while still on trial.

Achieving Funding for ERT for MPS I & Fabry Disease

Alison Millar

Recently the MPS Society has referred to me a number of patients and their families who have experienced problems with their local Primary Care Trusts over funding for ERT for MPS I or Fabry Disease.

Commonly, there has been a lack of any local policy adopted in relation to the treatment of lysosomal storage disorders.

In the absence of such a policy, the Primary Care Trust concerned was either refusing the patient's application without looking at the individual merits or simply putting off making any decision at all. Various reasons were given for this – but were variations on the theme that the patient's application could not be looked at until national or regional guidance on the funding of enzyme replacement therapies had been developed.

I was asked to consider whether there were grounds for 'judicial review' – the legal procedure by which the Court reviews the lawfulness of a decision or action of a public body. I thought it might be helpful to discuss some of the strengths – and limitations – of this procedure. These issues are not only of relevance to patients and their families and carers currently struggling to get treatment for Fabry Disease or MPS I. It seems to me that they could well reoccur as enzyme replacement treatments for other MPS and related diseases become commercially available if healthcare decision-makers do not engage with the issues raised now.

The Court's role in claims for judicial review is to review the way in which a public authority has come by its decision. The Court will scrutinise whether the public authority has correctly understood and applied the law that regulates its decision-making powers or responsibilities. The Court can also look at the procedure the public authority has followed when making its decision and consider overall whether this was 'fair'. This is part of the Court's constitutional role to supervise the exercise of power by the State, and check that public authorities do not abuse their powers or carry out their duties in an unfair or arbitrary way.

Since 2 October 2000, all public authorities, including the Courts, have also been required not to act in a way that would violate an individual's rights under the European Convention on Human Rights.

It is important to realise, however, that the Court will not get into making what it thinks is the

A Legal View

'correct' decision for the public authority. If the Court finds that a public authority has made an unlawful decision, its main power is to set that decision aside and order the public authority to take it again, this time following a correct approach and fair procedure. Even if the original decision was clearly unlawful the Court will very rarely be persuaded to step into the shoes of the public authority and make the actual decision. That would be to overstep its constitutional role.

Primary Care Trusts are delegated to make reasonable provision of healthcare services for people within their area. That does not mean they have to fund all medical treatment and services that their patients could benefit from.

NHS authorities have limited budgets and cannot provide all necessary treatment. But they have to operate fair policies and procedures for deciding which treatment should be funded based on clinical need and taking into account the circumstances of a patient's individual case.

In the enzyme replacement therapy cases, we have had a considerable degree of success getting Primary Care Trusts to acknowledge that they have to consider individual patient's applications for treatment and it is not legitimate to put them off until some national or regional guidance is made. By and large most Primary Care Trusts, when challenged, have put into effect their procedures for considering cases where there is no existing policy or guidance on funding of the treatment.

It is at this stage, however, that the limitations of the legal process can become most apparent. The Court can look at the approach the Primary Care Trust should follow when making a decision on treatment – and in one case in which I have been involved we have had an important concession from the Primary Care Trust, after legal proceedings were issued, that the decision challenged had been made unlawfully.

However, the Court will not interfere with a Primary Care Trust's judgements on the likelihood of the effectiveness of medical treatment, or how its cost effectiveness may compare with other treatments. These are judgements for the health authority alone to take, and the Court will not arbitrate on this even if you feel the health authority got it wrong. This can cause real problems in cases like these where the medical condition is rare and the treatment is new and not well understood outside the national centres.

The MPS Society have worked very hard to assist patients and their families to challenge inaccuracies through the internal appeals processes operated by Primary Care Trusts. However, there are significant limitations with this procedure. Often the people involved in the appeal will have no more knowledge of the disease or its treatment than the original decision-makers. They may fall into the same errors or make further erroneous assumptions about the application. The ultimate decision may therefore still be very unsatisfactory, but, for the reasons I have explained above, it is likely to be very difficult to challenge.

So what can be done in these circumstances? It may be possible to reapply if further evidence of the clinical effectiveness and cost effectiveness of the treatment becomes available at a later stage.

However, the wider point arising out of all this is that it is not practical for policy about the funding of enzyme replacement therapies to be determined at a local Primary Care Trust level. The local decision-makers may well not have any previous knowledge of lysosomal storage diseases and their treatment and be having to acquire this expertise for the first time, and this causes problems. The current very high cost of the treatment cannot be ignored, and means that local decision-makers - not knowing much about MPS and related diseases and their devastating effects and struggling to engage with these issues - are inevitably going to be reluctant to approve funding out of their limited resources.

The current situation where decisions have to be taken at a local level also leads to a bit of a lottery in funding, which is clearly unacceptable. It therefore seems to me that national guidance needs to be agreed on the funding of enzyme replacement therapies. This becomes all the more important as further of these treatments are developed, putting extra demands on the budgets of local health authorities.

This is where the campaigning work of the MPS Society – and of individual patients, their families and carers – becomes absolutely crucial. As a solicitor at a 'campaigning' law firm, I am well aware how the law can only do so much, and often it is 'people power' that gets issues put up the agenda of Government and given due consideration. It is really important you continue to raise public awareness of MPS I and related diseases, and push for the funding of these treatments both so that national guidance is promptly developed and so that guidance properly reflects the high need for funding. ■

Alison Millar is a solicitor in the Human Rights Department of Leigh, Day & Co. (amillar@leighday.co.uk)

ERT News in brief

Ellie Gunary

In the last newsletter Christine wrote about the decisions made in both Scotland and Wales to not fund ERT.

Scotland

The Deputy Chair of the Scottish Medicines Consortium (SMC) in saying no to funding ERT for MPS I issued on 7 May 2004 the following advice... 'Laronidase is not recommended for use within NHS Scotland for the treatment of MPS Type I. Laronidase was approved by the EMEA under exceptional circumstances and has been granted orphan drug status. The evidence of its efficacy is therefore limited. No information is presented in the submission to support the therapy being cost effective and therefore the economic case is not demonstrated.' Whilst Genzyme are appealing to the SMC against this decision the MPS Society is supporting two individuals in Scotland who have been refused funding by their local NHS as a consequence of this decision. This is all the more difficult because one health authority in Scotland is funding ERT for a child with MPS I.

Wales

In Wales, Genzyme's appeal to the All Wales Medicines Strategy Group (AWMSG) was rejected on all counts. As a consequence the MPS Society has written comprehensively to Dr Paul Buss, Chair of the Appeal Panel, to Professor Walker in his capacity as Chairman of AWMSG and to Professor Routledge, Welsh Medicines Panel Director. Unfortunately Dr Paul Buss' response of 16 June 2004 indicates that the AWMSG have only noted the MPS Society's views, although the AWMSG have acknowledged that in the future, the involvement of stakeholders in its deliberations will be high on the agenda, this being as a direct result of the MPS Society's input. Rather like Scotland, postcode prescribing prevails and one child is receiving ERT funded by her Health Authority.

For the Society's members in general, however, the decisions of both the AWMSG and the SMC spell not only enormous disappointment at a personal level as their disease progresses but also we would argue clearly indicate discrimination by Health Authorities in terms of treatment options.The MPS Society in its support to members in Scotland and Wales is committed to supporting those individuals who approach us achieve ERT when this is clinically recommended by their tertiary centre doctor. Options being currently pursued include judicial review working with lawyers and highlighting concerns through Members of Parliament.

ERT

A Service for Adults with

Inherited Metabolic Diseases

Dr Phil Lee

Introduction

Much has happened over the last one hundred years since Archibald Garrod first recognised the existence of inborn errors of metabolism (IEMs). Enzymes have been discovered and the genes for these enzymes have been characterised. Understanding of the underlying problems behind many clinical syndromes has improved.

All of these have led to the development of a variety of treatments that have changed the natural history of IEMs. Many of these treatments have been special diets but more recently enzyme replacement therapy for some lysosomal storage diseases has become available. Not only can much more be done today for people with IEMs than ever before, but more are being diagnosed as laboratory technology improves and doctors become aware of these disorders.

Paediatric metabolic medicine has achieved what it has set out to do: prevent untreatable disease with genetic counseling and antenatal testing and provide effective treatment to children with IEMs.

The challenge that is now faced is dealing with these individuals as they pass through adolescence into adulthood and start to have families of their own. This article discusses the approach of one centre dedicated to the care of adolescence and adults with inherited metabolic disease.

Transition & Dependence

The transition of an individual from childhood to adulthood is a difficult process whether or not a chronic disorder exists. When the disorder has an impact on everyday life, that process is even harder. People with IEMs often have multiple physical, learning, psychological and social problems. In addition, many of the treatments for these disorders are difficult and complicated to administer.

More and more is expected of the young person: taking over the responsibility for the treatment and its monitoring. It is not surprising that adolescence is a difficult time and, on top of that, the individual is then asked to leave the children's services that they have grown to trust and rely upon. This is a time that many patients fall out of the system and become 'lost to follow-up'.

As well as the physical and emotional changes that occur as individuals grow older, there are great social changes that happen. The supportive school environment is left behind and replaced by a competitive working life and young people leave the family to establish themselves as individuals within society.

Parents have invested a huge amount, emotionally, physically and financially in their child with an IEM. Letting go is particularly hard to do as they see all their hard work potentially wasted. Indeed many of my patients still attend clinic with their parents even in their 30s and beyond! Much of our work is spent trying to separate the patients from their families so that care can be directed at them.

The Adult Metabolic Clinic at UCL

The unit within which I work was first established by Professor Charles Dent in the 1940s. He was a clinician scientist who introduced the amino acid analyser into the clinical setting, which allowed him to recognise many conditions and he even had a disorder named after him: Dent's disease describes a rare kidney disorder. He died shortly after he retired and for the last quarter of the 20th century, the unit was successfully run by Dr David Brenton at the Middlesex Hospital until 1998.

Currently the unit is thriving with over 1,000 patients attending, a number which is growing rapidly year upon year, as new referrals increase and few pass away. Close links with the paediatric metabolic service at Great Ormond Street Hospital for Children exist both at a clinical and laboratory level. Many of the biochemical and molecular services are shared too.

I joined the unit in 1997 and moved it to its current site at the National Hospital for Neurology and Neurosurgery, Queen Square (part of the same UCL Hospitals NHS Trust) in 2000. The purpose of the move was to improve the transition of the patients from Great Ormond Street Hospital, just around the corner.

It means communication between the adult and children's teams has improved and has also meant fewer blood and urine samples get lost going to the laboratory! Queen Square is also very suited to looking after people with rare problems, so we have not become lost in a large general hospital. Many of our patients have neurological, neurosurgical and/or neuropsychological problems so a neurological setting is an appropriate one for them.

I run a weekly clinic at Great Ormond Street Hospital and two of my paediatric colleagues, Dr Ashok Vellodi and Dr Maureen Cleary run clinics at Queen Square once or twice a month. The age of transfer depends entirely on the individual and their family, according to their physical and emotional development, varying between 13 years and 18 years.

As well as medical consultants there are also other very important members of the team including a clinical nurse specialist and a research nurse who are very much involved in enzyme therapies for Fabry disease and Gaucher disease; two dietitians and a dietetic assistant who look after the needs of those on special metabolic diets; a clinical research fellow who is examining nutritional interventions in glycogen storage diseases; and two research dietitians running local and national projects. We also have a full-time secretary who makes sure everything runs smoothly and Chemical Pathology/Metabolic Medicine trainee doctors pass through the unit for blocks of four months.

Facilities

Unfortunately, many adults with IEMs are still seen within paediatric services because there is no appropriate adult service to which to refer them. Although this may be satisfactory in an out-patient setting where clinics for older

The Metabolic Team

patients can be arranged separately from the children, difficulties may arise when in-patient beds are needed. This occurs when acute emergencies occur or there is a need to re-admit patients for dietary re-education. Day-case facilities for load tests and infusions (such as enzyme replacement therapy for lysosomal storage disorders) are also needed.

We are lucky that we are able to access appropriate ward facilities for our adult patients. We also find having a dedicated metabolic diet kitchen a particularly valuable resource. It can be used to provide metabolic diets for those requiring in-patient care which is extremely hard to achieve from hospital diet bays. It also allows patients to be educated practically about their diets.

Another facility that can be very helpful is a dedicated 'Adolescent Clinic'. We are fortunate to have access to a service within the Hospital Trust which is run by the first appointed director of adolescent medicine in the United Kingdom and is run by specifically trained nurses with appropriate clinical psychological support.

Conclusion

The specialty of Inherited Metabolic Disease is growing up rapidly into adulthood. Increasing numbers of young people with IEMs are surviving and it is essential that their needs are met in a patient focused service.

This article has described one such unit in London which could act as a template for other units to model. Much work is needed to develop these facilities, particularly as more therapies become available for groups such as those with mucopolysaccharidoses. ■

Dr Philip Lee, Charles Dent Metabolic Unit
The National Hospital for Neurology & Neurosurgery
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Special Interest

Are we getting it right?

A Quality Assurance Package designed for Children's Hospice Services

ASSOCIATION OF CHILDREN'S HOSPICES

People who provide services for children and their families need to find out if what they offer is right for those who receive them. Children's hospice services attempt to give personalised, individual care to children with life-limiting conditions and for this their staff often receive gratitude and praise. Staff gain pleasure from families' relief and thanks for being helped through periods of great pain and stress, as well as for much needed respite.

But, these expressions of gratitude, welcome though they are, do not give hospice services a clear indication of where the strengths and weaknesses of their organisations lie, or enable them to plan improvements in a systematic way.

It is because children's hospice services recognise the importance of having a systematic means of finding out if they are getting it right for children and families that their umbrella organisation, the Association of Children's Hospices (ACH), has designed and developed a quality assurance (QA) package specifically for them.

The package is called **'Are we getting it right?'** and ACH is delighted that it is now published and children's hospices are starting to use it.

If your family uses, or is thinking about using, a children's hospice service you can look at a copy of the package which will give you an insight into the values underlying what the service offers.

ACH started with a clean sheet of paper for their quality assurance project which I have co-ordinated since 2001. The Steering Group and I decided that a tool of this kind should embody the values of everyone who had a part in children's hospices. We therefore embarked on a consultation of all these groups, that is the staff, the children and their parents, and those who work closely with children's hospices such as special needs teachers, consultants, paediatric community nurses, art and music therapists.

Twenty six hospices took part in the consultation. Each appointed members of its care team to work on the QA project and to form a think-tank of ideas. The first thing they did was to find out what staff thought were the essential aspects of the service they offered and the replies showed what a breadth of support their hospices offered children and families from the time of a child's referral.

It was agreed by all the hospices that we should find out what children and young people value most about hospice support since they are the focus of children's hospice care.

We recognised that there is a large number of them who cannot express their views verbally and felt strongly that ways should be found to include them in the consultation. Sometimes drawing and painting are a means through which this group can express themselves, as for most children. We therefore invited all the children to show what they enjoy most about hospice visits in some form of artwork and we hoped that some of these pictures could be used to illustrate the package.

The Quality Assurance Link People (QALPS) in the hospices came up trumps gathering replies from eighty children, asking them what they enjoyed most about their visits and what, if anything, they would change. Amongst them were fourteen children who had little or no verbal ability.

The QALPS found it challenging to communicate with this group but enlisted the help of family members, special needs teachers or carers who supported the children in their homes – all of whom knew the children so well that they could interpret their reactions to many aspects of the hospice environment from spa pools and sensory rooms to family link people and night staff. Another important group were healthy siblings and thirteen brothers and sisters were consulted and gave some very moving replies to the questions.

The QALPS also asked some parents in each hospice what difference the hospice service made to their lives and the three things they valued most about it, as well as anything they would change.

In addition, six professionals associated with each hospice completed questionnaires; we were delighted to receive so much rich data and a wonderful collection of very colourful, expressive children's artwork.

When the responses from children, parents, staff and associated professionals were combined a large degree of consensus was apparent. Among the most valued things was care tailored to the individual needs of each child by staff who are welcoming and who understand children, as well as being professionally skilled. Children see carers as 'friends' who accept them as normal despite their condition and enable them to do things which may not be possible at home with their family, such as trips out with peers and a wide variety of leisure activities.

The emphasis for the group of children who cannot communicate verbally is on knowing their carers well and being known by them, in familiar, child-friendly, safe surroundings. Alongside this is the wish to experience new and different things which are introduced at their pace.

Parents appreciate staff who they can talk to at any time and who are there for them when emergencies arise.

All the groups value care which meets the needs of both the child and family members as fully as possible, including those of healthy siblings.

They see a collaborative way of working between hospice teams and other professionals involved in a child's care as offering the best support for children and families, avoiding as far as possible the necessity to repeat the child's story and making hand over from one source of care to another as seamless as possible. As to changes they would like to see, one stood out above all – the wish by children, parents and professionals for more and longer visits!

After much careful reading of the responses it was decided that the content of the QA Package should fall into six main areas, or *key aspects*, namely: access, the child, the family, the staff, environment and communication.

For each *key aspect* hospice staff are invited to answer several focus questions about care for the child, care for the family and so on. They do this by gathering evidence from within the hospice relating to a number of value statements and then making a judgement about how far they meet them. For example, the first focus question for *key aspect* 'The Child' asks how successful staff are at recognising and respecting each child as an individual.

Evidence can be found in all sorts of records and paperwork which children's hospices keep; there are child notes and care plans, and many hospices write individual diaries of each visit which let families know details about their child's day.

There are four **value statements** for which evidence needs to be found, as follows:

There are **sufficient staff** to give each child individual attention.

The child's need for **continuity of care** by known staff is recognised and observed whenever possible.

Each child receives **personalised care**.

Staff are successful in promoting children's best interests and **respect** their potential autonomy.

However, the people who can provide the best information, or evidence, are often children and families. Hospice staff are therefore keen to obtain their views about all these matters and plan to ask them whether their child has sufficient individual attention and whether he or she knows some staff well. Staff will plan these conversations having regard to confidentiality, and then reflect on what they hear. Once they have collected evidence from different people and from their records, they should be able to discern where and how improvements can be made to their service.

Children's hospice teams have a tool to help them develop their service and an approach to quality assurance which offers children & parents opportunities to contribute to continuing improvement.

In addition they are acquiring evidence which demonstrates what they do well and which they can use to enhance the standards and professionalism of children's hospice care.

For more information contact the author of this article Eve Herd, Association of Children's Hospices QA Co-ordinator, at

qaproject@childhospice.org.uk

Special Interest

React

Rapid Effective Assistance
for Children with potentially Terminal Illness

The need for React

At least one in 1,000 British children suffers from a life-limiting (terminal) illness. Nearly all of these children are cared for at home by their families day and night throughout their short lives. Most life-limiting illnesses are degenerative – a child's health will deteriorate over a period of years and their needs will change.

For many of the families we help it is the pressure of caring for a very sick child that has brought financial hardship; the breakdown of marriages forcing single parents to cope alone, or the need to care for a sick child 24 hours a day meaning that parents can no longer work.

About React

Founded in 1989, React is a dynamic small charity working to improve the quality of life for financially disadvantaged children with life-limiting illnesses across the UK. We are all passionate about our work and believe that every child should have comfort, dignity and the opportunity to participate in life as fully as possible.

Our work

Based on professional advice and our 14 years' experience, we provide children and their families with specialist buggies, car seats and wheelchairs, adapted computer equipment, sensory equipment and toys, electric beds, washing machines and tumble dryers and holidays in one of our five mobile homes

All of the above, along with hoists, stair lifts, mattresses, and many other items including, very sadly, help with funeral expenses, are provided in a quick and caring way. We help children only where support is not available through the National Health Service or Local Authority, and all applications must be endorsed by a medical or social work professional.

How can you contact React?

W: www.reactcharity.org
T: 020 8940 2575
E: react@reactcharity.org

ACT Information Pack

ACT has produced an information pack for families which outlines the main sources of support available to children with long term or life limiting conditions. Individual packs can be obtained free of charge from ACT at www.act.org.uk or phone 0845 108 2201.

Carers (Equal Opportunities) Bill

ACT was delighted at the news that the Carers (Equal Opportunities) Bill has become an Act, as it received Royal Assent on 22 July 2004. The Private Member's Bill was introduced by Dr Hywel Francis, MP for Aberavon and has been dubbed 'Sam's Act' in memory of his disabled son, who he and his wife, Mair, cared for until he died at the age of 16. The MPS Society was one of 300 organisations that gave support to this Bill.

Under the new law, carers will now have to be told about their rights, have more opportunities for work, education and leisure and there will be greater collaboration between statutory services to help them in their caring roles.

Early Intervention Feasibility Study

Ellie Gunary

Representing the MPS Society I was invited on 20 May 2004 to a planning day for a new centre for early intervention for families with very young disabled children. Mencap is leading a feasibility study commissioned by the Department for Education and Skills to set up a national centre for early intervention. Early intervention has been shown to have the potential to make a real difference to the lives of young children with disabilities and their families. The vision is for a national centre that will provide a focus for information, practice, research and training for children with disabilities aged from birth to 5.

This day brought together parents, practitioners, researchers and representatives of voluntary organisations. Presentations were made and workshops held. During the workshops we discussed and presented thoughts and ideas on what a national centre should do and ideas on how it could be run. The findings of the planning day of the most popular functions of the national centre are: Influencing education and training in the field of early intervention; Providing and distributing information; Carrying out or commissioning research; Developing standards or benchmarks for early intervention.

E-Forum launched to inform the Centre for Early Intervention Feasibility Study

As part of the feasibility study, a discussion forum has been launched. To join in the discussion MPS Society members need to:

Email password@earlyintervention.org.uk or call 020 7696 6954 for a password. Then go to www.earlyintervention.org.uk and click on 'e-forum'. The views posted on this form will be analysed and used to inform the final report on how the national centre will take shape.

The topics currently being discussed focus on four main areas: How a national centre could influence education and learning; What information could a centre provide and how could it be distributed; How a centre could carry out or commission research; How standards and benchmarks could be developed.

The outcome of the consultation is being reported back to the Department of Education and Skills.

www.earlyintervention.org.uk

Technical Equipment for Disabled People

Remap

www.remap.org.uk

Paul Bird, father of Lewis and Jack Bird (MPS IV), wrote to us about Remap saying 'Nothing seems to phase them'. Can Remap help you?

Remap is a registered charity producing technical equipment for children and adults with a disability. Using the voluntary services of around 1500 professional engineers, technicians and healthcare experts, Remap produces over 3000 bespoke aids each year to enhance independence in day to day living, education, work and leisure for people with disabilities. Remap volunteers design and manufacture, or adapt, equipment for people with disabilities providing it is not available commercially. The equipment is supplied free of charge to the client. Remap receives requests for help from occupational therapists, other healthcare professionals, charities acting for people with disabilities, the individuals and their families or carers.

For more information contact Remap
0845 1300 456 www.remap.org.uk

Ceiling Track Hoist

Free to anyone who can collect it from Kent. Gordon and Alison Rowe have an 18ft ceiling track hoist with bolts. Recently serviced, they are offering free to an MPS Society member.

Clearway Automatic Wheelchair Lift For Sale

Alison and Gordon Rowe also have a Clearway Automatic wheelchair lift for a VW Caravell Van for sale. Recently serviced. Offers from £200 to £300.

Any members interested, please contact the MPS Advocacy Support Team.

Mashed Food Recipes

The Advocacy Support Team have recently received a request for innovative and tasty recipes for mashed up food. Can you help?

Contact the Society now on
01494 434156

New Publications

The MPS Society has recently published several new booklets in the 'Guide to Understanding...' range. The ML II, ML III and Maroteaux-Lamy booklets are now available. The MPS VI booklet has been produced with the kind support of a grant from BioMarin Pharmaceutical Inc.

To order your copies contact us now on 01494 434156 or check out www.mpssociety.co.uk.

A new **progression** booklet for **MPS VI**, Maroteaux Lamy Disease, is in the pipeline. Do you have a series of good quality photos covering the life of an individual affected by MPS VI? If you do, and you would like more information, please contact us now.

Travel Insurance Information Sheet

Free to all members

50p to non members and professionals

In response to requests by our members, the MPS Society has compiled a list of Insurance Companies who have been recommended to the MPS Society by other members. The Society cannot recommend any of these companies specifically and has no direct experience of them. However all the companies listed are a result of positive experiences.

Disability Living Allowance Information Sheet

Free to all members

50p to non members and professionals

As part of the Advocacy Support Servicewe complete Disability Living Allowance (DLA) forms on behalf of individuals and families. The information sheet covers the general principles and requirements for making a successful claim. This information sheet provides details on the DLA and how it is assessed and explains the different components that need satisfying in order to make a successful claim.

It is not a step-by-step guide on how to fill in the DLA form from an individual perspectiveas each individual has varying needs. It provides an understanding of the DLA and the process.

Psychosocial Outcomes of BMT

Cheryl Pitt

All families with children or young adults affected by MPS I Hurler disease who have had a bone marrow transplant will now have received a formal invitation to volunteer to participate in the research project: Psychosocial Outcomes of Bone Marrow Transplant for Individuals Affected by MPS I Hurler Disease.

This research is very important as no data currently exists, which charts the psychological and social progress of affected children post-BMT. We therefore do not know the kind of challenges that children and young people may face throughout their lives, what factors influence their psychosocial development, and what resources they have to cope. Your participation in this research is important to us, and we would greatly appreciate any time you can give.

We would like to help improve the outcomes of bone marrow transplant for individuals affected by MPS I, and in order to do that we need your help. If you would like to take part in the research please complete the reply slip that was recently sent to you and return it to me in the envelope provided. Your participation will be kept confidential. If you did not receive information about this research and you think you should have, please call me at the MPS Office and I will send you details.

If you would like any other information about the project, or you had difficulty understanding the information you were sent, please also do not hesitate to give me a call. I look forward to hearing from you and to seeing you all soon.

Equipment available

Kirton chair

In very good condition, reclinable, with footrest. Available free to MPS members.

If you are interested call the MPS Society. 01494 434156

Jeans for Genes

We would like to hear what you did to raise money on Jeans for Genes day, Friday 1 October 2004.

Our thanks go to our many members and supporters who, on Friday 1 October 2004, joined people throughout the UK by paying £1 to wear jeans for the day. By doing so you, like so many people up and down the country, helped raise vital monies to support research into causes and treatments of genetic diseases and to fund much needed support projects such as an MPS Society Advocacy Support Worker.

The strapline for this year is 'Denimstrate you care' and we would like to hear how you denimstrated! Perhaps you...

Spread the word – Jeans for Genes produced colourful flyers, posters and other advertising materials

Took a Jeans for Genes fundraising pack into your place of work

Sent a Jeans for Genes nursery, primary and/or secondary school fundraising pack into local schools and nurseries

Wrote letters to local businesses and schools to get them on board

Organised fundraising events and activities

Sold, bought and wore Jeans for Genes badges and other Jeans for Genes merchandise

Spoke to the media providing a personal story of how MPS affects you and your family

Became a school speaker and made a presentation at a top supporting school organised through the MPS Society's Advocacy Support Team

Got in touch with your local Matalan store and organized or took part in fundraising activities with this Jeans for Genes supporter

Or perhaps being very original you did something very unique and different

Whatever you did to raise money we really thank you.

And why are we asking you to let us know what you did?

The Jeans for Genes management team are asked by local media throughout each campaign for details of events or fundraising activities in their area. We appreciate and respect that not everybody welcomes media coverage and if this is the case we would still like to hear from you to include your contribution in the MPS Society's Jeans for Genes report

For more information about Jeans for Genes visit www.jeansforgenes.com or freephone 0800 980 4800

Alvema Trax

In very good condition. Available to MPS members with harness & protector pads.

If you are interested call the MPS Society now for more information 01494 434156

MPS 2004 Research Grants

The MPS 2004 Research Grant Programme is now open. If you would like to apply for an MPS Research Grant please contact the MPS Society for an Application Form and Terms and Conditions.

All applications will be externally peer reviewed and must contain a convincing scientific proposal.

The deadline for receipt of applications is **20 November 2004**.

Society for
Mucopolysaccharide and Related Diseases

Providing **support**
to those affected
by rare, genetic,
life-limiting
MPS Diseases

CAN YOU LEND A HAND?

Contact us
now for a
fundraising pack

01494 434156
mps@mpssociety.co.uk
www.mpssociety.co.uk