

# MPS

Autumn 2006

Care Today, Hope Tomorrow

Society for  
Mucopolysaccharide  
Diseases



## 9th International Symposium

on MPS & Related Diseases  
Venice

**SPECIAL EDITION**

Mucopolysaccharide and 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 1200 children and adults suffering from MPS and Related 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.

## What are the aims of the MPS Society?

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

## How does the Society achieve 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 Individuals 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

### Quarterly Magazine

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

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MPS Magazine

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**Newsletter Deadlines**

Winter	1 Dec 2006
Spring	1 Mar 2007
Summer	1 Jun 2007
Autumn	1 Sep 2007

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# CHIEF EXECUTIVE'S REPORT



It is always a pleasure to be able to write of news on the treatment front. In July treatment for another group of our members came a big step closer when Elaprase was approved by the Food and Drug Administration (FDA) in the United States as an Enzyme Replacement Therapy (ERT) for MPS II, Hunter disease. This brings to five the number of MPS and related diseases (Hurler Scheie, Scheie, Hunter, Maroteaux Lamy and Fabry) for which there is an Enzyme Replacement Therapy. Whilst this is good news no day passes in the MPS Office when we aren't reminded that there is a long way to go with much needed breakthroughs for the other sixteen diseases.

As those of you wanting ERT who live in Scotland and Wales will know only too well, the system is not as straightforward or equitable as it is in England and Northern Ireland. Members of the Advocacy Team and myself recently spent four days in Scotland meeting eighteen children and adults affected by MPS diseases and their families. The purpose of our visit was to consult on what is working and what is not working for MPS patients in Scotland. Meetings with members took us to Ayrshire, Glasgow, Edinburgh, Fife and the Highlands and provided the Advocacy Team with valuable information which we are now compiling into a report to be shared with decision makers in Scotland. During our visit several families used the opportunity to share their experiences with the press and, as a result, a father whose son suffers from MPS II contacted us for support.

On 28 June MPS families, Trustees, staff and volunteers travelled to the four day International Symposium on Mucopolysaccharide Diseases in Venice. There were over 600 delegates split between a lay programme and scientific programme. In this magazine and future issues we will produce some of the talks and review the scientific presentations. There were exceptional contributions from UK MPS Society members. Some of these speakers have been asked to present at the MPS Conference in Northampton 29 June - 1 July 2007. Information about this Conference, which is open to MPS families and professionals, along with a booking form is enclosed with this magazine. 2007 is the start of the Society's 25th Anniversary year and we are planning to start this very special year with a conference weekend that has something for everyone. On Friday there will be a Fabry Meeting followed on Saturday by three separate conferences on MPS including one for those affected by the rarest of rare related MPS conditions. Please do book early as we expect demand to outweigh accommodation in the Hilton Hotel and will use the very nearby Marriott Hotel to provide additional accommodation.

The MPS Society has been awarded a BBC TV Lifeline Appeal that is expected to go out on Sunday 19 November 2006. We are currently working on the style of the message designed to persuade as many people as possible to support the work of the MPS Society by making a donation to the Society following the broadcast. We have some ambitious plans as to whom we would like to present the appeal and as soon as decisions are made they will be announced on our website ([www.mppsociety.co.uk](http://www.mppsociety.co.uk)). In order to continue to provide a high level of support services to over 1200 affected MPS families and develop new initiatives as well as fund vital research, we really do need as much help as possible. If you have family and friends who can help the Society by organising a fundraising event, taking part in sponsored events or selling Christmas cards please do let us know or put them in touch with the Society.

**Christine Lavery**  
Chief Executive

# News from the MANAGEMENT COMMITTEE

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Management Committee Meeting held on 15 July 2006.

## Governance

The Chairman advised Trustees that he had reviewed and checked the Charity Commission Annual Return completed by the Chief Executive and it was agreed the Chairman should sign on behalf of Trustees.

The Society's Risk Register was reviewed and it was agreed no changes are required at this time. However, Trustees did agree to seek independent advice regarding Trustee indemnity. The Chief Executive reviewed with Trustees the Governance Self Review Checklist and drew Trustees' attention to papers previously circulated relating to Learning through Board Performance and Payments and Benefits to Trustees.

## MPS Research Grants

Dr Brian Bigger, recently appointed Research Fellow and lead scientist of the MPS Stem Cell Group at the University of Manchester, gave an overview of his previous work at the University of Oxford and an update on the MPS Stem Cell research project.

The Chief Executive gave an update on each of the other research projects currently funded by the MPS Society. The Trustees expressed their thanks to Clare Beesley from the Institute of Child Health for her work on the Biomarker Project and all her efforts for MPS over the last few years.

Trustees considered an application for a capital research grant and part funding for a research project. It was agreed unanimously to fund two new PCR machines for the Willink Biochemical Genetics Unit at a cost of £6,616.

The Shauna Gosling Trust have agreed to fund the revenue costs and capital costs in Year 1 of a post doc scientist for the MPS Stem Cell Group in Manchester. The Trustees agreed unanimously to fund £60,000 in Year 2 and are committed to securing the funds required for Year 3.

## Personnel

The Chief Executive reported that Cheryl Pitt is on maternity leave from her post as Research Officer having had a baby boy, Jackson Robert Pitt, and that flowers had been sent on behalf of Trustees.

## Jeans for Genes 2006 Appeal

The Chief Executive brought Trustees up to date on issues including the good news that the MPS Society had been able to secure sponsorship of £85,000 for the 2006 Jeans for Genes Appeal from Genzyme and Shire. Wilma Robins reported on the plans for the Jeans for Genes Carol Service to be held on 20 December at St James', Piccadilly.

## MPS Events

The Chief Executive introduced plans for the celebration of the Society's 25th anniversary year. An outline of the Society's programme for the 2007 MPS National Conference was given.

### **You are important to us, please keep in touch.**

Please remember to let the Society know if you are moving and your new address and telephone number. In addition to helping keep the printing costs down, you will help us keep our database up to date. Keep us informed of new addresses, telephone numbers, email addresses and any interesting news about yourself, your child or your family.

### **New 0845 telephone number**

The MPS Society telephone number is 0845 389 9901. This number has been introduced as a benefit to MPS Society members as call costs will be charged to the caller at 'local' rates. Please remember to use this number when contacting us.

# AGM '06

## MPS Annual General Meeting 2006

The Annual General Meeting of the Society took place at the Splash Landings Hotel on Saturday 29 April 2006 at 6.30pm preceeding the Alton Towers Weekend Gala Dinner. 75 members, 7 guests and 6 employees attended. There were no apologies.

The minutes of the Annual General Meeting held on 2 July 2005 were distributed in advance to those members present and were accepted as true and accurate.

The Chairman, Barry Wilson, presented the Trustees Report. This is published in the Annual Report and Accounts for the year ending 31 October 2005.

The Treasurer, Judith Evans, presented the Statement of Accounts, for the financial year ending 31 October 2005, the details of which are also to be found in the Society's latest Annual Report. It was proposed and seconded that the auditors, McLintock and Partners, Chester, be appointed the Society's auditors for the financial year ending 31 October 2006.

There being five vacant places on the Management Committee, Wilma Robins and Bob Devine were re-elected and Bob Stevens, Paul Moody and Angela Brown were elected as Trustees.

Under any other business, the Chairman outlined the decisions that led to the purchase of MPS House and the Society's strategy to increase its income from Charitable Trusts to enable the Society to reach out to an ever-increasing number of individuals, families, partners, carers and professionals affected by MPS, Fabry and Related Lysosomal Storage Diseases. The Chairman then thanked the members and guests for coming to Alton Towers and making the weekend so enjoyable.



MPS Board of Trustees



Children's Entertainment

Just to say thank you for the Alton Towers weekend. We had a brilliant time even though the weather wasn't too good on Sunday. It was nice to meet up with friends we have made and talk to others about the MPS II ERT.

Ben had a good time at the children's entertainment and enjoyed meeting up with Tom the volunteer he has had previously.

Many thanks to you all  
Lee, Jackie and Ben Cooper (MPS II)



**LOTTERY FUNDED**

The MPS Society is very grateful to be awarded a grant for **£3,500** by the **Awards for All** programme to support the childcare activities which took place during the Alton Towers Family Weekend and Annual General Meeting in April 2006.

# Introducing our new Trustees

## Peter Conlin

My name is Peter Conlin and I am a co-opted trustee for this current year.

My association with the MPS Society started when my son Joshua Benjamin (Ben) was diagnosed with MPS I (HS) on Halloween 2003 by Dr Ed Wraith at Manchester. Ben is currently on ERT in Teesside.

In the early days I was not sure of what career suited me best so my early C.V. is quite varied including; Assistant Educational Consultant, Computer Operator, Accountancy and also Restaurant Management in Cambridge where I cooked 169 steaks one Friday night and on another occasion cooked for Jeffrey Archer. I learnt how to cook steak and more importantly how to look after a beer cellar.

In 1991, I became interested in Speech Therapy and in 1996 I qualified as a Speech and Language Therapist from the University of Newcastle upon Tyne. As a Therapist I was lucky enough to experience working at the Michael Palin Centre for Stammering Children in Islington, where I met the great man himself at the Christmas Party. I am now happily working as a Speech and Language Therapist (Expert Practitioner in Paediatric and Adolescent Stammering) in South Shields, in South Tyneside (somewhere UP NORTH for those that don't know).

My biggest change, apart from Ben's diagnosis, occurred at the start of this year when I decided to stop spending every evening staring at a box in the corner of my lounge and do something constructive. In which time I have given up

alcohol, written two children's books, one rejected by various agents, and about to be re-written and improved, started going to Ju-jitsu twice a week and read a lot more fantastic books.

I love Ju-jitsu as I get to spend time with Sarah (my daughter) and talk about how to hurt people in 100 different ways (although Ju-jitsu is really self-defence). We have both recently passed our orange belts. Ju-jitsu is a great way to reduce stress as if you don't concentrate you get hurt, you quickly learn to forget about work.

My other great passions include reading, especially Colin Bateman, Nick Hornby, Terry Pratchett, Ian Banks, books about stammering but most of all Spike Milligan (and obviously MPS information that Antonia keeps sending me).

I love music, although recently guitar lessons have stopped due to lack of time, I felt that at some point in the day I needed to sleep.

From my working experience and my personality I have a number of skills to bring to the Board of Trustees. I look forward to working with the other Trustees.



## Bob Stevens

I would like to introduce myself as one of the new trustees of the MPS Society.

My name is Bob Stevens and I am 38 years old. I have worked in the Plant Hire industry for 22 years, starting as a junior and working my way up to Director. I have also recently started a business with my father-in-law and best friend developing properties on the Isle of Wight; this is ultimately what I would like to do full time.

My hobbies include golf (although games are few and far between at present!) and going to the gym. Earlier in the year I swapped dumbbells for 'pavement pounding' and completed the London Marathon for the MPS Society, an achievement of which I am extremely proud.

I live in Farnham, Surrey with my wife, Claire, two children, Oliver and Samuel and our cat called Matey.

photo courtesy of David Betteridge  
www.dhbphotography.co.uk

Oliver (6) and Samuel (4) were diagnosed with Hunter Disease in 2002. As any parent with an MPS child knows, a diagnosis like that is shattering and with our world turning upside down, we faced a very uncertain future. Oliver also had a fairly extensive brain haemorrhage at birth and was very poorly, so this was a double blow, trebled by Samuel's diagnosis-he was only 19 weeks old at the time.

My aim as a Trustee is to fight hard to achieve what is required for all MPS sufferers and their families to have hope for the future. MPS families are one huge big, extended family and I would dearly love to be a part of the team who bring the 'Hope' in the MPS Society's motto, forward to 'Today'.



# Announcements

## Dear All

We always enjoy receiving the MPS Magazine and would like to contribute to the costs. Therefore I enclose a cheque for £20. I am not able to fundraise, due to health problems, so hope this is adequate for your excellent magazine.

Keep up the good work, and thank you for all your support to date.

Yours sincerely

**Margaret and son, Louis, and family**

Many congratulations from all at the MPS Society to Cheryl Pitt, Reseacher Officer at the MPS Society on the birth of her son, Jackson Robert on 1 July 2006, weighing 7lb 11oz.



**Congratulations to Isaac Turner (MPS I BMT).** In the first Sale inter Beaver colony football tournament, Issac's team (3rd Ashton) won the tournament in the final on penalties. Isaac played in four matches including the final and was very proud of himself, as were his parents, Adam & Lou Turner.



## New Members

Mr Hilliard has recently been in contact with the Society. Mr Hilliard has a diagnosis of Fabry Disease. He is 55 years old and the family live in the South East.

Ms Donna Halleron and Mr Wayne Clarke have recently been in contact with the Society. Enola has a diagnosis of Morquio Disease. Enola is 2 and a half years old. The family live in the North West.

Mr Harris has recently been in contact with the Society. Mr Harris has a diagnosis of Fabry Disease. Mr Harris is 64 years old and the family live in the South of England.

## Deaths

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

Kym Caines who suffered from Sanfilippo Disease and who died on 27 May 2006 aged 22 years.

Derek Denham who suffered from Hunter Disease and who died on 16 May 2006 aged 31 years.

It was with considerable sadness that we learnt of the death of **Susan Graham** after a brave fight against cancer. Susan, mother of Kerry and Laura who both lost their lives to Sanfilippo disease in 2000 and 2005 respectively, died on 17 August 2006. Our thoughts are with Stewart, Susan's husband, at this time.

### Matthew Di Ilio 1991 - 2006

It is with great sadness that we learnt that Matthew Di Ilio passed away on Saturday 19 August 2006 with his parents and his sister at his side. Matthew was aged 15 years and fought a courageous battle against Sanfilippo B disease. Lori, Matthew's mother, was until recently the Canadian MPS Society's Executive Director and the whole Di Ilio family provided a great deal of inspiration to the Canadian MPS Society and its members over the last 10 years.



## Congratulations!

We were delighted to learn that **Jo Wilson** (MPS I Scheie Disease) has recently got engaged to Alex Smale.

Jo is currently studying medical imaging at the University of Exeter, but met her fiancé Alex whilst working at the Hilton Hotel Bromsgrove. Alex is himself studying Hotel Catering and Management and is a qualified chef. Jo and Alex are planning to wait a couple of years before they marry but we wish them every future success and happiness. Many congratulations from the MPS Society!



# ANNOUNCEMENTS

## Introducing Charmaine

Hi, I'm Charmaine, the newest recruit to the MPS team. My role is also new; I am the Event and Volunteer co-ordinator which basically means that I am the main port of call for the National Conference and other events organised by the MPS Society.

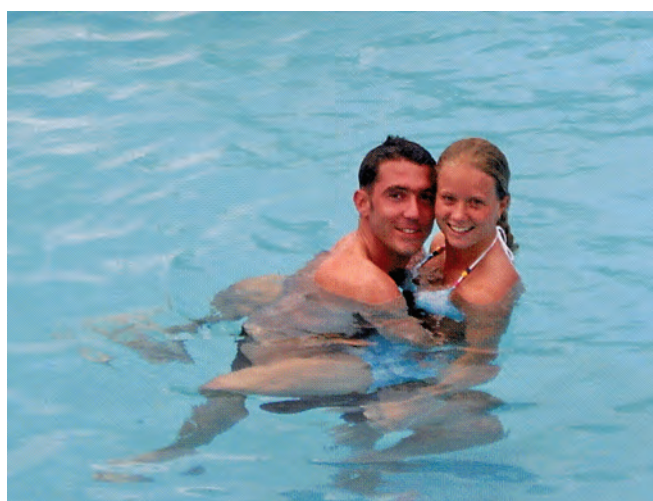
Now to tell you a little bit about myself. After studying a National Diploma in Health and Social Care I went on to further studies, graduating from the University of Westminster with a degree in Criminology and Psychology, a strange but very interesting combination. From University I went on and worked with the Youth Offending Team and in a special needs school as a learning support assistant. This was excellent experience for being hands-on with children with disabilities and I also worked with children who suffer from MPS diseases. This is how I found out about the MPS Society. Although working at the school was very rewarding and very enjoyable I was looking to utilise my skills in a similar area which is just as rewarding.

Now a bit about me and what I do in my spare time; I go to the gym and like to try out the classes such as Kick aerobics, Boxafit and Circuit Training, but I often regret it for the next day or two! I spend time with friends and my boyfriend, Matt, who I bought off eBay for £11.50 (yes, that is the auction website...). All the money was raised for the tsunami, but I'm sure he would have sold for more if the money was for the MPS Society. I must say that I bid for a joke (and because it was for charity) but when I met him we clicked. All in all I keep myself busy with the MPS Society, with the Youth Offending Team and catching up with friends and family.

I hope to meet you all soon and if you know anyone who wants to become a volunteer just call the office or email me at [c.scott@mpssociety.co.uk](mailto:c.scott@mpssociety.co.uk).



Congratulations to **Laura Patterson and Gary Rogers** on their recent engagement! Laura and Gary regularly support the Society as childcare volunteers and will be coming to the MPS Weekend Conference next year. They will be getting married on 4 August 2007 and we wish them all the best for the future!



## Your letters

We are always pleased to receive letters from all readers of the MPS Magazine and especially our members. We welcome letters on any subject and your views and comments would be very welcome.

The MPS Society was delighted to learn that **Mariam Saud** is going to the University of East Anglia to study multi media computing after achieving a distinction in her course and received £25 and a certificate.

All the Freemasons in Buckinghamshire, totalling some 4000 in all, subscribe to the Buckinghamshire Masonic Centenary Fund. They are delighted to assist the excellent work of the MPS Society by funding the training of volunteers by giving a grant of £3,450.

# ANNOUNCEMENTS



Jake (MPS I)

## To all involved with the MPS Society

It has been many weeks since we sadly lost Jake, and we would like to thank everyone for the support we have received, also the flowers and the £100 which went towards Jake's headstone, which has just gone up. Thanks also to Sophie and Neisha who attended Jake's funeral.

Please find below a poem which was read out at Jake's funeral. I would also like to add what a pleasure it was having Jake and we wouldn't have missed a minute.

To any parents starting out with an MPS child, all I can say is treasure every moment. As well as much sadness there will be much joy.

With sincere thanks from

**Clive, Carol, Ricky and Naomi Shaw**

Many congratulations to **Fiona McDowall**, sister of David (MPS II), who has achieved a place at Glasgow University to study Dentistry.



## At My Own Pace

My face may be different  
But my feelings are the same  
I laugh and I cry  
And take pride in my gains

I was sent here to be among you  
To teach you to love  
As God in the heavens  
Looks down from above

To him I'm no different  
His love has no bounds  
It's those here among you  
In cities and towns

Who judge me by standards  
That man has imparted  
But this family I've chosen  
Will help me get started

For I'm one of the children  
So special and few  
That came here to learn  
The same lessons as you

That love is acceptance  
It must come from the heart  
We all have the same purpose  
Though not the same start

The Lord gave me life  
To live and embrace  
And I'll do it as you do  
But at my own pace

Author Unknown

## Angela Lewis Retires

Angela Lewis was one of the original scientists involved in the setting up in 1975 of a specialised laboratory at St Michael's Hospital, Bristol led by Dr Charles Pennock. Charles retired a few years back and in July the time came for Angela to say goodbye and retire gracefully. Angela was a regular attendee at the MPS conferences and supported the work of the Society during its formative years. On behalf of the MPS Society and its members, particularly those in the South West of England who will have benefitted greatly from Angela's dedication over three decades, we send our very best wishes for a long and fruitful retirement.

Looking to the future, the same laboratory services are provided at Bristol Royal Infirmary. Dr Janet Stone who came to the laboratory in 1986 is now in charge of this specialised service and fortunately they have a 'new Angela' in the form of Helen who has taken over the enzyme work in the laboratory.



# Ashley's Great Escape!



I am writing to tell you about what our son can get up to. Ashley has Sanfilippo Disease, MPS III (he was only 3 months old when diagnosed). He is now 15 years old and has fewer trips on his own.

Ash was staying with his grandparents, which would normally be from Saturday afternoon until Sunday evening. It all started when Nanny was upstairs whilst Ash and his Grandad were watching videos in the front room. Then the phone rang and Grandad answered it. After a couple of minutes Grandad realised that Ash was no longer there!

Both Grandparents automatically went first to their neighbour's house, as he would quite often just walk in and sit down to watch television, or he would run across the road to feed the fish in the back garden. He will go indoors and join in with whoever is in the room. But however, this time he was not there. Grandad came to the top of the road where we lived to see if Ash had made it home. He was really worried as he found out that Ash wasn't here either.

He explained what had happened and we all came outside, including his godfather who was visiting. We did not know where to start as we had walked everywhere with Ash. His Grandparents went to the shops and alleyways, dad and his godfather ran towards the pond and headed towards the common. The children split up and went around the block. I was beginning to panic so ran back to get my bike, I pedalled like mad towards the main road.

I saw a number of people that know Ash and asked if I was heading in the direction, they said yes, he was wandering around by the main road. I was really panicking and my heart dropped in to my stomach, I was shouting for him non-stop, not even thinking about him being deaf! It was natural to call to him.

I was getting to the end of the road and didn't know what to do next or which way to go. I stopped and looked to my left and then heard "Mummy!" There he was, oblivious to what was happening, he was pleased to see me, and not at all worried. He was just slowly walking towards me in his Nanny's slippers. Grandad and Nanny pulled up in their car, he casually walked towards them and got straight in. I was glad he was not stressed!

When we got home we sat down and discussed what had happened. If anyone did approach Ash he would not be able to communicate with him or her, he may recognise some words but it is likely he will carry on walking. My main worry is what I can do to let people know about Ash. A talisman would be the answer for most people who are epileptic, but this is not suitable for a Sanfilippo child. He doesn't like things like that; we have tried watches, which he removes as soon as you put them on! Tagging would be a good idea, but you can't do this, as the police would have to be involved and they won't agree it. Identity tags like the ones for toddlers would also be good, however they too would be taken off within minutes. I have thought about having his clothing printed with something like "if I am on my own, please help me to go back home" also including our address and phone number, but this would cause other problems. Another idea was to write a piece for the local newspaper again, however I thought that maybe when or if he did manage to have a wander no one would remember. If anyone shares a moment like this and has any suggestions we would be grateful. We have now invested in a quad bike, so at least we can get to him a lot quicker, and go anywhere. The bonus is that he loves to ride it with his dad.

We can now look at Ash and laugh, I can still see the look on his face as he saw me, his Nanny and Grandad chasing after him, there he was, this quite big young man casually walking around in his Nan's flowers!

I did originally contact the MPS Society to ask if they had or knew of any ideas of how to identify Ashley's needs etc. They thought that this story would show a different temperament and determination and at the end of it he normally expects a chase!

## MEMBERS' NEWS

# Living Independently by Andrew Millar



Andrew (MPS III)

My name is Andrew and I am 36 years old. I live in Scotland. I have MPS III Sanfilippo Disease and have lived independently in my own home for two years. Before moving to my own home, I lived with my family for 28 years then moved into a group home setting. This did not suit my needs and in this day of inclusion, my mum's feelings were, why should I not have the same opportunities as others to live in my own home.

My mum found the home, which is in a small residential area for the elderly and disabled. My mum only lives a few minutes away, so is on hand whenever I need her. It is a ground floor two bedroom flat that suits both me and my carers fine. Before moving in everything was set up between my mum and the provider to provide a homely setting with all necessary alterations and adaptations.

I have a high package of care provided by the local authority to meet my daily living and medical needs. This package of care is what pays my carers to care for me. I have six carers who work on a rotational basis. Any pieces of equipment, repairs or decorating that needs doing, I pay for myself.

My manager and carer are in charge of my money and make sure that it is properly spent on me and all things I require. Mum went to court to appeal a welfare

guardianship order, due to the nature of my condition and the fact that it is a degenerative disease. This award has been given indefinitely, which is great as it means my mum will not have to go to court again to renew the order. It is common for them to need renewing every three years. The reason for her doing this was to ensure that my needs are met to the highest standard and that I had the same rights as everyone else.

My mum works very closely with all my carers some of whom have been with me since I first moved into my home. Together they have jointly set up the whole care package as well as care plans, a daily diary, medication, eating, seizure, toileting records, protocols and procedures for all carers to follow and abide by. Each new carer that joins us has a two-week induction, where they shadow the experienced carers to see how my needs are managed. For each shift there are two carers on hand to support me during the day and one sleeps in during the night to ensure my safety.

All carers before leaving have to complete all records in order that the next shift is fully informed of how I have been. This is vitally important in managing my medical needs and ensuring that people are fully informed of how I have been. Medically I am also well looked after by my GP's, and district nurses when the need arises.

I have a great group of carers who look after me and my mum is always on hand to offer support due to the amount of knowledge she has of my needs and me. We have a great laugh most of the time but my carers all know not to make too much noise if I am unwell or feeling a bit low. I have a great social life and attend a day centre three days a week where I have a good staff team and a large group of friends who I have known for a number of years. On my other days, I have to do the usual weekly shop and we go for walks around the local area or down to the beach. When out my carers take a mobile with them for safety. This way they can contact any person required in an emergency and can get my mum very quickly if needed.

I love having my own place and the independence it brings which I so rightly deserve. It's also good as I get to know a regular team of carers and they get to know my needs and me too.

I would recommend independent living to anyone, especially for young adults as there are not many places within the UK who offer the right kind of service in the right environment.

## Advocacy Support Team views

Members of the Advocacy Support Team visited Andrew, his mum and carers at home to see first hand what it was like for a young adult with MPS III to live independently. Andrew definitely looked happy and the level of care and support he received was second to none. While caring for Andrew the carers are there solely for his needs and everything in the home is set up for him. This is extremely unique compared to living in group accommodation where space, equipment and staff have to be shared and it is not always possible to provide a 1:1 care service to a person with extremely complex medical needs.

Andrew was definitely not isolated by living in his own home and had the choice of being sociable or having time out if needed. He still accesses his local day centre three times a week and does his own shopping with the help and support of his carers.

His home is fully adapted to all his needs with a second bedroom for his carers, which offers both Andrew and others privacy if needed. It is accessible and he also has his own garden.

It was great to see that this type of provision is achievable and that it works. It took a lot of joint working between Andrew's mother and the local authority but all the hard work has paid off and Andrew is very happy and content living in his own home.

If people would like more information on how this was achieved and what is involved, please contact the Advocacy Support Team: [advocacy@mpssociety.co.uk](mailto:advocacy@mpssociety.co.uk)



Andrew's Bathroom



Andrew's Bedroom



Andrew's Kitchen

## CLINICS

# MPS Clinics

### Morquio Clinic

12 June 2006 saw a Morquio clinic at Great Ormond Street Hospital. One concern from families was that more appointments meant more time out of school, and of course the cost of travelling to and parking in London. Niamh Finnegan (Metabolic Clinical Nurse Specialist) had been working diligently to arrange appointments so that more specialists could be on hand at each clinic, thus reducing the frequency of visits. The families in attendance found this to be a huge benefit.

### Sanfilippo Clinic

On 22 June 2006 there was a Sanfilippo clinic at Great Ormond Street Hospital. Following the same model of more specialists on the same day meant that families were able to mingle and chat between appointments. Some families already knew each other while other families were new to the clinic. The new families were able to gain some of the benefits of experience from the others and forge new friendships. Many of those in attendance were also going to be attending the family day at London Zoo.

### BMT Clinic

On 26 July 2006 Dr Ed Wraith and the MPS Society hosted a BMT clinic at the Royal Children's Hospital, Manchester. The clinic was quite well attended and it was certainly a pleasure to meet those who were there. The waiting room did become hectic from time to time, as traffic had led to everyone arriving at the same time! Fortunately we had crisps and chocolate for everyone. We would like to thank all those involved in organising this event particularly Dr Wraith, Jean and Dot.



This page clockwise from top right: Nathan Gremo (MPS III), Sophie Summerton (MPS III), Sam Hitchin (MPS III), Libby Fullalove (MPS III); Page 15 from left to right: Jasmin Heap (MPS III), Reece Culverwell (MPS II)

## Bristol MPS Clinic

The second of Bristol's MPS clinics was held on 29 August at the Frenchay Children's Unit. We had a full day of appointments and everything went very smoothly. We hope that all families found their time with both the doctors and our Advocacy Team useful and informative. It was great to speak with the families there. The next Bristol Clinic is planned for 31 October 2006.

In 2007 we are hoping to arrange four clinics so that we can ensure that families get the time that they need with the doctors and the advocacy team. We would like to thank all the staff involved in the organisation of the clinic and would again like to thank Dr Wraith and Dr Jardine for their support at these clinics.



## FOR SALE! A chance to get some wheels and get into action!

### Manger chair

Sarah Long has a Manger electric chair that she would like to offer to another individual who would benefit from its use. It should be available within the next couple of months (estimated October/November) and offers would be welcome. As you can see from the picture it extends to a fair height and is in good condition. It has not been serviced this year due to Sarah getting her new chair but it still has a good battery and is in good working order.

### Mercedes van for sale

Sarah is also selling her van and it should be available for purchase from Jan 2007. It is in good working order and has been adapted for Sarah to drive but these adaptations can be altered to suit. It is a 1998 Mercedes Vito van, automatic and it has approximately 63,000 miles on the clock. The lift allows direct access to the van and this can be controlled from the inside as well as the outside with a remote control. There is an automatic clamp system in place as well as manual fixing rails for securing wheelchairs. The driving seat is a six way seat (turns, raises and moves forward), but it is unclear at present whether the small driving seat for Sarah will be taken out to go

in her new vehicle so another driver's chair may need to be purchased. The seat mechanisms however will be remaining. There is an extended pedal block which can be removed for other drivers (the tall variety!), electric ignition, handbrake and air conditioning. It was new to Sarah so a full service history is available. Price available on contact.



Both in good working order with lots of life still in them, for sale because of changing access needs. Would like to see them go to good homes. If anyone is interested in any of the above, please contact Sarah on 07885 400492.



## EVENTS

# Family Day at London Zoo

By Ashley Siberini

Sunday July 9th was a fairly typical British summer day for many people, a large outing had been arranged, and there was rain, lots of it. Families from the southeastern parts of England were waking to find that the 'Prolonged Dry Spell' which has caused problems in the area had, in fact, seemingly ended. Ended on a day they were all planning to attend London Zoo! The skies over London Zoo were a little more considerate. Although darkened, early drizzle diminished and the sun began shining as only that sun can.

The day was funded by Help A London Child, a charity that helps less advantaged children. MPS Society staff arrived at the zoo early to greet the families and provide them with the essentials for the day which included money from the Society so that individuals could have better choice of what they wanted to eat. In the same area that day was a charity 'Race for Life' run. Fortunately, we were able to find our member families amongst the hustle and bustle. The whole process was made all the easier by some very kind staff at the zoo.

The zoo itself is currently undergoing some major changes. Some of the key attractions are now based at Whipsnade Wildlife Park where they have more room and better conditions to live in. Some parts of the zoo are located in listed buildings, which meant complications for wheelchair users gaining access. However, there are some fabulous newer buildings that are designed with easy access in mind.

The Bugs building is home to some rather intriguing and odd-looking creepy crawlies. The glass panels were all at a level that suited everyone. Many parts of the zoo had walk-through zones, such as Butterfly Paradise, an Aviary and the Squirrel Monkey enclosure. Other interesting and exotic animals inhabited pens with easy viewing access, so all in all there was plenty to see.

All of those in attendance were invited to colour in the invitation they had been sent from the MPS Society, there was a main and runner up prize for the most original.

MPS Families gathering at London Zoo





At 3pm the families gathered for pre-purchased refreshments and the results of the colouring in competition. William Summerton won the main prize for his very neat and tidy colouring. Michael Oyawale won the runner up for his use of bright lavish colouring in. MPS Society staff bought the prizes on the day to be sure that they suited the recipients.

Many families used this time to catch up with each other from previous meetings or form new friendships with families they had not met before. As the weather was now

nearing glorious we were able to take advantage of the outdoor seating and our younger members could stretch their legs, have faces painted, ride a merry-go-round or visit the gift shop or have a snack.

The day began drawing to a close, the hustle and bustle grew quieter, and families said their farewells. It was a pleasure to see some familiar faces and also some new ones. The main challenge facing us all now was navigating back through the busy London streets. At least the sun was still shining. ■



Photos clockwise from top right: The families of Sophie O'Connor (MPS IH), Roshani Nonis (MPS III), Ben and Emma Perfect (MPS VI), Sophie Summerton (MPS III), Michael Oyewale (MPS IV), Alan Dickerson (Fabry).

## Dates for your diaries!

# 2006/07

20 October 2006	Childhood Wood Planting
25 October 2006	Birmingham MPS Clinic (TBC)
27 October 2006	Midlands Family Day at Twycross Zoo
28 October 2006	Eastern England Family Day at Ferry Farm
31 October 2006	Bristol MPS Clinic
11 November 2006	Adult weekend in the South (TBC)
16 November 2006	Northern Ireland MPS Clinic
17 November 2006	BMT Clinic (Manchester)
18 November 2006	Adult MPS Weekend in the North (TBC)
24 November 2006	Cardiff MPS Clinic
26 November 2006	Northern Christmas Party in Leeds
30 November 2006	Bristol MPS Clinic
9 December 2006	Scottish Christmas Party at Almond Valley
9 January 2007	Bristol Clinic
18 or 25 January 2007	MPS III Clinic, Great Ormond Street Hospital
3 April 2007	Bristol MPS Clinic
10 July 2007	Bristol MPS Clinic
2 October 2007	Bristol MPS Clinic

Everyone in the relevant areas has been sent an invitation to **Ferry Farm and Twycross Zoo**. However, these events are open to all MPS members, so please contact the MPS office if you would like an invitation.

## MPS National Conference 2007

This will take place 29 June - 1 July at the Hilton Hotel, Northampton. On Friday 29 June 2007 there will be a conference on Fabry Disease. Running in parallel will be a children's activity programme for those aged 0-10 years who have Fabry disease and siblings of a Fabry sufferer. For Fabry sufferers aged 11-17 years and their brothers and sisters there will be a one hour mini conference on Fabry Disease led by Dr Uma Ramaswami and Dr Sally Davies, followed by a teenage social activity for the remainder of the day.

On Saturday 30 June there will be three full day symposia running in parallel. One will be focusing on clinical management and social care for children and adults with MPS related neurodegenerative diseases. The second will address clinical management, social development and independent living, whilst the third symposium will provide a unique opportunity to learn about the rarest of the rare diseases i.e. MLII, MLIII, Fucosidosis, Mannosidosis and Multiple Sulphatase Deficiency.

There is a full day programme for bereaved families and adult members which includes time to discover Clumber

Park, a hidden secret in the jewels of National Trust properties, and lunch, followed by a visit to the Childhood Wood and a short remembrance ceremony.

There is a full day children's activity programme planned. During the adult gala dinner childcare and activities will be arranged.

On Sunday 1 July the morning will be taken up with presentations on current research projects for MPS Diseases. We are delighted that Professor John Hopwood from the Department of Chemical Pathology in North Adelaide, Australia will close the programme with a presentation looking at turning research into therapy. This will be followed by Sunday Lunch.

Accommodation is in the Hilton Hotel where the conference will take place and the Marriott Hotel, 3½ miles away. There will be a regular shuttle between the hotels and priority for the Hilton will be given to MPS families and professionals booking for the full conference package.

Booking forms are available from the MPS office.

**A Jeans for Genes Carol Service** is being held at 7pm on **Wednesday 20 December 2006** at St James' Piccadilly. The service promises to be very special, with musical accompaniment and celebrity guest readers. Tickets costing between £15 and £60 are available through the MPS Office.

# CHILDHOOD WOOD

## Remembrance Day

On a wonderfully sunny day in July about 60 adults and children met for the annual Remembrance Day at the Childhood Wood, the Society's special area of Sherwood Forest. It was a day for reflection and celebration as we remembered those who have died from MPS and related diseases.

Most families had lunch at Rose Cottage and then everyone met up at Sherwood Pines Forest Park and walked to the Childhood Wood. It truly is a real wood now and a very peaceful place in which to remember lost loved ones.

The new memory boards which have replaced the plaques previously marking each tree are supported on a dramatic oak structure and form a fitting tribute to all those who have lost their lives to MPS and related diseases.

Coloured helium balloons were attached to the wooden structure (a somewhat tricky exercise as those who were there will recall!).

Barry Wilson welcomed everyone including Councillor Joyce Bosnjak from Nottinghamshire County Council who offered to help any families in the area encountering problems relating to County Council services provided in connection with MPS. Christine Lavery then talked about the recent changes which had been made to the Wood. It was a particularly poignant few minutes when Councillor Bosnjak read out the names of those being remembered that day, followed by Wilma Robins reciting the Christina Rossetti poem 'Remember'.

After releasing the balloons which floated up into the clear blue sky, families walked back and had refreshments picnic style before saying goodbye to each other.

All those who have sadly lost an MPS member in the last 12 months have received information and details of the next planting. However, if you have lost a loved one to MPS in previous years and don't have a tree but would like one at this stage, please contact us without delay.



# VENICE

## A testament by your intrepid Advocacy Support Reporter

Ashley Siberini



Ashley

Symposium originally referred to a drinking party (the Greek verb symptein means “to drink together”) but has since come to refer to any academic conference, whether or not drinking takes place. Fortunately the 9th International Symposium on Mucopolysaccharide and Related Diseases was to be held in Venice, Italy. There can be only few who have yet to hear of the stunning architecture and waterways in Venice and the passion for food and drink that the Italians so rightly boast. Add to that the world’s foremost medical practitioners and researchers for MPS, dedicated families and carers, worldwide MPS Society equivalents and you have a Symposium of potentially great magnitude.

Leaving home at 3:30 in the morning did not immediately conjure such inspiring thoughts however! Travelling with fellow Advocacy Support Officer Steve Cotterell and Trustee Wilma Robins would at least ensure that the perilous journey over the Alps would at least be a team effort. Ok, so we flew from Gatwick to Venice, but those Alps are still perilous!



Kush Tailor, Tom Graham and Richard and Liz Volk

Venice’s Marco Polo airport is about an hour’s river-bus ride away from the island of Lido where the conference was being held. The small island is particularly quiet and far less “touristy” than the main island. Venice’s Lido is an 11-mile long sandbar, home to about 20,000 residents, greatly augmented by the (mainly Italian) tourists who move in every summer. Four hundred years ago, the first casino in Europe opened in Venice and the city became famous for its gambling traditions. The Lido Casino, once the haunt of royalty and film stars is now a congress centre (King Farouk of Egypt, Adnan Kashoggi, the film director Vittorio de Sica, the film producer Carlo Ponte (husband of Sophia Loren), and actors such as Omar Sharif have all graced the building). Its spacious art deco design would be home to the various talks and workshops and some fine dining for the symposium.

Day one of the symposium started at 2:30pm, allowing Wilma, Steve and myself the chance to have a spot of lunch and a shower before arriving at the congress centre. The weather was truly glorious, hot and sunny. Opening speeches by representatives from Italy, Germany, Australia, USA and UK gave a clear sign as to how the international scientific community are working together to improve the quality of life for those affected by MPS. Talks ranged from epidemiology to limitations of current therapies. With most in attendance weary from travel the UK guests made their way to the Villa Mabapa hotel for a get together party at 6 pm (symptein indeed!).

Day two of the symposium (Friday) and the programme for the day was packed full of talks from and for professionals and talks from and for families. Under Italian law medicinal products may not be promoted directly to the user, so the workshops sponsored by pharmaceutical companies could only be attended by professionals in the field. There is tale of past events being shut down due to a breach of this law. Not wishing to incur any negative attention from the authorities all of those in attendance had colour coded identity badges so that the staff at the centre could spot who belonged in which talk. The professional aspect was chaired by our very own Dr Ed Wraith. Sitting in a huge lecture theatre at 8:30am waiting to hear all about the Pathogenesis of Lysosomal Storage Disorders was certainly a first for me. My GCSE Biology really hadn’t prepared me for the scientific assault my ears were about to receive. I blame the mixed comprehensive schools myself.

The family talk for the day started off with Emergency and Heart Problems with accounts from our Italian friends and our own Dr Vellodi (Anglo-Italian translation was provided by staff at the congress centre). Following was a series of talks about respiratory problems associated with MPS. Two

particular talks were by mothers whose children have such problems. Bettina Wildi from Germany delivered a very thorough and thought provoking account of having a child who uses CPAP.

Dr Tomatsu of the USA chaired a session on research into animal models, with very easy to understand talks such as "Morphopathological features in tissues of mannosidosis guinea pigs at different gestational ages". Could you imagine playing scrabble with these guys? The day was rounded off with family talks about feeding problems and orthopedic and skeletal problems. With the warm evening now upon us it was time to head back to the hotel for a little socialising with colleagues, members and their families. A few of us ventured over to St. Marks Square (aka Piazza San Marco) where delicious pizza was consumed and beer bought at roughly £7 per bottle. Seriously that waiter would make Dick Turpin blush! Robbery of the highest order! The pick-pockets who operate in the area have some real competition it seems.

Day three of the symposium was set to be the busiest by far. The entire morning was a combined families and professionals session. It was focused on updates of trials and various therapies with presentations from Doctors Wraith, Muenzer, Harmatz, Hemsley, Tomatsu, Sly and Beck, to name just a few. After lunch the programme was again split between families and professionals. While professionals debated issues surrounding effective delivery of treatment regarding the blood-brain barrier, the families met to discuss eye problems, neurological aspects and psychological aspects. It was great to see so many of our UK members and families talk this particular afternoon. A lot of the talks went over the scheduled time. A clear sign that those in attendance had something very important to say.

The Saturday evening would host the gala dinner, also to be held at the congress centre. With concerns in the air about overbooking of the dinner we had to time to return to the hotel for a shower and change before the dinner, oh we also caught some rotten over-rated football team lose on penalties yet again. During the dinner, which was quite delightful, the Italian MPS Association presented the "Together for MPS Award" to our Chief Executive, Christine Lavery. It was designed to recognise an association which had been the most active in protecting and assisting its members.

With the evening still fairly young and a much lighter schedule the next day a few of us from the MPS Society decided to take a scenic route back to the hotel. In beautiful places such as Venice, one really has to be vigilant and not



Symposium venue

miss any of the stunning scenery, it could be many years or even never that we would be there again. Imagine our delight when some of this aforementioned viewing led us to witness a very prominent UK doctor (not difficult to guess who) 'letting his hair down' and giving it plenty to some classic disco sounds at a party. Swayze and Travolta at their best had nothing on this guy!

The final day, a Sunday, held two morning sessions. One was a professionals session on gene therapy and late breaking news. The other was a family session on looking to the future, after which summaries were made.

A final spot of lunch at the hotel and people began to drift away. Riverboats and taxis took many back to Marco Polo airport. Steve, Wilma and myself had one last tour around the shops before making our journey back across those treacherous alps!



Children's outing to Venice

# VENICE

## A Personal Perspective of the Symposium

Sailesh and Nita Tailor

So why did we decide to go? Well, it started with our son Pavan's diagnosis of Multiple Sulfatase Deficiency in March 1999, one of the rarer diseases in the MPS group of conditions - something like 1 in 1,000,000. The prognosis wasn't very good either, and the fact that Pavan was the only one in the country. It has traits from MPS II, IIIA, IIID, ML and MPS VI, so hearing this just makes you wonder who would be spending money and time on research on such a rare condition. We were certainly proven right, there wasn't any treatment and nothing on the horizon except for symptomatic treatment.

Then one day out of the blue whilst 'Googling' the internet, back in August 2003, we saw the best news since the whole thing started. The news was the gene for MSD had been discovered by an Italian team headed by Andrea Ballabio. We were under no delusion what this meant, we knew any potential treatments leading on from this landmark discovery would still be some time away, possibly not even in Pavan's lifetime. However it was a giant step forward, for other researchers to build on.

In 2005 we heard the 2006 International MPS Congress was being held in Italy. Remembering the earlier discovery was made by an Italian team, we found out the professionals that were taking part and whose name should come up but Andrea Ballabio. Our eyes lit up and we instantly knew we had to be there.



We made the preparations months in advance. We booked Pavan into Acorns Children's Hospice knowing full well the logistics for his travel and care during the conference would be complex. There was one snag with this arrangement which hit us a week before the trip, and that was we have never left him for that length of time coupled with the distance we were apart. To say we were sad is probably an under estimation.

Finally we were there and our primary objective was to meet Andrea Ballabio, and find out what was happening on the MSD front. Secondary objective was to gain further knowledge from the other talks and meet and discuss with other professionals and families. To this end we had prepared several copies of Pavan's history and medical reports in order to hand it to the professionals that were either researching MSD or had potential treatments that may apply to MSD. The handout was also to alert these people to say we are here, and that MSD was not just a condition, but the fact our son has it and your research may help him.

We were pretty happy on meeting Andrea, and to learn mouse model gene therapy trials under way. Also when human trials do come about, he will inform us.

Christine Lavery and her team made our stay so much better and we were very privileged to be there when she was awarded the magnificent gift in recognition of her hard work. She is truly the force behind making MPS an international success story.

There was a further bonus with our elder son Kush who came with us and is not affected by the condition. He thoroughly enjoyed his time with the UK carers who came, to the point he was perfectly happy not to have us around. He is looking forward to coming with us to the UK conference next time.

Lastly, the Lido was a lovely location and our trip to Venice before we headed back home was fantastic.

Thanks to all the people in our group, whose company we enjoyed. See you next time...

Sailesh, Nita & Kush and not forgetting the young man himself Pavan.

# Volunteering in Venice

By Lucy Lavery (Hannah Donegani, Beth Nichols and Tom Graham)

Upon arriving at the Morrizini Child Centre in Venice, where the children's programme for the 9th International Symposium on Lysosomal Storage Disorders would be held, we were separated into groups represented with a coloured hat depending on disorder. Us four (Tom, Lucy, Beth and Hannah) have had a great deal of experience of MPS either through many years of volunteering or having siblings with MPS. With our knowledge of MPS children it made us laugh that the Italian volunteers thought that the children would conform to their strict ideas of organisation. To the annoyance of the Italian volunteers, we were no better at conforming than the children. They were not pleased when Tom in orange group (Sanfilippo) was helping to look after Jackson who has MPS II so was in blue group. The futility of trying to stereotype MPS children has never been more apparent than when caring for Andrew Hawkins and Jackson Whittaker.

Jackson's an 8 (almost 9) year old Australian boy with MPS II. He's very fun, sometimes outrageous and can be extremely hyperactive. On the other hand, 23 year old Andrew Hawkins has Sanfilippo manifested in a form never seen by us as volunteers. Provided he is comfortable, Andrew is quietly affectionate, sensitive and content. The most obvious contrast between the two was their personal opinions on cleanliness. Jackson couldn't stay clean for more than a couple of minutes, whereas Andrew managed to ensure that not only himself but the rest of us remained clean. Andrew was probably the only person wearing an orange hat to do so.

After the first day at the centre we had already realised that four days in the one place probably wouldn't be fun for Andrew, Jackson, Sophia and Kush so on the first evening we made plans to break out of the centre at lunch time the next day and tour Venice mainland. After all we hadn't come all this way to stay in a children's centre. Luckily the morning before as responsible volunteers we had done a brief risk assessment of Venice. Of course we discovered it to be entirely unsuitable for the Irish Martina's wheelchair, Jackson's tendency to run off and Andrew's dislike for large crowds. We just saw this as a challenge! But before we pushed our volunteering skills to the max, the Italian's had one final shock in store for us: open glass flasks of red wine served on the children's tables at lunch in the children's centre. This came as such a shock to us that we all had to have a sniff before believing our eyes. We don't know whether it was our mature sense of responsibility or the amount of wine consumed the night before that kept us on the water.

Mainland Venice seemed like a specially designed obstacle course for MPS;

boats, steps, bridges, canals, pigeons and as always the hardest to negotiate - crowds. After pizzas, ice cream and coke we were ready to explore. Jackson walked the streets of Venice chatting up street sellers and making friends with anybody along the way. One of Jackson's best friends was a Chinese tourist in a sparkling white shirt. The man didn't have much choice as Jackson, armed with a choc-ice, sat so close to him that he was almost on top of him. Jackson and his new best friend had their photo taken as a reminder of their brief but beautiful friendship. And as if by some miracle the man's shirt remained spotless.

We made slow progress along the streets of Venice, covering 20 metres was an achievement what with toilet breaks, Jackson's bartering and us three girls lifting Marina and wheelchair over bridges whilst big, strong, Italian men looked on. This is why we valued the help of two siblings, Kush Tailor and Sophia Volk who became unofficial volunteers. Sadly for Kush he ended up resembling a packhorse carrying Martina's stroller and three rucksacks. Sophia quickly earned Andrew's trust and affection. Despite Andrew being almost twice Sophia's age they became the best of friends and Sophia was the envy of us who were rarely rewarded with hugs.

As if Venice mainland wasn't challenge enough, the next day we embarked on a parent's worst nightmare...the beach. No situation illustrated the contrast between Andrew and Jackson more than being on the beach. Whilst Jackson was content to be covered head to toe in sand and found nothing more pleasurable than doing the same to Tom, Andrew's obsession with cleanliness lead him to "sweep the messes off the (ocean) floor". This produced a large shell collection.

Volunteering in Venice was hard work but an incredible experience. I think I speak on behalf of Tom, Beth and Hannah when I say that Jackson, Andrew and Martina were great company and that the help of Sophia and Kush was invaluable. Thank you MPS for giving us the chance to meet some great people, experience really interesting situations and, of course, taking us to Venice!



# My trip to Venice...

Myles Broughton (MPS I HS)

I was asked to do a talk on reaching my educational potential for the International Symposium in Venice. My mother and I spoke of going to the Symposium in Venice as we shared an interest in Roman history and were going to make a holiday of it, but unfortunately, my mother passed away before Christmas. I felt that it would be best to go for her sake and so I agreed to do the talk.

With the expenses of the conference being so high my family couldn't afford to come with me, so I went alone. Before the flight out I was understandably nervous, having never been to Venice or Italy before and had limited information to go on. The flight went okay. My electric wheelchair was loaded aboard and arrived into Venice undamaged. This was a relief. I met with the other members of the Society who had flown on the same flight as me but we were scattered about all over the plane, because we didn't all check-in together.

We caught the bus to the boat transfer. They were both wheelchair friendly. The bus had ramps and even the controls for them were located in the wheelchair bay. The only trouble was the instructions on how to use the controls were in one language, Italian; so none of us knew how to operate it and had to rely on the driver. The boat transport was adequate, but best used when the tide was in when using a wheelchair! There were occasions at times when the chairs needed to be lifted from boat to land.

Venice's water transportation system was like having a water version of a motorway, there were speed limits marked out, markers marked out and people drove their boats in the same way you would on the road. The lifestyle was almost electrifying and infectious. The weather also compliments it.

We were collected and directed to our bus which had been arranged to take us to our accommodation. I was staying at the Villa

Mabapa which is part of the Best Western hotel chain. On arrival I was directed to the rear of the hotel where the wheelchair access to the hotel was supposed to be. Their definition of wheelchair access was a nice long wooden ramp with two steps at the top. First hurdle to overcome I thought. Sophie dealt with it though and I got a ground floor apartment.

My apartment had a double bed made out of two single beds and I had two bathrooms. I did forget that I'd given Sophie my passport, which the reception staff kept hold of. So later that night I turned my room upside down, because I thought I'd lost my passport. I was in a real frenzy and called Neisha and Neisha reminded me that the reception might have it, which of course they did, but I had them return it to me. I had also forgotten to make a photocopy of my passport in case I lost it. I registered myself at the Casino (the venue for the Symposium), which was an old building. I was given a red backpack, talk timetable, tourist information, tickets for a boat trip and gala dinner.

The casino had a large theatre with a speech stand and a table where speakers can sit to answer questions. The seating area was large. I didn't spend much time in this room, although most of the seminars was held in there. This was Thursday and later that night I had a meal with staff members of the MPS Society. I gave them a giggle.

I had been booked on a boat trip the following day. A minivan picked me and some others up and took us to the boat. The boat took us on a very scenic tour and the sea looked very blue and vibrant. I took lots of photos and followed the tour. I was glad to get out as the diet coke in my room was really expensive and I was anxious to find a cheaper source. As it turned out Venice are selling coke light.

Later I went back to the casino to take a look round the building. The main entrance had



steps up to it and I had to go round the back and up the lift to get inside. The building appeared to be historical. We were kept to three floors. The first is the ground floor and nothing really happened there. The second floor had the reception area, and the stands for the pharmaceutical companies. The third floor was where the food was served.

I went out for dinner with some members of MPS staff near Piazza San Marco. We tried to get to St Marks Square, but when we got to the stop where we were meant to get off we found a bridge which didn't have any access for wheelchairs and so we tried going down some streets to find a way round. We ended up at a restaurant where we ate outside listening to the Italians cheering their team on. We ate and talked for a while and went back to seeing if we could get to St Marks Square again. We got back on the boat and got off at the next stop which got round the problem with dealing with the bridge.

We found the Square and went for a drink. Whilst relaxing, people approached us numerous times trying to sell roses. I eventually gave in and bought Neisha, Antonia and Sophie one each as they had been kind to have me with them. The stroll from the boat port to the hotel was pleasant. We met up with other families who had gone to a live classical concert on the way back to the hotel.

The next day was Saturday and the day of my talk. I was nervous and itching to get it over and done with. I had spent most of my time in my chair as I didn't want to leave it anywhere and so I was starting to ache. So I thought I'd talk standing up, which was fine to start with, but then the heat and pain started to get to me and I was struggling to focus on my lines. I got a standing ovation, but I felt I had messed it up so much I didn't notice it.

The gala dinner was good and Christine was given an award. It was a unique experience to see multiple cultures meeting in one place and having translators translating the language so everyone could understand what was being said in the seminars.

Sunday I stayed in the hotel and watched a new cruise liner sail past. I didn't feel like going to the Casino. I had one last lunch with the MPS group and later after lunch we said our farewells to everyone staying behind.

We had arranged for a water taxi to take us to the airport. On the way we hit a wave which sent our baggage bouncing in the air, and my bag nearly took a dive into the sea. Luckily it was saved, but it had us all in stitches, especially Sophie, Antonia and Christine who knew it was my bag and laughed at me for ages! On the way an aircraft was landing as it was passing over us and the air currents from under its wings pushed the boat slightly downwards into the sea and it was a strange sensation.

Now in the airport, there was a mutual stand-off as I was checking in. I had to wait for wheelchair assistance as part of airport rules, but Venice Marco Polo Airport only had three members of staff on duty and so they were trying to make us wait. Christine Lavery soon put a stop to it. We ended up sitting on the baggage conveyor belt to stop anyone else from being served and Christine called for the supervisor. We sorted it and I made it safely back to Gatwick Airport having got some duty free goods on the way. The one shocking thing of the whole conference which really got my attention was how little information other countries had on MPS and how limited they are compared to us in the UK with getting treatment. ■



Christine Lavery, Myles Broughton and Barry Wilson

VENICE

# International MPS Network



Over 24 years ago the UK MPS Society was founded by a group of parents united by one common bond; they all had a child or children suffering from a Mucopolysaccharide or Related Lysosomal Disease.

Within a decade affected families from many countries in the developed world, United States, Canada, Australia, New Zealand, and much of Western Europe had an MPS Society or representative patient organisation to turn to for support and information.

Today there are MPS Societies in South America, Japan and the whole of Europe. Whilst united by one common cause and similar aims and objectives, no two MPS Societies are the same and each one is autonomous from the others. However, this autonomy may well be their strength – able to meet the needs of MPS families taking into account the economical, cultural and geographical situation of their country.

All MPS Societies offer a range of support and information whilst a growing number are also funding vital scientific and clinical research. The role of the MPS Societies is not exclusive to affected families; the Societies need and value the excellent support and help they enjoy from key professionals across the globe.

Today the MPS International Network is a collaboration of energised MPS Societies and the families they support, scientific and clinical experts in the field and a much welcomed and growing pharmaceutical industry working to make a difference in the lives of those affected by Lysosomal Storage Diseases.

## International MPS Network Meeting

29 June, Venice

Twenty organisers from thirteen countries participated in this lively half day meeting preceding the International Symposium on MPS and Related Diseases in Venice.

During the meeting, Steve Holland (USA) explained the process of using a Yahoo group to set up and maintain consistent communication between the International MPS Network. This is particularly important as several MPS patient organisations did not receive their electronic invite and missed out on the meeting. It was agreed that the UK MPS Society co-ordinate the Yahoo group in the first year and that two people from the recognised international patient organisation for

MPS in each country will have access to the Yahoo site.

A motion was put forward and carried recognising 18 patient organisations from Europe. From now on, groups being established in countries where there is no current MPS patient organisation can apply for membership by sending an application in the form of a letter to the International MPS Network, C/O the UK MPS Society for approval.

Following considerable discussion, a motion was carried to designate 15 May 2007 as International MPS Awareness Day.

A proposal to host the 2007 International MPS Network Meeting was made by the Polish MPS Society. A motion was carried to hold this meeting 24-27 May in Poland.

A proposal was made by Kirsten Harkins to host the 2008 International Symposium on MPS Diseases in Vancouver, British Columbia, Canada from 25-29 June 2008. Steve Holland put forward a motion to accept Vancouver as the host city of the 2008 meeting. This was announced at the conclusion of the International Symposium on MPS Diseases that followed Venice.

The Society has recently been sent copies of the Fabry news-sheet from the **4th International Fabry Patient Meeting**, held in Paris last October. The news-sheet gives an overview of information shared at the conference, as well as personal perspectives from patients. If anyone would like a copy of this news-sheet, please could you contact the MPS Society.

During the International Symposium in Venice, two very prestigious awards were made. Both awards are glass masterpieces produced by the most famous Venetian glass artist 'Archimede Segusa'. The artwork represents a hand-shaped tree carrying an egg containing the DNA double helix.



Christine Lavery and Professor Neufeld

The first award, entitled a 'Life for MPS' was awarded to **Professor Elizabeth Neufeld** of Los Angeles, California, in recognition of her achievements as a pioneer in the field of Mucopolysaccharide and Related Diseases.

Since the early 1970s, Professor Neufeld has worked to determine the pathophysiology and later the molecular mechanisms of these diseases, drawing the guidelines for the modern therapy.

The Italian MPS Association has also established a 'Together for MPS' award assigned to the organisation that has been the most active in protecting and assisting its own members.

The 'Together for MPS' was presented to the UK MPS Society's Chief Executive, Christine Lavery, in recognition of the many years she has spent in fostering links to other MPS Societies around the world. The founder and driving force of the UK MPS Society for the last 25 years, Christine's continuing efforts to help other Societies and encourage cooperation between those Societies, particularly in the field of research, has been the catalyst for the formation of the 'International MPS Network'. It is that network which I believe will encourage future co-operation in ensuring that the efforts from each MPS Society around the world is not duplicated. Also, that the governments of each of the member Societies realise that they have a powerful force to deal with when it comes to any legislation that may be required to ensure that each of our members, that could benefit from any available treatment, does actually receive it.

Well done Christine, we are all very proud of you and look forward to seeing the masterpiece on display at MPS House.

**Barry Wilson**  
Chairman of Trustees



# VENICE

**Judy Holroyd**, a Trustee of the MPS Society, attended the Symposium. Judy has summarised two of the topics covered in the sessions for readers of the MPS Magazine...

## Neonatal Screening

### Background

Newborn screening is one of the largest screening programmes in the UK and each year over 600,000 newborns are screened. The newborn blood spot screening programme is a very successful screening programme and the uptake is more than 99%. It was introduced in the UK in 1969 for phenylketonuria (PKU) and in 1981 for congenital hypothyroidism (CHT). In Wales, newborn screening for Duchenne Muscular Dystrophy was introduced in 1991 and for cystic fibrosis in 1996. It is hoped to extend newborn screening for cystic fibrosis to the rest of the UK by April 2007. Also in England there is a well advanced programme in place for the phased implementation of newborn screening for the inherited blood disorder, sickle cell disease.

### Summary

The session on neonatal screening for lysosomal storage disorders (LSD's) focussed on new technologies for the diagnosis of LSD's that could use the dried blood spot form the traditional heel prick test.

Professor John Hopwood from Adelaide, Australia, introduced the session on neonatal screening for lysosomal storage disorders (LSD's) with an overview of the new technologies for the diagnosis of LSD's that could use the dried blood spot from the traditional heel prick test. He was enthusiastic about the introduction of newborn screening for MPS disorders in Australia.

Dr Michael Gelb from Seattle, USA described a technique called tandem mass spectrometry that provides a way to analyse a large number of chemicals extracted from the dried blood spot sample. These tests are being developed for use in newborn screening laboratories in conjunction with a pharmaceutical company that is manufacturing some of the products needed for this new technology.

The next presentation from Dr Peter Meikle from Adelaide, Australia described the use of protein markers using immune-quantification assays and of metabolite markers using tandem mass spectrometry for the identification, at birth, of individuals who have an LSD.

Dr Maria Fuller, also from Adelaide, followed by describing a technique called electrospray ionisation-tandem mass spectrometry that is being used in her laboratory. It is hoped that this approach will provide a more detailed picture of disease activity in patients, leading to improved methods of diagnosis, prediction of clinical severity and biochemical monitoring of therapy.

The fourth technical presentation was from Dr Gabriela Nilzawa from Buenos Aires, Argentina who described a pilot study that showed it is possible to evaluate the activities of lysosomal enzymes in routine samples from a newborn screening programme.



Judy Holroyd

The final presentation was from Dr Fiona Stewart, Consultant in Medical Genetics, Belfast, Northern Ireland. She emphasised the ethical issues in newborn screening in that any new programme introduced should start from the premise 'to do more good than harm'.

Many of the previous speakers had emphasised the paucity of knowledge about the incidence and natural history of LSD's and the fact that there is only treatment for a proportion of LSD's. All the speakers raised the issue that the introduction of a newborn screening programme would need careful evaluation before it was implemented although it is likely that the technology will be developed in the not too distant future.

Dr Stewart suggested some advantages of newborn screening. These included the opportunity to implement early treatment especially with the advances in the treatments for MPS and other storage disorders. Secondly, if a diagnosis is made early, then accurate genetic counselling could be given to parents who could then be offered prenatal diagnosis for a future pregnancy. Thirdly, an early diagnosis may minimise the extensive tests that are given to a child before a diagnosis is made

Some disadvantages she raised were the affect on the parent-child relationship in that it could deprive the parents of the time they have with their child before the diagnosis is made. Also, how careful consideration would need to be given on how a positive diagnosis would be given to families and a clear management pathway would be vital. She also raised the issue of whether the early availability of genetic information and hence reproductive choice justifies giving the parents the knowledge that their child has a life limiting disorder for which there is no cure.

Dr Stewart ended by emphasising that it is important that the wishes and needs of families affected by LSD's are paramount when considering the ethics of newborn screening for these disorders.

## Update and Perspectives on Therapy for the Mucopolysaccharidoses

The leading clinicians from around the world who have been involved in clinical trials for MPS I, MPS II and MPS VI and therapies for MPS III, MPS IV and MPS VII presented their perspectives on the therapeutic potential of these treatments.

Dr Ed Wraith started with an update on the clinical trials for MPS I. He reported that recently, as a result of Phase 1, 2 and 3 clinical trials, Aldurazyme has now become commercially available as a long term enzyme replacement therapy for patients with MPS I. The results have shown the product to be safe and to have alleviated many systemic signs and symptoms of this progressive multisystemic disease. Sustained improvements in lung function and general mobility (improved joint range of motion) as well as sustained reductions in levels of the storage product in urine were found to be significant. Also reduction in the size of the liver and in the frequency of excessive daytime sleepiness and irregular breathing at night were found to be significant.

Dr Wraith also gave an overview of other studies that have been initiated to widen the experience of the use of ERT in MPS I. These included a study in patients under the age of 5 years, many of whom had the most severe form of MPS I. He reported that, even in this group a broad range of positive results have been observed in many organ systems.

Dr J Muenzer followed with an update of the clinical trials for the MPS II and the recent press release from the MPS Society announced that FDA approval has been granted for 'Elaprase' the ERT for the treatment for MPS II, Hunter Disease. Further trials are now in the planning stage.

Dr Harmatz from the USA reviewed the results of the clinical trials for MPS VI that showed patients on Naglazyme treatment had increased their height and had improved lung function as well as showing progression in physical development.

Dr Hemsley from Adelaide followed with an overview of recent laboratory research aimed at providing a safe and effective therapy for all patients with MPS III Sanfilippo disease. As yet, all these lines of research are still in the early stages of development.

Dr Tomatsu reported on the current approaches to the treatment of MPS IV. He described a potential ERT application that used a 'tagged' enzyme to enhance the delivery and clinical effectiveness of the product to the bone marrow.

Dr W Sly from the USA described how the work in his laboratory on animal models for MPS VII may have relevance in extending the benefits of enzyme therapy for all lysosomal storage disorders.

Dr M Beck from Germany concluded this session by discussing different methods of monitoring the efficacy of treatment for these complex multi-systemic disorders. Apart from measuring physical outcomes in patients he suggested that biomarkers (substances in the blood or urine whose levels can indicate the presence or extent of disease) could be helpful tools in diagnosis of a given disease and for monitoring the response at a cellular level to potential lines of treatment. He emphasised that the advantages and limitations of potential biomarkers have to be evaluated by observations in many patients.

If you would like to find out more about any of the issues covered here, published abstracts can be requested by contacting the MPS Office.

# INTERNATIONAL

## Einar Grønvik Bachke

31 December 1986 - 11 June 2006

In January 1988, Einar came from South Korea to Oslo, to be a son to Knut and Oddrun and a brother to Helga. He was just over a year old. In that year, he had been separated from his biological mother, lived with relatives, then in a children's home and finally with a foster mother to prepare for adoption, before he was given to them. When he died, he was almost nineteen and a half. For someone with a severe condition of MPS II, that is a long life. Einar had lasted past all expectations, and when he died, he did it in the best way possible - by drifting off to sleep in the middle of a family party, surrounded by people who loved him, and not waking up again.

Einar's first year is not an ideal start in life, but he quickly showed that he had come through it very well. After a while it was also obvious why he was better equipped to cope with such experiences than most of us would have been. He carried within him a wellspring of love for and trust in his fellow man that one couldn't help responding to. He had trust in life - that it is good to live. He trusted other people - that they were good and wished the best for him. And above all, he had trust in himself. Einar took utterly for granted that he could reach out, understand and be understood, share sorrows and joys - mostly joys. Right up to the end he strove to get to know every new person he met, and

to show that he saw, recognised and appreciated them, and there are many new faces to learn for those who need a lot of assistance, as Einar did.



He was always a kind and considerate boy. There are many memories of Einar trying to comfort someone in trouble, or get them help. He was deeply shocked the first time he met a boy with his own disorder at a later stage of development, not because he was frightened or repelled, but because nothing could be done to help, really help. He stayed beside that boy for a long time, trying to cheer him up, and often returned to the photos that were taken, showing that he remembered and thought about this visit.

After Einar was diagnosed in 1990, the family got in touch with the UK MPS Society, and for some years, MPS in Norway, indeed the Nordic countries, was fronted internationally by an adoptee from Korea. The connection to the MPS Society became very important, and not only because it gave Einar access to specialised medical advice and a lot of practical support. It also became a link between MPS in Norway and England, and in the end of important benefit to all Norwegian MPS families. But from Einar's point of view, the best thing was all the new people, the visits to England, and later trips to MPS symposia in Essen and Paris. His last trip abroad was to Stockholm a few months ago, where he enjoyed seeing a number of old MPS friends, and was allowed to take part in the conference dinner at the Vasa Ship museum!

Einar had to carry a heavy burden through his disease. He grieved deeply and for a long time, when he at six years lost his balance, and with that, his mobility. He could no longer run and play together with other children. He cried his heart out then more than once, and his family and friends grieved with him. But he came through that, accepted that this was the way it was, and that his illness had to be lived with, and in the end MPS and all it implies, matured him ahead of time. One might say that he mastered the role of an invalid. The driver who took him to school remarked last winter that he could feel Einar keeping an eye on his driving, and that he knew Einar wouldn't put up with any sloppiness.



This was a feeling shared and now affectionately remembered by the staff at Einar's care home where he spent 50% of his time. He knew every detail in his own routine, and they could trust him to let them know if anything seemed wrong, but also to take pleasure in all they did to keep him interested and entertained.

Einar's courage and joy in life made a deep impression on many, and some put this impression strikingly into words. One au pair from the Phillipines who looked after him, thought that Einar's coming to live with his family should be seen as a vote of confidence from Our Lord. A close friend once said that Einar's task in life was to make us all see what a human being IS, when you remove the external decorations. And that was in fact a very cheering sight.

All people are different, including those with MPS. Einar was able to show us that his illness, though a dominant force in his life, was only a small part of him. He was glad to have the life he had, but at the end, also probably glad to be able to go. He had done as much as he could, and he will not soon be forgotten. ■

## INTERNATIONAL

## Our Journey into the Unknown...

# Fucosidosis



Suzan, my dearest sister, was a perfectly normal child born in 1967 in London. My family decided to move to Cyprus permanently in 1972 when she was 5 years old and I was 7.

Suzan had red spots on the knees and elbows and her lower back as well as on her chest. We later discovered that they were called angiokerotomas. We could not find a cure for it and the professionals in Cyprus did not know the cause or the reason for them.

When Suzan was 14 and studying in secondary school, she failed her exams and could not pass her class, so had to study the same class the following year. We didn't think it was abnormal, as we knew that she had other talents like embroidery.

Suzan started having ear infections and her tonsils were removed at around the age of 16. She also had an operation on her adenoids.

As she was bored at home she started to do work experience in a family friend's factory at the age of 18.

Suzan soon stopped eating and drinking at home saying that she was eating at work. She lost weight and her periods stopped. She soon developed hallucinations.

Cyprus is a small country and the doctors we visited did not know why she suddenly stopped eating and was acting the way she was. Suzan was terrified all the time. At one stage she also refused to go to the toilet. At one stage Suzan was so bad we could not cope with her anger and temper. She kept talking about killing herself, she complained that she was very ugly and thought she had hair growing on her arms, she thought that she had no teeth in her mouth. She was saying that there were soldiers following us and they were going to kill us. On our way to the doctors, she was worried that we were going to get lost.

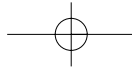
Her temper became so bad that she threw a knife at my mum at one stage. She was actually going through anorexia (no-one knew). We visited every psychiatrist, psychotherapist, counsellor, general practitioner, medical consultant and hospital in Cyprus. At this stage she was taken in as an inpatient at a mental health clinic. The antidepressants and medications were making her situation worse.

In 1986 my father decided to come back to London to seek help. They registered at a local GP. The GP didn't know what her condition was, so he referred her to St George's hospital where Suzan went through a full check up and was finally diagnosed with Fucosidosis.

We did not know anything about this disease. There was not enough information around and the only thing we knew was that this was a very rare disease and there was no cure for it. As soon as we heard the bad news my mother and I decided to move back to London to care for Suzan as her condition was deteriorating rapidly.

Over the years Suzan had lost hearing completely in one ear and had limited hearing in the other. Therefore she is now wearing a hearing aid. She also developed brittle bone condition in the spine, neck and hips, which is affecting her mobility. Suzan was on antidepressants and painkillers ever since we can remember. But she was still complaining from pain in the hips, knees and legs.





Suzan's angiokeratomas were getting very severe on her genital area. It was becoming very uncomfortable for her. Suzan also developed obsessive-compulsive disorder through her life. Therefore, she went through several laser treatments to remove these unpleasant berry red spots.

Suzan developed incontinence problems and was finding it difficult to control her bladder and in some cases her bowels.

We were left in the dark for many years until 2000. This was when I learnt how to use the computer and the Internet and started to use the Google search engine for information on Fucosidosis. Suzan was 33 years old when I found out that the life expectancy for people with mild Fucosidosis was 40 years. I was gutted but also determined to overcome this. The more research I did on the Internet the more questions were answered in my mind.

We finally knew what to expect and that every symptom Suzan had was linked to this genetic disorder. There was still no cure for this disorder but there were definitely things to comfort Suzan and I was determined to find it. It wasn't until I typed Fucosidosis in the search engine again one night when I came home from work and found out a website called ISMRD that our whole life was beginning to change.

I discovered that this was an umbrella organisation for similar disorders and there were real people that really cared and really wanted to help. There was a conference about this disorder and I was determined to go. I emailed Paul Murphy and got a response immediately. Suzan and I were invited to this conference. We had financial difficulties. So, I searched for financial support and grants towards our travel from organisations in London but unfortunately could not succeed. I told Paul about this and Paul agreed to sponsor us. I also managed to arrange a medical assessment for Suzan while we were in America at John Hopkins Hospital. We packed up and travelled to America to find out everything about this disorder and to meet other families and learn from their experiences.

This was an eye-opener for us. I was amazed with the whole idea. I met so many kind and helpful

people. I found out so much information. I made so many friends in such a small time. Best of all, I found out about an infusion that might be a cure for Suzan's ongoing pain. Pamidronate was an infusion Jenny Noble was very keen and happy to use with her children. I wanted to give it a go. Jenny gave me all the information and the protocol for using this infusion.

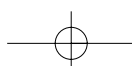
At the medical assessment I discussed this with the consultant and he was happy for Suzan to try it. Suzan was discovered to have a murmur in her heart which caused sleep apnoea, which was not discovered in London. I was gutted. My darling sister could have died in her sleep and we would not have known the cause to help prevent this. Our consultant at John Hopkins Hospital gave us a 12-page report on Suzan's condition and he also included a future plan for her. I gave a copy to all the doctors that Suzan was seen by in London so that they would know and would follow.

I came back to London and immediately changed Suzan's Specialist Consultant, as he had no experience in this field. Again, thanks to the Internet and the conference, I read and heard a few case studies by Dr Vellodi. I found his email address and emailed him regarding Suzan's condition and asked for an appointment. This was arranged and we were in the hands of an experienced consultant. He agreed to treat Suzan with pamidronate and this was arranged in January 2005. Suzan had a bone density test and this was -2.5. Suzan has been on this infusion for over a year every month and is completely free from painkillers. She is now much more active, happy and pain free.

With the way Suzan's bones were deteriorating, had we not known about this infusion at the right time, it would have been too late for the infusion to help improve Suzan's condition.

Therefore I owe a huge thank you to Paul Murphy who supported us all the way, to Jenny Noble for her endless encouragement and support and to Dr Goswall for arranging us the appointments at John Hopkins Hospital. I also want to thank everybody I had met at the conference and wish you the best of luck with your loved ones. ■

**Aydin Kulle**



## INTERNATIONAL

# Rare Diseases Workshop

31 May 2006, Brussels

It was an enormous privilege to be invited by the Austrian Presidency to the European Union to speak at a meeting organised to explore and consider the impact of rare diseases on the European population and what needs to be done by the European Union to address rare diseases.

Mucopolysaccharide Diseases were brought to the attention of the Austrian Chancellor by Michaela Weigl, President of the MPS Society in Austria and her daughter, Maria, who has Morquio disease (MPS IV). Convinced of the unique challenges facing families throughout Europe affected by MPS and other rare diseases, the Austrian Government decided to prioritise rare diseases during the Austrian Presidency and act as a co-ordinating body in Europe encouraging research and treatment.

The British MPS Society expresses nothing but gratitude to the Austrian Presidency for bringing such vital awareness of MPS to the decision-makers in the European Council.

## BUGGY FOR SALE

Rosemary and Harry Nurse have an Alvema Buggy for sale. It has a low back, 15" wide seat, a harness and four new tyres.

£900 new, they are looking for offers in the region of £100. Please contact the MPS office for more information.

Republik Österreich  
Dr. Wolfgang Schüssel  
Bundeskanzler  
Gesellschaft für MPS  
Gesellschaft für Mukopolysaccharidosen  
Und ähnliche Erkrankungen  
Finklham 90  
4075 Finklham

26 June 2006  
GZ BKA-405.975/0006-IV/5/2006

Ladies and Gentlemen

Austria as having the presidency of the EU in the first half of 2006 aims to accomplish realistic goals for the citizens of the European Union. I intend to give notice that the chiefs of state of the EU have agreed at their meeting on June 15 and 16 on a topic which is especially important to me: I personally support a special programme fighting rare diseases like i.e. Epidermolysis Bullosa (EB) or Mucopolysaccharidosis (MPS) which affect mainly children. The European Commission is ready to function as a coordinating instance and to support the efforts of all members.

The additional profit of a coordinated effort to fight rare diseases shall be visible by the efficient use of finances which have been provided for by the 7<sup>th</sup> EU programme for research and development to bring diagnostic, preventive and therapeutic measures.

I believe this conclusion is important because the number of patients affected by such diseases is so small in every European country that national programmes can only be developed with many compromises. Therefore I am very pleased by the readiness of all members and also the European Commission to choose a new common approach.

My great appreciation goes especially to you who achieve so much through your daily work for all those affected by these diseases. I wish to inspire your courage by those conclusions of the European Council to successfully keep up with your work.

## MPS Research Programme 2007

The MPS Society is pleased to invite applications for its 2007 MPS Research Programme. Applicants are advised to familiarise themselves with the Society's Terms and Conditions of Medical Research Grants, submission instructions and application process before submitting an application. All guidance and application forms are downloadable at [www.mpssociety.co.uk](http://www.mpssociety.co.uk).

Deadline for submission of Grant Proposals: **24 November 2006**

# INFORMATION EXCHANGE

## Benefit updates

### You may qualify for a reduction in your council tax

If you are disabled or have a disabled person (adult or child) living with you, you may be entitled to a reduction in your council tax. If you were successful in an application it would mean that you would be charged a band lower than what your property is worth and what you are currently paying. (i.e. if you are band C you would go down to band B).

To be considered you need to have one of the following:  
An adapted room for the disabled person (i.e an extended bedroom, living space), an additional bathroom and/or have had to make provisions for extra space to allow for a wheelchair to be used in the home.

### So how do you apply?

To apply you need to contact your local council who will send you a form to complete and return. A Council Officer may then want to visit your home to see what adaptations/alterations have been made and see records of benefits received, for example, DLA. They may also contact your doctor for more information. All applications for the council tax benefit will be backdated to the date the application was originally made.

### VAT relief, the facts

You do not have to pay VAT on equipment designed for a disabled person or if you have had something adapted. This can cover things such as;

Building work, servicing and maintenance, installation of equipment, hire or purchases of disability equipment such as vehicles, wheelchairs, beds, hoists, gadgets (for example grabber stick, kettle tipper) chairs and computer software or hardware especially for a disabled person.

However, this can be difficult as the rules are complex and unfortunately not everything will qualify for VAT relief. It is therefore important that you check whether you qualify to receive VAT relief and that the product, service or adaptations being purchased or provided, qualifies for the exemption.

### Am I eligible for VAT relief?

To qualify for VAT relief the law states that you must be chronically sick or disabled (i.e. have a physical or mental impairment that has a long term severe effect on your ability) or have an illness that doctors treat as a chronic sickness or be terminally ill.

### So what do I do?

Before you purchase services or equipment always check to see if it qualifies for VAT relief first. You may have to complete a form declaring that you have a chronic illness or disability and what is being purchased is for personal and domestic use.

You do not have to purchase the service or equipment first then reclaim the tax back from the government. You can see an example of a VAT relief form on the HM Revenue & Customs website ([www.hmrc.gov.uk](http://www.hmrc.gov.uk)).

### Help paying for heating & insulation improvements

You may be able to get help from the government's warm front scheme if you receive income or disability related benefits (such as working tax credit, child tax credit, attendance allowance, DLA, income support, housing benefit, council tax benefit, pension credit, income based jobseekers allowance) and need help to pay for heating or insulation improvements to your home. This can apply whether you privately own your home or rent it.

### What can you use it for?

You can use the benefit to improve things such as loft insulation, draught proofing, hot water tank insulation, gas, electric or oil central heating, converting open fires to glass fronted fires, timer controls for electric space and water heaters and energy advice.

### How do I apply?

You can apply for a grant over the phone on freephone 0800 3166011. They will then send you an application form to complete and return. A Home Energy Adviser would then want to visit you to assess your property.

## The Human Tissue Act

New legislation relating to the use of human tissue, the Human Tissue Act, comes into force on 1 September 2006.

In summary, the Act sets out the requirements for consent of human tissue to be used for various purposes, including research. At present, asking for consent to use tissue in medical research is part of good research practice, but the Act will make this a legal requirement, along with the requirement for ethical review.

The Act will also establish the Human Tissue Authority (HTA) who will issue licences for storage and usage of tissue for the purposes of the Act, again including research. The HTA will have the power to inspect premises where human tissue is stored and used.

One MPS Society member has recently told us about a recruitment consultancy called Talent Match Ltd. **TalentMatch Ltd is a specialist HR consultancy supporting employers to attract and recruit talented disabled people.**

With an emphasis on merit based recruitment and a commitment to an employer led approach, TalentMatch Ltd works with employers, recruiters and specialist brokers to attract disabled people with the ability to do the job and the desire to make a difference.

Their consultants are experts in the field of HR, disability confident search and selection, and reasonable adjustment management, ensuring a business driver resourcing solution every time. Phone 01442 877406 or visit [www.talentmatchrecruiting.co.uk](http://www.talentmatchrecruiting.co.uk)

# Holly Bank School

By Donna Lowther



When my children were first diagnosed with Sanfilippo Disease, MPS III, the whole family was devastated, as I am sure most families who have children with MPS are at the beginning. We know how these diseases progress and what will eventually happen but it is quite difficult to envisage how these changes will affect the family and how these changes will be managed. We all want the best for our children and most of us can make sure that they have the correct level of care whilst they are at home, but how can we be sure that they are getting the same level of commitment to their needs elsewhere.

Before my children were diagnosed, my daughter, Siobhan, attended mainstream school (reception class) where aspects of her behaviour changed from the way she behaved at home. Obviously we did not understand the reasons for this and my daughter was diagnosed with learning difficulties and was transferred to a Moderate Learning Difficulties Special School. I was pleased to see that her behaviour settled down at this stage. In hindsight I don't think that she could cope with the amount of children who were in the mainstream reception class. When she was six and her brother, Shaun, was four, they were both diagnosed with Sanfilippo. I have to say the Headmaster was a different matter. He saw this as an opportunity to get funding for physiotherapists using my children's illness. Needless to say I was annoyed at his lack of compassion (that's putting it mildly).

I realised at this point that I wanted somewhere for my children to go that would be able to accommodate them throughout the stages they would inevitably go through so that they were not constantly moving from school to

school as their condition progressed. The children's Social Worker, OT and Educational Psychologist all agreed and we started to look for different schools for them. It was the teacher at the school they were attending who saw an article about a school in Yorkshire that might be able to accommodate their needs so we requested more information from them. When we received the information pack I must admit that it seemed too good to be true. However, we contacted them to arrange a visit.

The school was Holly Bank and was situated in Mirfield, West Yorkshire. The thought of sending my children to a school that was far away was daunting. It was a two hour drive from our home in Gateshead to the school. The children's Community Nurse and Social Worker accompanied the children and myself for a visit. The staff were very welcoming and made us feel at ease. They even looked after the children while we had a look around. We all agreed that the school seemed ideal for both of them. The only thing I was worried about was that all of the pupils were in wheelchairs and at this point my children were at the hyperactive stage and needed to be able to run around. I needn't have worried because they bought some soft play equipment and a ball pool and made sure that they had plenty of freedom to run around.

Originally they attended Monday to Friday coming home at weekends, but as their condition deteriorated they came home one weekend every half term and school holidays. The school has Speech Therapists, Physiotherapists, Occupational Therapist, and Nurses all on site as well as a multitude of care staff, teachers and classroom assistants. Everyone is known by his or her first name and they are extremely dedicated to their roles. They have a flat where families can stay if they need to visit their children. Nothing is too much trouble for any of the staff.

Sadly, both my children have passed away and I cannot begin to tell you the amount of support the family received from everyone at Holly Bank. My son attended the school from the age of six to thirteen and my daughter from the age of eight to nineteen when she left school. The school has gone through many changes in the years that I have been associated with it and all for the better I might add. The decision was hard to send my children away to school but it was the best decision that I could have made for both them and myself. Being a single parent I do not think I could have coped as well as I did but for the staff at Holly Bank. If any other family is looking for a new school then I can thoroughly recommend Holly Bank.

# INFORMATION EXCHANGE



Siobhan (page 32), Shaun (above)

**Hollybank Trust**  
 Far Common Road  
 Mirfield  
 West Yorkshire  
 WF14 0DQ  
 Tel: 01924 490833  
 Fax: 01924 491464  
 Registered Charity No. 1043129  
[www.hollybanktrust.com](http://www.hollybanktrust.com)



### Whose needs do we meet?

Holly Bank is both a school and a 52 week children's home. We work with young people aged between 5-19 years and their families. All our learners have complex physical disabilities with associated and often severe learning difficulties. These include serious sensory dysfunctions and degenerative medical conditions requiring significant medical and therapeutic input. Our provision is complemented by the seamless transition from childhood to adulthood through the continuity of our adult care both on site and in the community.

### How do we meet these needs?

Our holistic multi-disciplinary approach ensures every young person receives the guidance and support needed to live life to the full. We provide a homely environment within a safe and secure setting. Our on-site facilities are second to none with a newly opened state of the art multi-activity hydro and therapy centre complemented by innovative assistive technologies, including Eagle Eyes and Camera Mouse, for which we are the UK designated centre. We provide each young person with a stimulating individually tailored 24-hour programme delivered by a committed team of staff in a tranquil, caring environment. Within each 24 hour programme young people access equipment, facilities,

expertise and experiences that enhance opportunities for inclusion in the wider world.

In school we deliver a sensory and developmental curriculum based on the National Curriculum. Students in our post-16 department develop empowering life skills. Achievement is applauded both internally and externally through national accreditation.

In our Ofsted inspection in March 2006 it was noted that 'pupils are extremely well supported through excellent work with an extensive range of professionals including occupational therapy, speech and language therapists, physiotherapists, ICT technicians and medical personnel. The input of these groups is very well documented, recorded and fully supports pupils' educational and personal progress. Similarly, very good links with parents increase adults' knowledge of pupils'.

### Who meets these needs?

Hollybank Trust employs over 300 staff, including education staff, all those mentioned above and social care staff. The teams are supported by administration staff and a wealth of visiting consultants from medical, educational and social care fields.

# INFORMATION EXCHANGE



*Your local children's hospice*

Acorns provides care, help and support to hundreds of families living in the West Midlands and Gloucestershire regions whose children have an illness for which there is no cure - a condition that prevents them reaching adulthood. On average, every week one of the children that Acorns cares for will die.

Acorns provides respite, emergency and end-of-life care in three hospices located in Birmingham, Walsall and Worcester. A dedicated community team offers further 24-hour support to the whole family in their homes including practical and emotional support for parents, special help for siblings and bereavement counselling. Acorns Hospice Walsall and Acorns Hospice Worcester have specialist adolescent facilities, the Walsall Hospice being the first in the country to introduce these.

Acorns services are available to all families within the West Midlands and Gloucestershire regions and the demand for these vital services grows daily.

Acorns is the largest provider of high quality and unique care and support for life-limited children and their families in the UK, offering support to over 500 families in the Heart of England. Acorns has helped well over 1200 families in the last 18 years.

With 30 beds, and given the present average lengths of residential stay per year, Acorns plans to be supporting around 640 children plus a further 300 bereaved families by 2010. Acorns also provide active support to at least 150 siblings.

Acorns has around 1000 volunteers and 300 paid staff. Volunteers work in all areas, in the hospice, with families in the community, in fundraising, in commercial offices and particularly in Acorns retail shops. There is no charge for Acorns services but running costs stand at £4,500 per day per hospice, that's £450 per bed. Only 9% of its funding comes through statutory sources and Acorns is therefore reliant on fundraising and donations to aid its work.

In January 2006, Acorns Children's Hospice Trust sadly announced the temporary closure of 12 beds in its three hospices due to lack of funding. In May 2006, despite a marked increase in donations it was announced that the beds would remain closed until further notice.

Closing beds within the three hospices was undoubtedly a tough decision for Acorns to make but one that was needed to ensure no compromise was made to the overall care or the quality of its services in the longer-term.

In May, the Department of Health announced that it was providing £27 million of funding for children's hospices across England, over three years. Through the Association of Children's Hospices, Acorns is working with the Department of Health to work out how the funding will be distributed. While this is fantastic and hugely welcome news, Acorns will continue to rely on voluntary support for the majority of its income.

It seems likely that the funding Acorns receives will be around 10%-15% of its overall costs. Put another way, it would fund Acorns services for less than two months of the year.

To find out more about Acorns, please visit [www.acorns.org.uk](http://www.acorns.org.uk)

## Information Session at Acorn's Hospice

On 23 August, Neisha Hall and I were invited to Acorns hospice (Worcestershire site) to share information on the MPS Society and MPS diseases. I had given a talk earlier in the year at the Walsall site which was very well received and was something that they were wanting to incorporate into all their training programmes to ensure all staff were informed and up to date with recent developments.

Unfortunately due to a bad accident on the M5, Neisha and I arrived a little later than planned. However we still had time to share information about the Society and the support that we give to our members and an overview of many of the MPS conditions, how they affect individuals and what treatments are currently available. This generated a lot

of discussions and staff at Acorns were able to gain the information that they wanted from this. While we were there, we also offered to come back on a separate occasion to give more information on specific diseases. This was well received and staff spoke about us coming back in March 2007 and doing a whole day of training on MPS and related diseases.

After the training we had the opportunity to look around the hospice and sample some of chef's fabulous lemon cake.

We would like to thank all the staff at Acorns for making us so welcome and we look forward to returning again next year. **Sophie Denham**

# RESEARCH & THERAPIES

## Enzyme Replacement Therapy (ERT) for patients with Mucopolysaccharide Type II, Hunter Disease, approved by FDA

Shire plc today announced that the US Food and Drug Administration (FDA) has granted marketing approval for idursulfase (ELAPRASE™) an enzyme replacement therapy for the treatment of Hunter Disease, also known as MPS II.

Hunter disease is a rare, life-limiting, genetic condition, mostly affecting males, caused by the deficiency of the lysosomal enzyme iduronate-2-sulfatase. Without this enzyme, cellular waste products accumulate in the tissues and most organs. Hunter disease comprises a wide spectrum of severity and some children with the severe form of the disease have progressive developmental delay and usually die before reaching their mid teens whilst others have an attenuated form of Hunter disease suffering little or no central nervous system damage and can live well into adulthood. In all cases, Hunter disease causes progressive cardiac disease and physical disability and makes it very difficult for adults to live independently.

'I was disappointed to be a couple of years too old to be included on the MPS II clinical trial. Over the last two years my physical condition has deteriorated. News of the FDA approval of Elaprase™ is just what I wanted to hear and can't wait for the EMEA to give their approval so that I too can start on this much needed treatment' – James

'After waiting years for this treatment, I am so glad it has been approved so that others like me but not

### Hunter Disease – did you know?

Charles Hunter who gave his name to Hunter Syndrome first described the condition in two brothers in 1917. After graduating from the University of Aberdeen, Charles Hunter undertook postgraduate training in London and Berlin before emigrating to Canada where he settled in Winnipeg practicing at Winnipeg General Hospital and specialising in internal medicine.

During the First World War Charles Hunter served in Europe as an army medical officer and in 1910 was appointed to the Faculty of Medicine at the University of Manitoba. In 1928 Charles Hunter was appointed Professor of Medicine, however, he was unhappy with the tedious administrative responsibilities and resigned the following year. Charles Hunter who was regarded as the leading diagnostician in Western Canada continued teaching and retained his private practice until only a few years before his death in 1959 at the age of 82.

fortunate to be on the MPS II clinical trial for Elaprase™ can now benefit' – Colin

This therapy will have a profound impact on the lives of children and young adults in the UK with Hunter Disease when it is approved by the European Medicines Evaluation Agency (EMA) hopefully before the end of 2006.

## Early access to Elaprase for MPS II in Europe

Following FDA approval of Elaprase, early access for Hunter patients not involved in the MPS II clinical trial have been granted to European patients funded by the particular countries health service:

3 patients in Germany on treatment and 17 being assessed  
3 patients in Spain  
2 patients in Sweden  
17 patients in Italy

### Access to Elaprase for MPS II in the UK

As it stands all UK MPS II patients not on the clinical trial will have to wait until Elaprase is licenced. All MPS II patients and their families known to the MPS Society were recently invited to indicate their expectations of treatment. Over 70% responded suggesting 21 patients living in England in addition to the patients already on the clinical trial expect to access treatment. In fairness to the NSCAG centres, particularly Manchester who have by far the largest cohort

of MPS II patients, the Society asks that MPS II patients and their families realise that not everyone can be first and that the treatment will have to be phased in. We would urge you to be patient and if you are worried or uncertain at the situation that you share this first and foremost with Dr Wraith and his team. It is wonderful that at last there is hope in this way and we all need to work together as over 30 MPS patients in England get on treatment and are satellited out to local hospitals.

In Scotland the access is more complex as the Scottish Medicines Consortium (SMC) expect a company to submit an application for their product to be funded. The SMC will then evaluate Elaprase against cost effectiveness and decide whether to recommend a drug for funding. In Wales the procedure is similar through the All Wales Specialist Medicines Group (AWSMG). In Northern Ireland, they follow a process mirrored on NSCAG for England.

## BREAKING NEWS!

It has been agreed that NSCAG will continue to commission the LSD service for a further year, from April 2007 to March 2008. This is good news for MPS Patients!

# How are you going to make an impression?



Everyone likes to get dressed up.  
So on **Friday 6th October** jump into your jeans and make a donation to help raise £4 million to fund pioneering research and vital support services for children affected by genetic disorders.

**Make an impression on a child's life today.**

**For your FREE fundraising pack visit:**

**[www.jeansforgenes.com](http://www.jeansforgenes.com)**

**or call freephone:**

**0800 980 4800**



**Jeans for Genes Day**

**Friday 6th October 2006**

**Dress to Impress**

Ten national charities working together to help children affected by genetic disorders

The net proceeds from the 2006 Jeans for Genes Appeal will be distributed among the charities

Jeans for Genes Campaign Reg. Charity No. 1062206. Logo and 'Jeans for Genes' © CGDRT. Reg. Charity No. 1003425