MPS

Care Today, Hope Tomorrow

Autumn 2008

Society for Mucopolysaccharide Diseases

CARE TODAY, HOPE TOMORROW

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. Our aims are to:

To act as a **support network**

To bring about more **public awareness**

To promote and support **research**

What we offer

Advocacy Support

Telephone Helpline

MPS Befriending Network

Support to Individuals with MPS

Regional MPS Clinics

Information Days and Workshops

National & International Conferences

Sibling Workshops

Information Resources

Quarterly Magazine

Bereavement Support

Research & Treatment

'Mucopolysaccharide and Related Diseases are individually rare; cumulatively affecting 1:25,000 live births. One baby born every eight days in the UK 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.'

Cover photograph: Dave and Laura Brodie and their son, Will (Hunter Disease, MPS II) with Tom and Kim Whitecotton and their son, Scotty, who also has Hunter Disease. Both families attended the International MPS Symposium in Vancouver, Canada, Summer 2008

MPS Society

MPS House, Repton Place
White Lion Road, Amersham
Bucks, HP7 9LP, UK
T: **0845 389 9901**
Out of Hours: **07712 653258**
F: **0845 389 9902**
E: **mps@mpssociety.co.uk**
www.mpssociety.co.uk
Registered Charity No. 287034

Magazine Deadlines

Winter	1 Dec 2008
Spring	1 Mar 2009
Summer	1 Jun 2009
Autumn	1 Sep 2009

Become a Friend of MPS

Subscriptions may be taken out from the UK or overseas by contacting the MPS Society's Office. The articles in this magazine do not necessarily reflect the opinions of the MPS Society or its Management Committee. The MPS Society reserves the right to edit content as necessary. Products advertised in this magazine are not necessarily endorsed by the Society.

CONTENTS

CHIEF EXECUTIVE'S REPORT

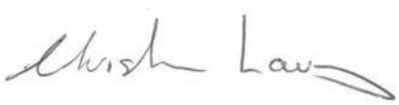
Autumn 2008

Following the announcement of two natural history studies preceding clinical trials for MPS IVA, Morquio Disease, the Society hosted its first single disease conference for those affected by MPS IVA, their families and professionals, 29 – 30 August 2008 at the Hilton Hotel Northampton. Families and professionals from sixteen countries attended. Only a few weekends previously, 23 children with brothers and sisters suffering from MPS took part in an action-packed sibling weekend at Longleat Center Parcs in Wiltshire. You can read more about these events in this MPS Magazine.

Later in the year, some of our young adults will be enjoying a weekend together in London and some of our North East families will be making a special visit and having lunch at Newcastle Football Club. The event preoccupying our thoughts at this moment is the programme for our National MPS and Fabry conference to be held 26-28 June 2009 at the Northampton Hilton. If any of our members would be interested in speaking we would love to hear from you.

There is a variety of news from the MPS office. Those of you who have attended events will know that Miriam Blowers is expecting a baby in November. Miriam will be leaving us shortly to go on maternity leave. We wish Miriam and her husband, Duncan, every good wish as they await the arrival of their first child and look forward to welcoming Miriam back in 2009. We have also said goodbye to two of the advocacy team, Neisha Hall and Chris Murphy. Neisha who has been with the Society over 3 years and Chris a year, are moving on for personal reasons and we wish them well for the future.

Finally, these are difficult times for us all and we do appreciate that there is little spare money for charitable giving once the household bills have been met. However, MPS does rely on your help particularly in these bad economic times. Income has dropped substantially in the past few months so if you can help with fundraising or encourage family and friends to raise funds for MPS this would be most welcome. Christmas cards are now available and we would also like to hear from anyone interested in taking part in the London Triathlon next year. Whatever the case, please be assured that your donations and fundraising, however small, will be put to excellent use; supporting children and adults with MPS and related diseases and their families; providing individual advocacy, bringing people with MPS together at events around the UK; providing sibling and young adult weekends and investing in important research.



Christine Lavery
Chief Executive

HIGHLIGHTS 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 12 July 2008.

Election of Officers

Barry Wilson was re-elected as Chairman, Wilma Robins and Bob Devine were re-elected as Vice Chairs. Judith Evans was re-elected as Treasurer.

Personnel

Approval was given to arrange for the MPS office partitions to be changed to accommodate a fourth individual office for the Senior Advocacy Officer.

Research

The MPS I Survey carried out by Natalie Durkin, a summer intern studying medicine at Birmingham University was presented as a poster at the International Symposium on MPS and related diseases.

The MPS II Longevity and Mortality Study carried out in 2007 by Lucy Lavery, a summer intern studying Human Genetics at Nottingham, in collaboration with Dr Ed Wraith of the Royal Manchester Children's Hospital has been accepted as a poster at the American Society for Human Genetics meeting in Philadelphia, 10 – 15 November 2008.

Trustees who had previously approved the student internship to carry out the MPS III natural history study, were advised that the questionnaire had been approved and interviews would commence 14 July 2008.

Trustees were given an overview of 'Tipping the Lens', a psychosocial research project for Morquio being carried out at the University of Bath.

Clinical Management and Treatment

Trustees were updated on the situation regarding access to Enzyme Replacement Therapy in Wales and Scotland.

Jeans for Genes

Trustees were advised that MPS is working well with the Jeans for Genes charity and jointly had been successful in securing Genzyme and Shire as corporate partners for the schools packs and Fabry video.

Risk Management

No changes were made to the Society's risk management register at this time. Trustees discussed the Society's reduction of projected income and Trustees agreed to review this monthly.

Policies

Two new policies were agreed, Whistleblowers and Professional and Personal Boundaries, subject to some amendments. The title of the Grievance Procedure (Trustees) was altered to Grievance Procedure for Trustees.

Society for Mucopolysaccharide Diseases

The MPS Logo

The MPS Society has become aware that its logo is being used on a number of websites to link to the MPS website and to promote the MPS Society. The MPS logo is a registered trademark and can only be used in situations where the MPS Society has given prior written consent. The MPS Society limits the use of its logo because it does not want to be seen as endorsing either an individual or an organisation. The MPS Society appreciates that the logo is a convenient way to link a website to the Society's own website but this can be easily achieved by attaching the link to the reference to the website. We hope this clarifies the situation.

MPS Events 2008

CLINICS

Friday 10 October	Manchester BMT Clinic
Friday 17 October	Manchester BMT Clinic
Tuesday 4 November	Bristol MPS Clinic
Friday 21 November	Birmingham MPS Clinic
Friday 5 December	Cardiff MPS Clinic (tbc)

REGIONAL PROGRAMME

Friday 24 October	Childhood Wood Planting Day
Sunday 7 December	Newcastle Christmas Party
Sat 29 - Sun 30 November	MPS Adult Weekend

NEWCASTLE CHRISTMAS PARTY
Sunday 7 December 2008

The Newcastle Family Day Event has been moved from 30th October to 7th December 2008 to provide a Christmas Family Day. We are looking at having a lunch together, Children's Entertainer and some other Christmas Festivities at the Newcastle Stadium!

PLEASE CONTACT THE OFFICE BY PHONE 0845 389 9901 BY 31st OCTOBER TO BOOK YOUR FAMILY PLACES.

Once you have contacted the office there will be a booking form that will be posted out to those wanting to come.

Please Note:
The maximum number we can subsidise within each family booking is 2 Adults, MPS Sufferers, and Siblings under the age of 18 years.

Additional adults and childrens can be booked in for the Newcastle Christmas Party at £20.00 per adult and £10.00 per child.

More details regarding the timing of the day and directions etc will be sent out once we have finalised numbers.

The MPS Society must have a minimum of **six** families booked in to attend the event to make it viable to take place.

If you have any queries please contact the MPS Society on **0845 389 9901**.

New Members

Mr Simon Greening has recently been in contact with the Society. His son Ethan has a diagnosis of Hurler Disease. Ethan is nine months old. The family live in Wales.

Mrs Joan Lyons has recently been in contact with the Society. Joan has a diagnosis of Morquio Disease. Joan is 51 years old. She lives in the South East.

Mr Dale and Miss John have recently been in contact with the Society. Marshall has a diagnosis of Sanfilippo Disease. Marshall is eight months old. The family live in Wales.

Mrs Hudson has recently been in contact with the Society. Her son Harry has been diagnosed with Hunter Disease. Harry is four years old. The family live in the North West.

Mrs Abdul Hamid has recently been in contact with the Society. Adnin and Aqilah have a diagnosis of Morquio Disease. Adnin is 8 years old and Aqilah is 5 years old. The family live in the East Midlands.

Ms Pemberton has recently been in contact with the Society. Samuel has a diagnosis of Mucolipidosis II, I-Cell Disease. Samuel is 10 months old. The family live in the North East.

Mr and Mrs Butt have recently been in contact with the Society. Their son has a diagnosis of Hunter Disease. Ali is 5 years old. The family live in the South East.

Mr Michael Illingworth and Ms Fiona Goulding have recently been in contact with the Society. Oliver and Benjamin have a diagnosis of Sanfilippo Disease. Oliver and Benjamin are five years old. The family live in the South East.

Deaths

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

Lauren Wainman who suffered from Sanfilippo Disease and who died on 2 February 2008 aged 15 years.

John Purdham who suffered from Morquio Disease and who died on 24 July 2008 aged 45 years.

Robert Glover who suffered from Sanfilippo Disease and who died on 13 September 2008 aged 12 years.

Christopher Leask who suffered from Sanfilippo Disease and who died on 20 September 2008 aged 35 years.

Reehan Ali who suffered from ML II and who died on 24 September 2008 aged 3 months.

Births

Congratulations to Dr Ed Wraith whose daughter Jenny gave birth to a baby boy, Dylan James Hughes, on 7 July 2008 weighing 7lbs 5oz.

Congratulations to MPS Trustee, Judy Holroyd, on the safe arrival of her first grandchild, Jack William Holroyd, on 21 July 2008, weighing 8lbs 1oz.

This is a picture of Craig and his first niece Maddison (we call her Maddie) who was born 24 March 2008 weighing in at 7lbs. She is gorgeous and also clear of any MPS disease. **Julie Hone**

Congratulations to Joanne Johnston, daughter of Margaret Coyles and sister of Aiden Kearney (MPS IV). Joanne gave birth to Niall Brian Johnston, born on 4 September 2008 weighing 7lb 8 oz.

ANNOUNCEMENTS

Victor McKusick, the Father of Medical Genetics, has passed away

Many of us will remember him for his pioneering descriptions of mucopolysaccharidosis lysosomal conditions at a time when clinical medicine was the art of history and physical examination and without the benefit of sophisticated laboratory testing. His observations were tremendously insightful and stood the tests of time and the molecular genetics revolution.

Public services were held in Baltimore and Maine during the first week of August.

More information can be found by visiting http://en.wikipedia.org/wiki/Victor_A._McKusick

Ann Canton remembers MPS

Those who have been members of the MPS Society since the 1980's and 1990's may remember Ann Canton, from West Wales who was diagnosed with Scheie disease. Ann and her husband Mervyn attended many of our MPS conferences and family days, taking a great interest in the younger families. Sadly, Ann's husband died a few years ago and Ann battled on losing her life to MPS on 15 September 2007.

Ann was a keen supporter of the MPS Society in her lifetime and remembered MPS in her will. The Society was most grateful to receive a legacy from Ann of £40,649 in recent weeks which will help considerably towards support for MPS members at a time when raising funds is particularly difficult.

If you or family members are writing a will, a donation to the MPS Society however large or small would be wonderful. For more information, please request our Fundraising Factsheet on Legacies.
fundraising@mpssociety.co.uk

If you would like help, guidance or information from the MPS Society's advocacy team please do phone us on **0845 389 9901** or email **advocacy@mpssociety.co.uk**

In Memory of Victoria Headland

Here are two poems that were written many years ago by Mary, a night nurse who helped to look after our daughters. Victoria died when she was 21 and Katie was 20, they both had Sanfilippo disease. Mary and Ray, her husband, became very dear friends of ours and were such a great support to us when we were caring for our two daughters. One poem was written during the night before Victoria's 21st birthday and the other the night before Katie's 17th birthday.

Even now, after all these years, Peter and I find these poems very moving. **Pauline Headland**

Victoria's thoughts in the wee small hours
On this my very special day
If I were able I would say
Thank you for my twenty-one years
Of tender care through laughter and tears
I may not clearly see or hear
But I feel your love as you draw near
You hold my hand and smooth my hair
Buy me pretty things to wear
You're never cross when I start to cry
Two arms wrap round and I feel you sigh
Then I am safe and sound again
Your devotion is plain for all to see
We love you both

From Katie and me xx

You know the Nurse who comes to our place?
You know the one, keeps washing my face
Well, she said, '1st August Kate, what do you say,
How about thanks for your Special Day'
So, thank you God for my Mum and Dad
For my two Nanas and one Granddad
Big brother Andrew 'cos he's real cool
Who else? Oh yes, those up at the school
Watch over Tora, my sister and friend,
Please keep her beside me right to the end
Tell Mum and Dad I love them the best,
They know how I'd love to be like the rest
Wear make-up; go for a drink with the boys
Sing, dance and make one hell of a noise
But, I mustn't grumble for you to see
I love all the love that is given to me
O.K. so I'm not like all of my peers
But I've had seventeen very special years.

Love Katie xx

Life without Luke

It is almost one year now since Luke David Morrison passed away. It was on 17th October 2007 at 5pm that Luke could no longer fight his MLII condition and passed peacefully from this world into the next. Luke died in his mother's arms with his daddy looking helplessly on at Stirling Royal Infirmary, where he had been born three years and three months earlier. We miss him very much and our family of one older son, James, and one younger son, Finlay, is now incomplete. We miss him every day and his presence, smile and fun-loving nature has left our hearts and home much emptier places.

Following his death we received much support and good advice, especially from parents who themselves have been bereft of a child in similar or more unexpected circumstances. To date some of this advice has been followed and most is now being acted upon as time passes - ever the more quickly. In many ways we wish that time, work and domestic life would stand still to give us a chance to reflect more fully on our precious time with Luke and the magnitude of his loss. We get caught up in the business of every moment and we can feel guilty of not thinking as often of Luke as we ought.

On the other side, time is a healer, which is lasting truth, but you never 'get over' the loss of a child and we do not ever expect to get over the loss of Luke. We are learning to live life without him, 'gone but not forgotten' as the saying goes. James was 5-years-old when Luke passed away and he has probably coped better than us with the loss. We tell him that Luke is now in Heaven and that explanation for now is sufficient for a young, uncomplicated and uncompromised mind.

We have been thankful for the support of our families and church. Our faith in God has put things into perspective and allowed us to make sense of why Luke was born with such a degenerative condition and why he lived for such a short time. There will always be questions and little in the way of answers but we think it better not to ask questions that we will never fully know the answers to in this life, our hope is that one day all shall be revealed and made clear.

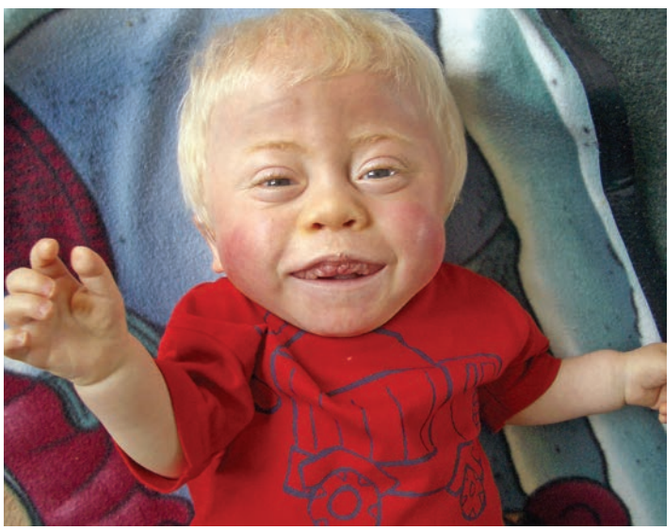
On Luke's headstone we have inscribed the verse from Isaiah 40: "He gathers the lambs in His arms and carries them close to His heart." We take much comfort from the hope that today Luke's soul is in the everlasting arms of Jesus and he is now in a better place than we are.

Luke taught and continues to teach us many things about ourselves and life. We try to remember as best we can what things we can learn from his three years with us. Patience and spending time with one's own family is of paramount importance as is enjoying the simple things in life. Luke loved trees. His sight was impaired, and he defiantly would not wear his trendy glasses, but he would frequently 'talk' to the trees overhead on his many walks in the pram. He also loved being around people; we think he even liked being in hospital as he got plenty of attention from his entourage of adoring nurses and the undivided attention of mummy and daddy.

Luke had a simple trust in people and treated everyone as a potential playmate. He would knock his tower of cups over so as to get your attention and then give a mischievous chuckle, knowing that you would build them back up for him to demolish again. He also had a helter-skelter musical toy where he deposited a ball at the top and collected it at the bottom. He loved music and would join in his favourite songs with actions and rhythmic tapping of his feet. Chocolate buttons were a huge favourite of his and the mere sight of a packet resulted in a broad grin. At night he had two simple requirements: his soft toy Dou-dou the cat and his shawl. With such simple pleasures he was content.

We would like to take this opportunity to say thank you to the consultants and nurses that cared for Luke in Stirling Royal Infirmary and for giving him every opportunity to get better and get back home. They treated him like a normal child with all the potential to thrive and did not discriminate on the grounds of his prognosis. We would also like to say thank you to the consultants for listening to us and asking for our advice when necessary; that concerted effort probably gave Luke a little longer to be loved and cherished by his family and many friends. We are also grateful to the MPS Society for their support both when Luke was alive and in his passing. We appreciated Neisha and her colleague coming the long distance to Luke's funeral and for the flowers we received in sympathy.

To end we would like to have written a nice poem to encapsulate our love of Luke but words fail. Going back to trees, we liken Luke to a Cherry Blossom. In spring the tree flowers and shows its full and beautiful radiance for a very short period of time, the winds come and the flowers are no more. Luke lived to his full potential and he was beautiful. **Ewen and Hazel Morrison**



Oliver and Samuel's Story

Back in March 2002, we were thanking our lucky stars that our 2 and a half year old little boy, Oliver, was doing so well. He had had a traumatic birth which had caused a 'fairly extensive' brain haemorrhage. We had to watch and wait to see if he would see/hear/walk/talk etc but he got there on each count in his own inimitable style, huge brown twinkling eyes and the cutest smile! Well, the speech was very slow, but he could say a few words, although at times it seemed as if I had to yell to get him to hear me…

I felt the luckiest girl in the world. I had Oliver who had escaped major health problems and a cutie of a baby in Samuel who was 16 weeks old.

Oliver had had a few bad chest infections and on a regular visit to our GP, I was told he had a heart murmur. Our paediatrician who was doing regular development checks would be informed, so he could check it out.

So convinced I was, that this was all going to turn out fine, I announced to Bob, Oliver's Daddy: "He's only gone and got himself a heart murmur…"
Bob's response was one of complete worry, to which I responded: "He'll be FINE!"

The Paediatrician seemed to want to do a scan, as the heart murmur was 'longer and louder' than he had expected it, had it been caused by his chest infection and so he booked us in for a scan the following week and would do urine tests 'for screening' in the meantime. Tuesday 23rd April 2002, we took Oliver for his scan. It seemed Oliver had a couple of valves which had thickened. This sounded a bit worrying, especially when the sonographer apologised it wasn't better news. Back in to Dr Evans we went and we discussed how these valves had become thickened, as Oliver's heart had been fine at birth, he had had so many tests while he was in the Special Care Baby Unit. We were reminded of the urine tests from the week before. They were testing for something called Mucopolysaccharidosis (MPS). The results would be another couple of days.

I asked for 'that word' to be written down. My Mum and Dad would only ask me what MPS stood for, and I hadn't a hope of remembering! Dr Evans wrote it down after I assured him we wouldn't look it up on the internet. He didn't want us worrying ourselves unnecessarily. Before we left, Bob asked a question.
'Would it alter life expectancy?'
'In severe cases, yes,' came the reply. Words spoken so gently, but which ripped through my heart and as we left I thanked him, I think I even tried to smile, trying to be terribly grown up, as the silent tears trickled down my cheeks and we left for home, still unaware of the horrors that awaited us.

At home, Bob snatched the little piece of paper from me with 'that word' on. I reminded him that we weren't to look it up. Off he went, regardless. Back from the computer some time later, he appeared in tears. There were different 'MPS diseases' but he had clicked on one called 'Hunter Syndrome'. Oliver might live until he was eight, but would gradually become more crippled and would lose all his skills. At best he would make his 30's but would be in a bad way. I asked him to go back to the computer and see if he could find a better one.

Everyone remembers, however blurred the experience was, the day they found out the heartbreaking truth about their child's future.

I spent the next few minutes hysterically trying to phone people. My sister was at work but I spoke to her. Mum and Dad were at golf until later. Golf? What were they doing at GOLF? Surely when there is a crisis, everyone is supposed to be just where you want them to be? I phoned my friend Vicky and ended up in her lounge, crying uncontrollably as we looked at Oliver trying to work out how this could all be happening.

As with all problems, I met Bob at Mum and Dad's later, with Estelle, my sister. We discussed the day's findings and clung to the possibility that this hadn't yet been confirmed. I had an unpopular thought to share. A friend had seen a photo of Samuel recently and noticed he had the same little 'dent' in his chest that Oliver had. My thinking was that if Oliver had this terrible disease, then Samuel would have it too. Nobody wanted to believe it, but surely if we prepared ourselves for the worst, we wouldn't have any nasty surprises?

The following day, Bob spoke to Dr Evans and asked if he was testing 'as a precaution, or as a suspicion'. When I heard the answer had been 'a suspicion' I knew the result would be positive. Dr Evans was a clever man, and if HE thought Oliver had it, then he did. I took myself

down to my church after work and, unable to sit and quietly pray, I knelt down where I had taken my vows nearly five years before and sobbed as I begged the Good Lord not to take my children away from me.

When the phone rang the following morning and Dr Evans asked if I would like the results over the phone or if we'd like to go in to discuss them, I bravely said over the phone was fine. He wasn't going to tell me anything I didn't already know. My dear friend, Vicky and her husband Ian had come round as they knew it was 'results day'. They took Oliver and Samuel into the garden as I spoke to Dr Evans who was trying to let me know all was not lost. I held the tears at bay, just like a 'real grown up' until I came off the phone when they appeared once more in free fall. This was it then.

As the weeks passed, Oliver was diagnosed with Hunter's, the same disease Bob had clicked on, quite by chance. Samuel who was only 19 weeks old, was left alone for a few weeks before he was diagnosed, and the last shred of hope was torn out of my heart.

Great Ormond Street had filled us in with the full story and possible outcomes of the Hunter scenario and let's face it, none of them were good. The doctors didn't like to predict if a child was going to be 'severe' or 'mild' although generally children diagnosed very young were usually severe. What made us feel a bit better was the fact the metabolic doctor said it was lucky Oliver was under Dr Evans' care, as the condition might have been missed until later had he been under the care of another doctor.

There was a ray of light in that a trial had begun its early stages to replace missing enzyme which caused the 'Hunter' symptoms. Maybe they could hurry this along, although it seemed it would be a good two years before anything came available to the boys.

The years ticked on and Oliver and Samuel started having a few operations; tonsils and adenoids out, grommets in. Oliver had development delay but had grommets inserted only a few months before he turned 4, with hearing aids following a month later. A week after his grommets were put in, he asked his first question. Previously it had been phrases like 'Mummy juice'. Now it was 'Radad (Grandad) what you doing?' A few months later a carpal tunnel release operation followed. Poor Samuel had three operations in two months, with Oliver having one in between and after watching Samuel go under a general anaesthetic for the third time and not crying, I worried I was getting hardened to it all, and chastised myself for being a bad mother!

As Oliver started the Special Needs Unit at his local mainstream school, Great Ormond Street offered me the cherished words 'he's not severe'. Now here was a good reason to cry. How I wished everyone could be so lucky. Yes, lucky. I count myself very lucky to be in that situation.

The clinical trials for the Enzyme Replacement Therapy had gone extremely well, although for families waiting to get at this drug, EXTREMELY SLOWLY! However, although it seemed the goalposts of time were shifted every time I thought we were nearly there, finally Elaprase was licenced in January 2007 and funded thanks to NSCAG from April 2007.

Oliver and Samuel had visited 'Dr Ed' up in Manchester who had made the difficult decision about a portacath very easy for us. With their veins, the boys wouldn't get weekly infusions without one! That was settled then and in March they had their ports put in, ready for D-Day two weeks later!

I had tried to prepare Oliver and Samuel for their infusions by letting them know they would be having a special 'magic medicine' which would help them grow, and run faster. It was difficult as they hadn't really had any problems they were aware of. They knew they had had operations and Oliver was taking heart medicine but there wasn't anything glaringly obvious which needed treating. They both love Calpol, but on hearing this magic medicine involved needles, they were not at all impressed!

Thanks to Rosa, the play specialist at Great Ormond Street and Bob the teddy bear who also has a port, the boys had needle therapy to prepare themselves for their weekly infusions. This seemed to go well, although the fear was still there.

On Thursday 5th April 2008 we made our first trip up to Great Ormond Street for an ERT infusion. We were all nervous, but I had got the boys Game Boys for the train journey and that took their mind off things as they excitedly played with their new gadgets.

Once on the ward, they had their magic cream put over their ports and a sticky plaster to keep it in place. Over the coming weeks it became clear that taking the plaster off was the worst part of the whole infusion! Oliver and Samuel hated this bit although they weren't too impressed with the needle going in, either!

However, once 'plugged in' to their pumps, the next few hours were wiled away moving from the Nintendo

Oliver and Samuel's Story...

GameCube, to the Playstation and onto the X Box, while the little enzyme of life slowly dripped into their bodies, giving us all hope for the future.

Lunch was served with a good choice of children's favourites so in a way, it was a children's paradise! There were also two other little boys, George, 3, and James, 4, who were having enzyme too. All four boys had a bond and they took to each other immediately. After infusions had finished, they would race around Kingfisher Ward making lots of noise and who could blame them? The day always seemed to finish with a game of football and Rosa would watch them with a smile. Happy children in hospital, the way it should be. Job done.

After eight weeks it was time to say goodbye to Kingfisher Ward and go our separate ways for infusions at our local hospitals. Lots of photos were taken of the 'four Amigos' and lots of tears shed by me. I hate goodbyes. The nurses had helped so much and as for Rosa, she was an absolute star. And George and James, I was going to really miss these two special boys. I left in a right state!

The following week we arrived at The Royal Surrey County Hospital at Guildford. Oliver had been admitted a few times over the years for his chest infections and it was good to see some of the nurses who recognised him. I soon settled into the pattern of picking the boys up from school at lunchtime putting their cream on and whisking them to hospital for the afternoon. The choice of dinners was limited, and the chips were nowhere as good as Great Ormond Street's, but that really wasn't a big complaint! We were so much closer to home and the boys were only missing half a school day each week. More to the point, no train or taxi fares to pay each week!

Weeks passed and the boys were definitely getting braver with the needle and even the sticky plaster. This was progress!

More importantly, people were starting to comment on the boys' tummies which were shrinking, their hair and skin becoming softer and their facial features seemed to be changing too. This stuff really worked! Yes, okay, we knew it worked from the trials, but there is NOTHING like seeing it work on your own children.

After eight weeks at Guildford, we had everything in place for home infusions. Now this was scary. There's nothing like already being in hospital just in case something goes wrong! I had been told that I would love the freedom that came from home infusions. I probably would, but I wasn't convinced about the safety until a nurse from Guildford's Children's ward admitted that they hadn't seen a full anaphylaxis and therefore would probably panic a bit too! Home it was then!

Two big boxes of ancillaries had been delivered and a mini fridge to put the drugs and Ametop 'magic' cream. The drugs deliveries were to be made each month and the fridge temperature monitored regularly. The fridge was most definitely NOT to be emptied in order to chill beer in the summer! It had only been a fleeting thought…

Lynn, our infusion nurse started coming each week and the boys grew to really like her, and not just because she quite often brought chocolate! They trusted her and were getting really relaxed about the needles and the plaster too. After a few months Lynn left Careology to work at Great Ormond Street, but was replaced by Caroline, who we had met up at the Willink in Manchester, when Oliver had taken part in a two year Natural History Study. The boys took to her straight away and treatment continued without a hitch.

I was beginning to get quite intrigued by the big boxes of tubes, needles etc and wanted to understand which bit went where. I started increasing the speed of the pumps carefully instructed by Caroline. One evening before infusion day, I went into the boxes and got everything out I thought would be needed for the following day. Caroline was impressed and thought I was ready for training. I had always said I wanted to learn how to do the infusions, but it was all too easy to just get the dinner on!

On 13th December, I inserted the boys' needles for the first time. Oliver's took two goes but Samuel's went in first time! For the last 5 and a half years I had been helpless in doing anything for my two boys, but now I was actively helping them to stay as healthy as they possibly could. And boy, did that make me feel better.

Within a couple of months, I was signed off, independent in treating my children. I was almost like a proper nurse! I look forward to every Tuesday when I prepare everything ready for the infusions. There is SO much to remember, so many bits and pieces and everything has to be kept so sterile but I'm on a roll.

I had trouble accepting that I had passed Hunter's on to my children, however unwittingly, but it makes me feel good to know I can help them get better. Me. Their Mummy. The one person who SHOULD be able to make it all better. There is so much hope for the future, and although I am very proud of my achievements as a 'nurse', I will be forever grateful to the clever scientists who made the drug, to NSGAG for funding it, Shire for marketing it, and to the MPS Society who have battled since 1982 to make it all happen. Thank you from the bottom of my heart. **Claire Stevens**

Lisa's Holiday

Lisa who suffers from MPS III, Sanfilippo Disease, with her parents Rosemary and Harry from Kent went on holiday in July this year to Butlins in Bognor. They were joined by Tracey, Jan and Jim from Cambridge. Both families had a brilliant time and even the weather was good!

Tracey and Lisa enjoyed themselves in the fun atmosphere, it was nice for the families to get together and they look forward to meeting up again soon.
Rosemary, Harry and Lisa Nurse

Janet and Jim Orrin and Tracey Guy (MPS III) and Rosemary, Harry and Lisa (MPS III) Nurse at Butlins July 2008

If you would like help, guidance or information from the MPS Society's advocacy team please do phone us on **0845 389 9901** or email **advocacy@mpssociety.co.uk**

Your news and views

We are always pleased to receive news, information, letters, stories and poems from all our readers, especially our members. We welcome letters on any subject and your views and comments would be very welcome or perhaps you would like to share some information? Email us at **newsletter@mpssociety.co.uk**

25th Anniversary celebrations at the Palace of Westminster

My name is Marina, grandma to twins Francesca and Josephine, whose parents are Julie and Chris. The twins suffer from Sanfilippo disease. Sadly, Francesca died in February 2007 aged fifteen. We all live in Bristol and I run a shop selling donated goods to raise funds for research into Sanfilippo disease.

Imagine my surprise, when returning one evening from a long day at the shop, I discovered amongst my post an invitation displaying a portcullis crest from the MP Jeremy Hunt on behalf of the MPS Society, requesting the pleasure of my company for Afternoon Tea to mark the end of the 25th anniversary year of the Society on Wednesday 14 May 2008 at the Palace of Westminster.

Immediately I rang my daughter and found to my relief that they had also been invited. It was all very exciting but what were we going to wear?

Julie and I decided on trouser suits, as we would be travelling on public transport. Then a few days before the event there was a mini heat wave and we were convinced that our outfits would be too hot. However, on the day it was cooler and we were glad of our jackets.

The day started really well, bright and breezy, but not too hot. The train was on time and we arrived at Paddington where we had a snack before taking the tube to Westminster. We decided to walk across Westminster Bridge to get a better look at the London Eye and Big Ben and to view our venue, the Terrace Pavilion.

Going into the fenced grounds of the Palace of Westminster and looking out and seeing so many people outside looking in made us feel really special.

We had to wait in line to clear security and could observe the police presence, some of them were heavily armed. Our bags were checked and there was a body search (I didn't mind because the policeman was good looking and at my age, 73, that can't be bad). Now we could actually enter the Palace of Westminster and found ourselves in the Great Hall, a vast cavern of dimly lit space with an imposing flight of stone steps and magnificent stain glass window at the opposite end. This Hall is where the Queen Mother lay in state in 2002.

At the top of the steps we passed through an impressive arched doorway and into a passageway which led to the ornate chamber, often seen on TV, between the House of Commons and House of Lords. This teemed with people and atmosphere. There's even a Post Office so we took the opportunity of sending some letters that would be franked with the House of Commons mark.

We descended to a lower level which turned out to have a number of rooms where MPs could meet and dine with visitors, before arriving at the doorway to the terrace. Here we were greeted by Christine and the MPS team who distributed ID badges and assorted information. It was nice to finally meet Gina Page, somebody I have spoken to many times on the phone.

The Pavilion Terrace is a large room opening onto a section of terrace overlooking the Thames with a wonderful view of the London Eye beyond Westminster Bridge.

The space quickly filled as people arrived at the appointed time of 4pm. Wine and soft drinks were available and it was not long before Jeremy Hunt MP was welcoming us all and spoke about his involvement with the MPS Society before handing over to other speakers who outlined the Society's work and research achievements over the last twenty five years and ongoing goals. It was then time to cut the cake, a task undertaken by Sarah Long and Myles Broughton with a little help from Jeremy Hunt. Then on with the conversation, canapes, cake and drinks! Chairs were hard to come by but I was lucky enough to find one and shared some special time with a lovely couple exchanging stories about our children's escapades. Hope you enjoyed it as much as I did!

As the party came to a close we slowly made our way back through the corridors towards the Great Hall taking the opportunity to browse in the gift shop and purchase some souvenirs.

It was a wonderful day and I feel so pleased to have been invited to join in the celebrations and to have sampled the hustle and bustle of London for a while. Best wishes to all of you and here's to the next twenty five years.

PALACE OF WESTMINSTER

Marina Foster and Julie Kembrey on the Terrace Pavilion of the Palace of Westminster

Marina Foster and Chris Kembrey in the Great Hall of the Palace of Westminster

Editor's Note: Apologies to Dr Ed Wraith and Dr Fiona Stewart for an error in the last edition of the MPS magazine. It was Dr Fiona Stewart who spoke at the Society's 25th Anniversary celebrations, not Dr Ed Wraith.

10th International Symposium
Vancouver, Canada

I couldn't believe how many of us there were, all gathered in Terminal 5 not only booked in, with tickets, but on time, on schedule and everything seemed to be going to plan!

It was getting all exciting as we headed through security and had some time to get a snack or do some shopping. There were 49 of us in total mainly MPS families, some wonderful MPS volunteers and some MPS staff to help make sure everything ran smoothly.

The British Airways Team was fantastic and let us board all together and were helpful with the buggies and wheelchairs. Once on board we were all sat together in a block booking across the plane! I think we were all well behaved and the flight seemed to go quickly enough! Nearly 10 hours later we landed and this is where the tiredness seemed to kick in.

Ben Lavery ran outside to find the bus for me and we were set about boarding the coach and finally arriving at the Sheraton Wall Centre Hotel where the conference was set to begin on Thursday after lunch.

We had time to check everyone in to the hotel and then all gather for a team meeting in the North Tower Lobby. We went over the plan for the next morning's outing and what was expected of everyone for the trip! The volunteers got to meet the families that they would be caring for during the childcare programme. I got to boss the Trustees around for two seconds as I explained their roles on the MPS Stand and if anyone had any questions this was the moment to ask them! Before I knew it I hit a brick wall and was ready to go to bed, shame it was only 5pm Canada time. I headed to my room to catch up with some sleep before another very busy day on the coach and being in charge of the group! Room service came in handy this night although I could have shared it with about four other people.

The following morning it was breakfast and then heading for the coach with a lovely bus driver who was friendly and unfazed by our large wheelchairs and buggies. He was great at organising everyone and best of all I got to speak on the loud speaker to the whole bus (I had hoped I would be allowed to do this)!

So, first stop was Capilano Suspension Bridge. I knew this would get busy by about 11.30am so we arrived at 9.30am and got in there before any of the other tourists! I am pleased to say that I didn't cross the bridge this time although everyone that did had a great time! There was an amazing gift shop there to enjoy and some shopping for family and friends back home happened. Alan Milligan (from the Royal Free Hospital) who was volunteering for us brought a brilliant faux racoon hat! Another coach checklist to mark to make sure everyone was on board and we set off to Grouse Mountain where the most amazing buffet lunch I have ever had was served for us all! Up Grouse Mountain you can see the most incredible views of Vancouver City and bay area, it was just beautiful!

We had time to explore the mountain before and after lunch with a brilliant Lumber Jack Show and some grizzly bears to visit (that looked a bit miserable) and the strangest thing of all is there was real snow on the mountain still! I couldn't believe it! I was walking on real snow... so strange coming from Australia!

By the time we got down the cable car journey and loaded back on to the coach we were running out of time to do much more so we had a drive through Stanley Park which was lovely with over 200 year old pine trees. The bus driver even let us jump out quickly to take a photo of a famous bridge!

We arrived back at the hotel for 5pm. Most of us were shattered apart from the volunteers that thought that was the perfect time to head down town for dinner! It was free time for all and an early night for a lot of us as we recovered from the jet lag! The MPS staff team entered a meeting which lasted until midnight which I'm sure cured my jet lag as I worked through some hysterical moments with Christine, Sue, Barbara Wedehase (USA MPS Society) and Laurie (also from USA MPS Society). Good memories those were.

The following morning (thank goodness) there was time for a small lie in and many of the families I'm sure appreciated this.

There was a special announcement while we were away in Vancouver that involved one of our MPS Society members and Mum to Will Brodie MPS I, Laura Brodie! Laura had been nominated for a Teaching Award by her fellow teachers at Allen's Croft Children's Centre where she is head teacher. This was a lovely evening with much celebration for Laura Brodie and her family and friends.

The conference programme started after lunch and the children were registered into the Childcare programme with the MPS UK Volunteers. The children ventured out to the Aquarium and Grouse Mountain and in addition enjoyed the beautiful Stanley park with Water Park and pools! It was stunningly hot weather the whole week so it was lovely to be able to have the children running around outside.

VANCOUVER

The conference programme was jam-packed with lots of cutting edge talks covering a range of the MPS diseases. Gordon and Anne Hill who are members of the UK MPS Society were speakers at the International Symposium and spoke brilliantly on palliative care. It was one of the first times that parents, individuals and patients were able to take part in electronic voting specifically on new born screening.

There was a special part of the conference that was for the bereaved families where they were able to release butterflies after a short memorial service was held! It was a special moment for many families as they gathered outside to remember those that have passed away from MPS conditions.

The Gala Dinner was amazing. There must have been 400 plus of us in a huge room seated at tables of 10. We ate lovely food including a chocolate torte for pudding!

We also played head and tails with the different professionals giving us questions that we had to answer with either heads (hands on heads) or tail (hands on bottoms) to answer the question. This was rather exciting as James Stewart (MPS I UK Member) and Sophie Thomas were left in right to the end of the game. James Stewart went home with a framed original artwork which had been especially designed for the conference in Vancouver as the prize for the last one standing in the head and tails game.

By the time it came to head home we finished the week off with a farewell lunch in the hotel for our group and boarded the bus bound for the International Vancouver Airport. It wasn't long before we were on the plane headed home… I slept the entire journey, I heard that others didn't sleep so well but I'm sure it didn't help with the jet lag. Again the British Airways staff were fantastic and we arrived safely and on time back at Heathrow. Another International Conference done and dusted, the next one set for 2010 in Adelaide, South Australia, the dates are Wednesday 23rd - Sunday 27th June. **Miriam Blowers**

EVENTS

Mrs Laura Brodie
Teaching Awards 2008 Winner
The NCSL Award for Headteacher of the Year in a Primary School in West Midlands
Allens Croft Childrens' Centre, Birmingham

Laura Brodie has dedicated the past four years working on her vision for the brand new £5.2 million Allens Croft Childrens' Centre. From humble beginnings in a small, tatty hut, she has overseen the creation of a multi-agency, state-of-the-art hub for the community, providing day-to-day support, education and care. It offers a seamless transition between day care, nursery education, health services and the neighbouring primary school.

She understands the challenging circumstances in which families live, and the impact this can have on aspirations. She ensures that services do not begin and end at the nursery doors, so an outreach programme works with them to support their needs. She acquires extra funding for the Centre by giving lectures and sharing good practice.

Children at Allen's Croft are exposed to art and creativity in an amazing way, allowing them to blossom. Laura has been inspired by the work done with pre-school children in Reggio Emilia, in Italy, and believes children learn best through play and hands-on experiences. Ann Mcnutt, the school Senco, who nominated her, said the work done at Allen's Croft "is some of the most fantastic work I have ever seen undertaken with children of that age."

Laura makes her staff feel valued and always turns negatives into positives. She is forward thinking, leads by example and has developed a successful staffing structure that enables information to be filtered down to all individuals. She regularly updates the team on new innovations, investigations and research, actively encouraging them to further their own interests and professional development.

Laura is aware families live on tight budgets so she recently organised a free trip for 80 families - many of whom had never been to the theatre before - to see Peter Pan, at the Birmingham Rep, offering an opportunity for Lifelong Family Learning.

Judges described the work done by Laura as "something totally unique and rare". "Clearly she has succeeded in the toughest challenge possible - achieving effective cross-professional collaboration," they said.

Article taken from The Teaching Awards
www.teachingawards.com

The MPS Society was very proud to announce that Laura Brodie, Mum of Will (MPS II), won The NCSL Award for Headteacher of the Year in a Primary School in West Midlands. Miriam Blowers tells how the MPS Society was asked to present a copy of the award to Laura whilst in Vancouver Canada for the International Symposium.

I had secretly received an email and phone call from Lisa at Allen Croft Childrens' Centre saying that they had nominated Laura and would we announce the nomination while we were in Canada as Laura would be missing out on the ceremony! I thought this sounded lovely and agreed to it straight away. I then less then a week later received a highly confidential phone call from the Teaching Awards office letting me know that Laura in fact had won the regions award and although she would be missing out on the ceremony which was taking place while we were in Vancouver, would I mind taking a copy of the award to Vancouver to present to her in person. I felt very honoured to have such inside information and packed a rather large heavy award to transport to Vancouver.

Below is what her colleague sent to me to read out during the presentation that took place in Vancouver: 'Laura is held in very high regards in the education world and her achievements are like no other. Our Centre is one of the first in Birmingham to link Health and Education together.

Laura has been working towards the completion of our £5.2 million centre for the last 8 to 10 years which opened in April 2008. She developed this new centre to be a multi-agency hub for the community and provides support, education and care. All of this was done without neglecting her role as a Head Teacher. Alongside this she also had an OFSTED inspection (while in a portacabin!) and achieved 'outstanding' in all areas. Not only has she managed to do all of this with optimism but she is also a wife to Dave and the mother of two wonderful children, Lily and Will, who continue to be her motivation and inspiration.'

The MPS Society wishes to send their congratulations to Laura and her school Allens Croft Childrens' Centre.

2008 MPS Conference, Vancouver:

A View from the Leonard Family

We were very pleased to be able to arrange with work and Chris's school to take the time in June to travel to Vancouver and meet so many old friends at the MPS conference.

The conference was very well attended. Two groups of lectures: Scientific and general lectures ran in adjacent rooms and family delegates invited to attend what ever they chose. Most of the topics were focused on MPS, but it was interesting that some of the invited lecturers talked on medical topics, such as arthritic changes, bone/cartilage problems or damaged brain cells, but from the perspective of medical teams treating or researching disorders such as Alzheimer's. We found the talk given by the vet that ran the menagerie of animal models very interesting. And yes! Those animals do have a good life!

Anne and Gordon Hill gave a very moving talk on their daughter, her life and their care. We presented a poster on 'Parental Care of two children with Fucosidosis', this generated plenty of takers for the handout and interest in the bone marrow transplant undertaken by Christopher 16 years ago!

The UK was well represented by MPS staff, families and volunteers, thank goodness! The conference plan for the children and young people appeared to be overwhelmed, but luckily UK volunteers instantly improvised some excellent sessions and outings for the UK children and teenagers.

Socially we had a great time! All of us Brits (apart from those running the MPS stall all day) were taken on a coach to visit Grouse Mountain, ride on the ski lift gondolas & visit a suspension bridge. And we had a great meal in the restaurant at the top of Grouse Mountain. The gala dinner was enjoyable, possibly our hosts had watched too many Awards ceremonies? It was very pleasing when John Hopwood was given a special award for his life-long work and encouragement of research collaboration. For our family, John Hopwood is very significant because his team in Adelaide carried out the early research on Fucosidosis.

Vancouver city centre is very modern. Glamorous glass concrete and steel, but very easy to walk around and reach the waterfront, park or shops. Quite by chance we were there on a certain Friday each month when all the bicycle users take their bikes onto the centre streets and literally stop the traffic. There were droves of them and many in fancy dress or very camp! The occupants of the stationery cars were 'not happy' as Christopher would say!

All in all we had a great time and would like to thank all the MPS Staff, Trustees & Volunteers.
The Leonard Family - Jean, Paul and Chris (Fucosidosis)

Chris in Vancouver with MPS volunteer David Murphy

Childhood Wood Remembrance Day

On Sunday 13th July 2008 the Society held its annual Remembrance Day at the Childhood Wood. Nine families attended the day to remember the children and adults they had sadly lost to an MPS or related disease.

It was a beautiful day and it was one of those rare occasions this summer where the sun was shining.

We planned things slightly differently this year and we arranged a Carvery lunch in the picturesque grounds of Rufford Country Park before taking a much needed walk down to the wood.

The Remembrance celebrations went beautifully and families released a balloon in Remembrances of the loved ones they had lost. Even the balloon stuck in the tree gradually made its flight into the sky with the perseverance of the family and their skills in stick throwing!

It was a lovely day and a privilege to have been able to be a part of it. **Sophie Thomas**

Remembering Kim

Another Childhood Wood Remembrance Day has gone by and unfortunately we were not able to attend it. Sadly it is now six years since we lost Kim and I am sad that we have not yet been to any of the Remembrance Days. Living in Surrey it is quite a distance for us. But we know that there are many who do go and that includes some from the MPS Society who go on behalf of those who can't attend.

Chris Murphy was there on our behalf. Because of the six years of passing, I asked for six balloons to be released which she was able to do. I know in future, releasing balloons may have to stop but at least this year it was O.K and I am grateful to Chris for releasing them.

Chris and I spoke on the phone prior to and after the Remembrance Day. We talked about Kim. About what she was like and particularly that Kim was diagnosed in the 1970s when there were only 12 known cases of MPS. Of course, Kim had loads of medical problems as the years went on and like many with MPS, she started out normal but things altered as years went by.

Of course, it would have been lovely if she had never had MPS, but she did and we learned to live with it and to have as normal a family life as possible. Kim had a younger brother and sister.

We never knew what Kim felt emotionally about life nor if she was even aware of her family and surroundings, but she seemed happy. As she was not a disruptive child we could take her anywhere and did. Luckily for us she was a 'Peter Pan' and never really grew larger than an 8 year old child, although she made it to 30. Great for me as I had to carry her most of the time.

Besides the love for Kim that I had and still have, I can only think of her in terms of having MPS. I can't know what she would have been like had she grown to a normal adult, like her brother who is married with three children and a sister who is nearly 24.

The oldest granddaughter (10 years old) Tiegan-Marie (Marie was given as it was Kim's middle name) knew Kim and was only four when Kim died but she loved Kim and still remembers her fondly. The other two younger ones were well aware of Kim by name and photo. Also because all the regular toys are in Kim's bedroom. Kim's own toys are respected and kept carefully, not damaged.

Having grandchildren is great. It gives me the chance to do things with them. Things I could not do with Kim.

Now with six years passing, I don't remember all the hard times just that Kim was with us, but I do wonder what she would have been like without MPS.
Marilyn Eggleton

Center Parcs Sibling Weekend

How lucky we were with the weather, it's hard to believe we had a very hot, sunny and DRY four days in Centre Parcs, Longleat, 25 - 28 July 2008.

Thanks to some great travelling organisation as children came from all over the country we somehow all managed to arrive at Center Parcs around the same time.

Once settled in to our villas, the children were still full of energy, it's hard to believe that some had been travelling for most of the day. Everyone had a hearty appetite and the pizzas and Danish pastries went down a storm.

After a good night's sleep we were ready to enjoy the weekend ahead. The children tried lots of different activities, some they were familiar with and some very new. Those of us who went to the bowling alley were surprised when we had to go behind the counter and find our own shoes! Everyone tall and small had great fun there and became experts by the end of the game.

Our challenge of the day was to get to our villas for lunch and then back to the swimming pool ready for the afternoon. The pool area was really nice, very tropical with lots of palm trees. All the children enjoyed the swimming, and as we had previously found, the time goes so quickly when having fun!

Party in the Park was a fabulous evening with tables set out with huge decorations of balloons. The food was excellent and the entertainment was enjoyed by all ages with a magician, live music and then the disco. This was a good excuse for most of the children and volunteers to hit the dance floor and have a good 'boogie'. But then the tiredness of the day kicked in and it was time to embark on the little train back to our villas.

I was beginning to realise that other visitors using the train were travelling at different times to us. It was beginning to sound like a quiz: how do you fit 22 children and 16 adults on a small train? Answers on a post card! The good news was we did and by the end of the four days we had the commuting down to a fine art!

Sunday morning we were still going strong and some of us were in the Activity Centre while others were building a raft by the Lake. It was very hot, so suntan lotion was being slapped on by everyone - seems a lifetime ago now with all the rain we have had since then.

After lunch it was back to the pool. By now we had this down to a fine art, managing towels, locker keys, spare clothes, bags and all getting out of the pool at the same time!

We even managed to celebrate Ryan's birthday during the weekend.

By 9am Monday morning we were washed, dressed, packed up and ready to move to Longleat Safari Park. With cars and the mini bus in convoy, it was "wagon's roll". We had a fabulous morning in the park where some of us managed to visit the maze. This was a real adventure to say the least, as some of the group were successful enough to get to the middle, most of us got lost and would probably still be in there now if we hadn't received directions from those who made it to the centre tower! They were giggling at the rest of us all bumbling about getting totally lost, but their directions got us there eventually. But there were the few who cheated, no names mentioned but you know who you are!

With visits to the Mine and Pet's Corner there was something there for everyone and still the weather held out.

As the weekend was drawing to an end, we had time for a picnic lunch in Longleat Park and say our goodbyes. It was an emotional time for most, with tears and laughter, but everyone agreed what a fab time we had together. I am sure everyone slept well that night.

From my own personal reflection I will always remember the laughter as we all took up our places on the train, it was like an MPS takeover, although the children were so well behaved and polite to other visitors.

With all the rain we have had since, it's hard to believe how lucky we were with the weather that weekend.
Linda Warner

SIBLING WEEKEND

"The sibling weekend was a really enjoyble experience, feeling like an older brother to the siblings was just one of the highlights of the weekend and I can't wait for the next trip. The activities were great and the organisation was excellent and I would just like to thank MPS for the opportunity."
Vishal Ram

EVENTS

"Thank you for taking us to Center Parcs in July.

I really enjoyed high ropes, even though it was a bit scary!

It was funny walking to raft building when Ian tripped over the fence! Also I really liked raft building because ours kept on breaking, so we got to swim with the fish!

I had fun at the swimming pool because the rapids were really fast.

The Latin night was really great because there was lots of entertainment. The magic man and the dancers were cool.

My favourite food was the lasagne. It was delicious.

At Longleat I enjoyed Old Joes Mine because I really liked the bats.

Thanks to everybody that looked after us for making it such a great weekend." ***Charlotte Home***

Dear MPS

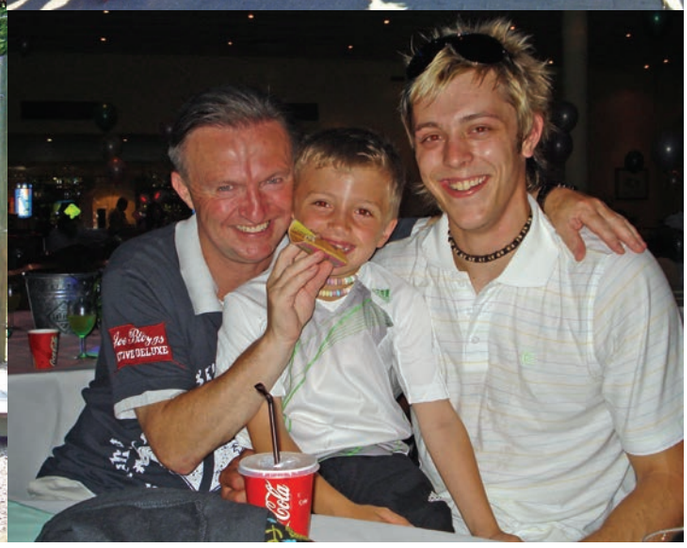
Thank you so very much for taking us to Center Parcs. Freya and I had a lovely time and made a lot of new friends with siblings with Sanfilippo and other MPS diseases.

We had a lot of fun with both the volunteers and other siblings especially in the water.

Freya and I loved doing the rapids, high ropes, challenge course and the archery.

Thank you to the MPS Society and the volunteers for taking us.

Mollie and Freya Chisling



Volunteers needed for research

Do you live in the North West of England?

Would you like to help improve services for families with genetic diseases or conditions?

Your views about a new questionnaire to measure outcomes from clinical genetics services.

I am carrying out research to develop ways of measuring how people with genetic diseases or conditions may benefit from seeing a geneticist / genetic counsellor or attending a genetics clinic. As part of this work, I am developing a questionnaire that will examine patients' thoughts, feelings and attitudes about the family disease or condition.

I am looking for people from families with genetic diseases or conditions to help me with this. It would mean coming to a meeting that will last for up to 2 hours to discuss the wording of the questionnaire, and to make sure that it addresses the issues that are really important to people from families with a genetic disease or condition. To help with this, you **do not** need to have been to a genetics clinic yourself.

There has been very little research in this area and this meeting provides an opportunity for **you** to help design this questionnaire to ensure that it is easy to use and relevant to families affected by genetic diseases or conditions. The questionnaire will be used to help improve services for affected families.

The research is funded by the Medical Research Council, and has ethical approval from the University of Manchester.

IF YOU WOULD LIKE TO KNOW SOME MORE ABOUT THIS, WITH NO COMMITMENT TO PARTICIPATE UNLESS YOU LATER DECIDE THAT YOU WOULD LIKE TO, PLEASE CONTACT ME.

Marion McAllister PhD, Genetic Counsellor and MRC Research Fellow,
Tel: 0161 276 8979 E-mail: marion.mcallister@manchester.ac.uk
The Nowgen Centre, 29 Grafton Street, Manchester M13 9WU

http://www.nowgen.org.uk/stories/232-dr_marion_mcallister
http://www.medicine.manchester.ac.uk/staff/151821

Expert Meeting on MPS IV, Morquio Disease

It was the first ever meeting for professionals, patients and their families on Morquio Disease. There was certainly an excitement for the families as to what was to be shared for the future treatments of Morquio Disease.

Over 180 adults, 45 children and 24 volunteers attended the Morquio Conference, 29 - 30 August 2008. There was also an International Network Meeting that took place following the Morquio Conference at which 13 different MPS Societies were represented.

Once again thanks to the MPS childcare volunteers the children that attended the conference with their families had lots to look forward to as they arrived on Friday 29th August. The day was set aside starting with lunch for everyone and then the parents, professionals and speakers headed to the conference room whilst the children paired up with their volunteers headed to 'Little Miss Makeover' for the girls and 15 or more animals to hold and learn about for the boys! It was a great day for the volunteers and children alike with an excitement in the air as John (partiesforanimals.co.uk) shared his knowledge of the animals and Gayle (mymakeoverparty.co.uk) pampered the girls with nail painting, hair extensions and a photo shoot to finish with.

The conference programme was full of professionals that gave up their time to come and share their expertise, the speakers presented well with plenty of time for questions and answers for both the professionals and the families present.

The Gala dinner followed where Sarah Long presented the 'Tipping the Lens Project' participants with certificates and Barry Wilson explained in some detail what the project was and is all about. It was great to be

able to acknowledge all the participant's hard work in documenting what life is like and about for them, day to day! The MPS Society also thanked Sarah Long for all her hard work and presented her with an orchid. That evening we all ate a delicious dinner of chicken and vegetables and cherry cheesecake for pudding!

The children and volunteers joined us for the meal and the children were entertained at their tables by some great colouring-in packs and puzzle that Sarah Long provided. Then we all got to enjoy some wonderful Rat Pack music to finish the night off.

It was a lovely evening with the option of an early night if need be for some of us!

The following morning it was an early start for some with volunteers and the children heading off to Ten Pin Bowling until lunch and the conference programme beginning at 9am sharp!

Another full day of conference for the parents finishing with Vivendy and Biomarin presenting their Enzyme Replacement Therapy and how it will translate to the clinic. Many families had waited a long time to hear of some hope for treatments for their Morquio Children.

The conference therefore finished on quite a high with a healthy discussion about where and when things would start to develop regarding Enzyme Replacement Therapy.

The childcare finished on a huge high with 'Mad Science' showing the children and volunteers how periscopes work and learning to make their own, playing with slime and learning how fairy floss is made and being able to sample it! Let's just say there were lots of Bright Blue Tongues!

Thank you to all the speakers that gave of their time to present at the conference, to the families and professionals that attended the conference.

The conferences would be impossible to run unless we had the amazing support of the MPS volunteers so a huge thank you to them for giving their time and energy to care so brilliantly for the children.

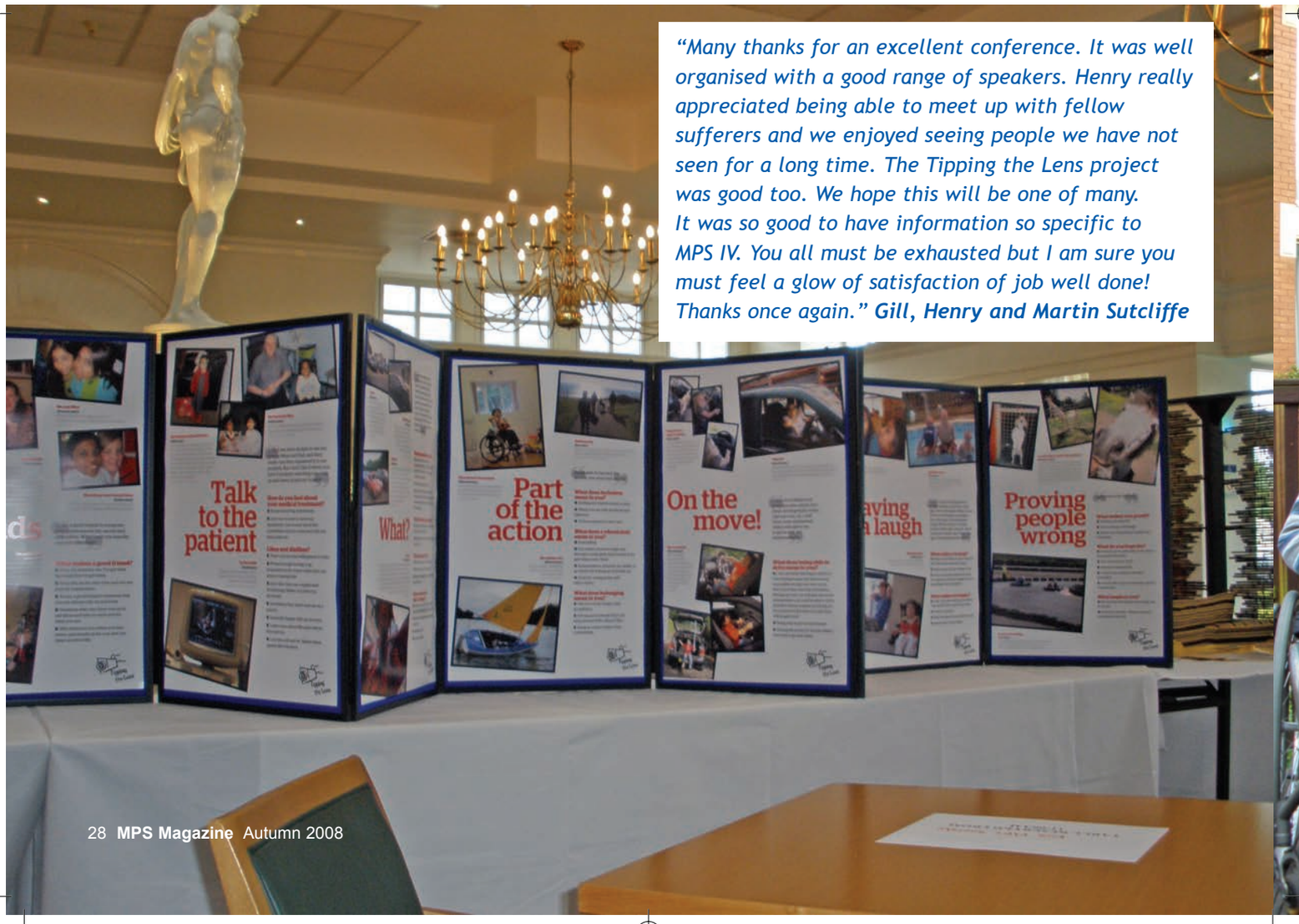
It was a wonderful conference and we are working hard at getting things ready for the next big event next year, the National Conference, 26 - 28 June 2009!
Miriam Blowers

Tipping the Lens

"My Words, My Images, My Views"
At the international Morquio symposium held on 29 August 2008 the Tipping the Lens Project launched with an exhibit which was placed in the Hotel foyer. The display consisted of a series of twelve A1 size posters which used the words and photographs of ten individuals living with Morquio (from a variety of different backgrounds and aged 12-42 years). These were designed to give an insight into what it's really like "living it", how we view ourselves and our world.

What is the Tipping the Lens project?
The Tipping the Lens project (TTL) is designed as a participatory project aiming to tip the lens away from a medical focus e.g. CT, MRI scans and X-rays etc., to focus on the narratives of people living with Morquio's. I was inspired to design the project as I live with Morquio and following a number of conversations with other people also living it. I aim to write this work to contribute to academic social research. During the conference I put together a number of TTL activities. Firstly, the main part was the exhibit in the foyer called "My words, My images, My View", secondly a "dining views" activity which provided the opportunity for professionals and families to define a good quality of life for people living with Morquio; and thirdly some children/young people's activities. (Anyone who was at the conference it would be great if you could please return questionnaires and children/young people activity books as it would be great to see them.) I am currently working with five further individuals on the project. If anyone else living with Morquio wants to be involved, do contact me or Sue Cotterell in the MPS office so we can chat about it! My next stage is to start visiting participants to talk through their work and develop their images and words to ensure I represent them in a way they are happy with. We are currently seeking funding to develop a book.

How were the Exhibit themes chosen?
The twelve themes shown in the posters emerged as the twelve strongest messages when all the materials were reviewed after the ten participants who featured completed their Viewpoint book (which contained a range of questions) and submitted images. All the words



"Many thanks for an excellent conference. It was well organised with a good range of speakers. Henry really appreciated being able to meet up with fellow sufferers and we enjoyed seeing people we have not seen for a long time. The Tipping the Lens project was good too. We hope this will be one of many. It was so good to have information so specific to MPS IV. You all must be exhausted but I am sure you must feel a glow of satisfaction of job well done! Thanks once again." ***Gill, Henry and Martin Sutcliffe***

and images used in the posters were taken directly from participants' responses; those selected were considered representative of a number of other similar comments.

What was in the exhibit?
The Posters contained a number of themes. These ranged from "Me!" a description of themselves e.g. "bubbly and chatty!", "beautiful petit!" Another explored how people themselves define Morquio, "I feel normal, never known anything different" as well as reflecting on their bodies the good and bad bits! The posters also showed the importance of family and friends, of being accepted and supported. Next to get the spotlight were issues relating to health care. This highlighted the importance of being involved alongside likes and dislikes "don't like they use complicated terminology…". Highlighted was what it is like living in a world were you are stared at, called names, and have to deal with other unfortunate situations. This was contrasted with a poster outlining what inclusion and belonging means "when you are fully involved and take part", "…where I feel comfortable". The importance of driving was raised, as participants wanting to, were learning and driving. "Being able to get out and about" independently was stressed as important. The importance of people's faith also came out strongly as did the similarities in their beliefs. The majority in our group had a faith and were either Christian or Muslim. The importance of enjoying life, finding inspirations and ambitions for the future were also captured.

What were the reactions and comments about the exhibit?
"Very poignant. Great images and comments very relevant and real…" A family response
"Excellent idea and project."
"It seems like a fantastic project, and it should be presented to the public at large…"
"Great idea - need more time to look at the photos and stories."
"A good way of creating 'togetherness'" Someone living with Morquio
"Life is more than what we see of our bodies. It is what we feel." A professional
"TTL has been a great project for (our daughter) to do. Helped her express her feelings, fears and also to come to terms with her own condition."

There was a lot of positive feedback about the exhibit from all the conference attendants, including those living it, family members, professionals and even hotel staff! Participants seemed pleased about being part of it and some of their families felt it had been very valuable talking about the issues the project raised. **Sarah Long**

MPS in Turkey

I come from Turkey and have been a member of the MPS Society for the last 24 years since our daughter, Natalie's, diagnosis with MPS III, Sanfilippo Disease. I always had this great desire to help establish a Society or help group in Turkey. Christine has been very instrumental and helpful in my efforts to achieve this goal. We have not formed a Society yet, but a lot has happened and I am in touch with lots of families out there.

The International MPS Symposium in Vancouver was especially exciting and important for me because I was told that two brothers with MPS VI from Turkey would be there with their mother, as well as two Turkish doctors.

I met with them on the second day of the conference during one of the breaks. It was such a busy conference for me that next time I could meet up with the Turkish group again was the gala dinner. We even managed to have a picture taken with Christine Lavery and Adriana

Montano, from Saint Louis University, USA (who happens to be married to a Turkish nuclear physicist who comes from my hometown. (What a small world!)

Both the brothers have started ERT a few months ago, which is fantastic news. I have also been told that they don't need to travel to big medical centres. They are able to have their treatment locally. One of the brothers speaks a little English and goes to university and is away from home.

I spoke to the family on the day of our departure and we exchanged addresses and e-mails. I spoke to them while I was in Turkey in July.

I have also been to the Morquio Conference in August. It was very informative and helpful. Not only did I learn more about this condition and the latest medical developments in this area, I had a MPS IV patient from Turkey contacting me as a result of my chat to an Irish mother who had met someone from Turkey at the Venice Conference. Thanks Mary and Elena McGauran. This is what networking is all about.

If there is anyone out there who knows Turkish people with MPS, please ask them to get in touch with me. Thanks to everyone for their help. **Fer Pidden**

MPS in Brazil

APMPS, Associacao Paulista de Mucopolissacaridoses, was founded on 5 June 2001 is a non-profitable civil entity with social ends, of national extent, that aims to provide the constitutional rights of the carriers of this syndrome, who face a situation of social vulnerability. The main objective of the association is to provide individual and group support regarding the areas of psychology, physiotherapy and social service in order to improve their qualities of life. Not only this support is provided but also family orientation is given, specially related to the

limitations and potentialities of each carrier. This is done so that the family can be supportive in terms of the difficulties.

After discovering that their children were carriers of a rare, harmful syndrome, called mucopolysaccharide, some families decided to create this association. Instead of complaining about that situation, those families joined their energy in order to save their 'special' children. No sooner were the fear and experiences shared, they decided to help each other and other carriers and their families. APMPS was created in this feeling of positive energy and the will to win.

Initially, the association headquarters was in one of the carrier relative's house, where all the infrastructure was 'sponsored' by the family. Later, they thought they were in need of a more appropriate place due to the necessity of helping not only more families from Sao Paulo, but from all over the country. Currently, APMPS's main office is in Campinas – S.P. and there is a branch in Guarulhos – S.P.

Based on the needs of the families, the main objectives of APMPS are: Improve the quality of life of all the carriers; Improve the diagnosis in Brazil; Keep the exchange between carriers, families and associations, nationally and internationally; Raise the Society's awareness for the problem; Provide updated information about the disease for the carriers and families; Establish a relationship with Medicine Universities; Focus on institutional partnership; Encourage partnerships between health services and professionals involved in the prevention and treatment of the disease; Change institutional support from the government; Support conferences, courses and symposiums and other events involving health and diseases; Provide support to the disease carriers and their families giving them all the information necessary; Judicial support and counselling; Encourage education campaigns for the society.

With the help of the ministry of health, APMPS's current main objective is to get the inclusion of the medication approved by ANVISA, specially the high cost ones. We intend to get those medications as fast as possible and perform the registration formal channels of the medications that have not been approved yet.

When a national policy of attention in clinical genetics is adopted in our country, the early diagnosis and the appropriate treatment for these children are going to be easier, offering them a better quality of life. There are medications and treatments, the only thing that is missing is the commitment of the authorities to make things happen. A great many of the Brazilians would be incredibly relieved if they found more people supporting the sacred right of life, warranted by our Constitution.

After some time of experience, we noticed that we needed a closer attendance due to the fact that most of the families ignore their rights as Brazilian citizens and consequently do not get the appropriate treatment. In order to help more families in Brazil and other countries, the alternative we have is to join our strength and power to other assocations.

MPS Brazilian carriers also face problems in obtaining support treatment (pulmonary therapy, speech and physiotherapy, hydrotherapy etc). Health insurance companies refuse to pay for these treatments so we are always in need of checking the rules and rights regarding the disease.

Most of the children do not have financial conditions of having a private treatment so they end up in public places, where there are no professionals in this area. Worried with such lack of information about MPS, the APMPS has been looking for all possible alternatives for making it public for the press, government and all the classes of society. As a result of our approach, some reports and articles in magazines and newspapers were published. We also had some programs on TV and we participated in public auditions in the senate house, we had meetings with some parliament people from Brasilia, including the health minister Dr Jose Gomes Temporal, and the minister of the federal high court, Ellen Gracie and we promoted national conferences in 2003 and 2007 with 280 and 450 participants respectively. We are also promoting meetings with the families to provide up-dated information about new treatments, therapies and medications. Nowadays, we have the help of volunteer professionals to fight our cause.

Unfortunately, the Brazilian government does not have interest in us. We have been fighting for a high cost medication called Laronidase (MPS I) and also for the liberation and registration of medications for MPS II and MPS VI in ANVISA.

Currently, APMPS supports about 120 carrier families. According to our statistics, there are 90 MPS I carriers, 150 carriers of MPS II and 100 carriers of MPS VII and 3 carriers of MPS IV. About 80% of these carriers use the medication through 'compassionate usage' or through temporary restraining orders.

We are far away from having a deserving quality of life. Most of us do not have good conditions of living, transportation, basic food and medication.

We have been working hard to get and keep in touch with other support groups and associations in order to create a net of political and social information, including medical news.

We with the help of volunteer work, we created our site www.apmps.org.br where we created a channel of communication between families involving health professionals and volunteers. We have also created some folders in order to provide information about MPS and the appropriate precautions that need to be taken with the carriers of the syndrome.

We know that we have a long way to go. However, we do believe that we can give hope and quality of life for these 'special' people.

MPS in Taiwan

The first educational forum for families and caregivers on Mucopolysaccharide Diseases for Asia-Pacific Countries, took place on 17 - 18 May 2008, in Taipei, Taiwan.

The aim of the meeting was to bring together representatives of MPS Societies and closely related MPS patient organisations to exchange information and learn from the experts. Seven countries participated, Hong Kong, Indonesia, Malaysia, Phillipines, South Korea, Australia and the host country Taiwan.

Over two days the delegates heard presentations on all aspects of the diagnosis, clinical management and treatment of Mucopolysaccharide diseases as well as participating in question and answer sessions.

The role of the patient association was presented by the founder of the Taiwanese MPS Society, Mrs Virginia Tsai. Virginia's presentation is reproduced below.
Christine Lavery

Role of a patient group
Mrs Virginia Tsai
Founder of Taiwan MPS Society

First of all, on behalf of the Taiwan MPS Society, I would like to give a warm welcome to the representatives of each country that joins us for today's first Asia-Pacific MPS Conference. This is the first time the Taiwan MPS Society has initiated and sponsored the Asia-Pacific MPS Conference. MPS is the rarest disease among all other diseases.

However, I sincerely hope that we could use this conference to share our medical experiences in helping, treating and reducing suffering for children with MPS diseases in Taiwan with other countries.

For this conference, we have invited medical doctors from all the best teaching hospitals in Taiwan. They will present a series of reports based upon their MPS research, medical treatment, and care experiences. I profoundly believe that we could learn from one another through our exchanging and sharing of ideas.

Now, I want to introduce how the Taiwan MPS Society was founded, what the role of the Taiwan MPS Society has played and what the Taiwan MPS Society has done for children with MPS diseases and their families.

My very good friend, Dr Wraith, wrote a preface in my second book as follows: 'Providing help and support for children and families with MPS disorders is a challenging but rewarding task. It requires help from a wide range of individuals including doctors, nurses, therapists and family. An increasingly important role is being played by parent support organisations who can provide friendship, information, advocacy and practical help.

The Taiwanese MPS community benefit from a very active parent support group. Over the years, I have come across many Taiwanese MPS families and it has been a great pleasure for me to try and help the families and their physicians deal with these difficult disorders.'

I have lived in England for three years. During these three years, my first son, David was diagnosed with Hunter's. He received the best medical care from Dr Wraith and his team as well as unlimited assistance from the UK MPS Society. I was so touched by the unconditional help, love and care given by the people of England, that I vowed the rest of my life to serve children with MPS diseases as well as their families in Taiwan.

Over the past eleven years, I have shared a concept I picked up from the UK MPS Society with the people of Taiwan. I learned that by combining the efforts of medical doctors, nurses, therapists and social workers together into a single unit, we could have a better chance of combating MPS. I can proudly acknowledge that we owe most of our success leading up to this day by rooting this concept I mentioned above in Taiwan.

I also would like to concisely present the successful story of the Taiwan MPS Society. How we are acknowledged by the Taiwanese government, recognised by the Taiwanese society, supported by medical personnel, and MPS families in Taiwan. Not only did we use the concept of 'walking in someone else's shoes' to assist parents and MPS children but also, more importantly, we did whatever we could to fight for children with MPS disease who have the same right and dignity to live as healthy children.

Because many MPS children are disabled and are not prone to long distance travel, the Taiwan MPS Society annually sponsors three carnivals located in the north, middle and south of Taiwan. Carnivals held at the north and south ends of Taiwan are only one-day events. However, the carnival held at the middle of Taiwan is a two-day, one-night event.

During these events, we have a one-on-one policy. Each individual MPS child is accompanied and well taken care of by a volunteer. However, two volunteers are required to take care of an MPS child with MPS III, Sanfilippo disease, who may be hyperactive.

The main reasons to have volunteers for the MPS children are as follows. First, we want the parents to be able to relax and enjoy themselves during the carnival. Second, we want the parents to have the opportunity to share and exchange the experiences that they have acquired whilst raising and taking care of their MPS children. Lastly, our carnival enables these families to be able to encourage and help one another to continue to have the strength to face each day with such courage and dedication.

Whenever the two-day, one night carnival event ended, our volunteers always say that,

'We finally understand how parents with MPS children suffer. Although many of us will need to take one week to recover from the exhausting task of taking care of the MPS children during the event, these parents spend a whole year without a break. The parents are the true heroes with a spirit that has gone through multiple challenges but has always persevered.'

Since they were college students eleven years ago, the volunteers of the Taiwan MPS Society have made their organisation stronger by devoting their time to the Society and never missing any of the events. After witnessing their absolute devotion, I have realised that once you join our society, we become like glue and bond tightly to one another.

Without the financial support from many enterprises and non-profit organisations, we would have never been able to sponsor our annual carnivals. Whilst conducting our fundraising campaign, we plan out our budget and itemise every single spending to ensure financial control. Because we want to let our donors understand that every dollar will be used for MPS children, we invite all of our sponsors to attend the carnivals and see all the progress we had accomplished with their generous funding.

In addition, we mailed our brochures and publications in 376 Public Health Centres as well as to all the teaching hospitals in Taiwan in order to update MPS information collected from around the world.

In order to avoid the newborn babies carrying MPS genes, we also arranged special prenatal screenings conducted by doctors with MPS expertise for our member parents carrying the MPS genes who wish to have a healthy baby.

The Taiwan MPS Society was established from ground zero eleven years ago. We are moving forward due to so many great supporters. Although we sometimes feel totally hopeless facing MPS disease, I want to encourage you with 'Live life one day at a time' which is always used to encourage my member parents.

Once again, thank you for attending this conference. If you need more information or consultations, please let us know. It is our great honour to work together and help these lovely little angels fight against the MPS disease.

MPS in Japan

The Japanese MPS Society was founded by eight families in 1986 and was known as the Japan Welfare of Children and Families Association (JWA). In 1992 Japanes Health Insurance began to cover bone marrow transplant for MPS diseases and in 2001 the Japanese MPS Society took its name. In November 2004 the members successfully petitioned the Ministry of Health, Labour and Welfare to enhance the development of enzyme replacement therapy for MPS diseases in Japan. Subsequently ERT for MPS I was approved in 2006, for MPS II in 2007 and MPS VI in 2008.

The share of phenotype of the MPS diseases amongst the membership of the Japanese MPS Society is unusual. It is estimated that there should be about 300 children and young adults with MPS of which 163 are members of the Japanese MPS Society. What is striking is that 90 have MPS II (Hunter) equating to 55% of the membership. This is followed by MPS III (Sanfilippo) at 22 (13%), MPS IVA (Morquio) at 17 (10%) and MPS I (Hurler, Hurler Scheie and Scheie) 10 (6%). The average age of the affected members is 16 years with 70% under 20 years of age. About four families of affected children join each year and about 5 members lose their lives to MPS each year. **Christine Lavery**

Photo above: Sue Cotterell (MPS staff) with Barbara Wilson (MPS family) and Nobuhiro Kasa, Vice President of the Japanese MPS Society at the International MPS Symposium, Vancouver

Lady of Steel

Tuesday 8th March was the most unforgettable day of my life. This was the day they wheeled me into the operating theatre at 9am in the morning and kept me there until 7pm that night. When I woke up my head and spine was fused together all the way down to T3.

Of course this does not happen without a lot of other stuff that goes before it. Prior to this I was having massive headaches every day and lots of numbness in my arms and legs. As a result of the operation I no longer have the headaches and the numbness is fading. The Doctor tells me that my arms are now safe from any spinal damage caused by my failing vertebrae in my neck and will retain the strength they used to have. However the big trade off is that my head is now fixed in place and I can't move it at all, no more looking sideways at the cute boys!

For me this is a big change, not being able to move my neck is really hard, once again this is one of the many things you get when you live with MLIII. The build up to the surgery was hard, there was a lot going through my mind like how am I going to deal with the fact that I can't move my neck? What is it going to be like? Am I going to be able to do the things I have always been able to do? I guess what I am trying to say is how do the Morquio kids cope with it? It's only been three weeks and I am finding it hard.

I spent about a week up in Auckland with my Mum and Dad who were there with me. Two of those days were in the intensive care unit and the rest of those days were in the high dependency unit. Most of that time was a blur to me which is good, but there were times that were hard, like all the drugs that I was taking made me think funny things. Some nights I would wake and not know where I was and on the last night I woke up crying my eyes out because I thought I was not coming home. Mum and Dad had to come and calm me down. I did end up coming home.

The doctors were really nice. I had two, John Ferguson and Andrew Law, they were really good to me. John is a young doctor, but he is really good at his job. The one thing he wouldn't do was shave the back of my head so he got another doctor to do it from his team - he didn't want to partake in that one. The rest of the team were great too. They came to see me in the high dependency unit but half the time I didn't know they were there because I was asleep. I would like to thank them for all they have done.

Going back to learning new things! Getting undressed night and morning is hard because I don't have the same movement in my arms, but it has got easy over the last six weeks. Talking to people I have to turn myself to face them, that's not a bad thing. I just have to learn that I have no movement in my neck because I have tried to turn and realised oop! Can't I do that? Eating dinner is the other one because I can't bend my neck down and it is hard to see what I am doing, but I have learnt if I bend my middle it is easy to see. But getting my mind around this all is hard and I know it will come; it will just take some time. Is there anything I need to know from the Moruio kids and adults?

I have to tell you a funny story! I can walk too but have been using my wheelchair more often because it's got a headrest on it, but I have started to get back up walking too and as I have noted my head sticks out more than it did before, and one day it just came out - I said - "I have a turtle neck" because that's what it feels like. Mum and dad laughed at this and they said that's because you have a normal neck now as before my neck was sinking down and I had no neck and now I do, so dad said to me that I can hang more jewellery around my neck now! So when I go back to see the Doctor I am going to tell him that he fixed me but now I have a turtle neck and he will just laugh.

I also have to say that the amount of family and friends who have been by my side and my mum and dad's too has been amazing. Family and friends mean the world to me and they have just helped me get though this and they are probably going to say I did all the hard stuff and that probably is true, but if it wasn't for all the support, I wouldn't have got though this. Before I had my surgery my mum set up this care page so people could post on there and keep up to date on my progress. It was good because now I can look back on it and say wow! Did that happen? And look back on all the loving and caring posts. So thanks to everyone for your support!

So what next I wonder? Perhaps mum and I will look at dealing with the contractures and I am now old enough to explore hip replacements. Not sure if I want to go down this track but it would be nice to be able to walk without a walking frame. In the meantime I have to learn to deal with my new neck, having contact with some of the Morquio people to ask how they do things would be wonderful.

Sarah Noble, Mucolipidosis type III, New Zealand

Joint Lysosomal Storage Diseases Patient Meeting

On Sunday 15th June 2008 Dr Atul Mehta and Dr Derralynn Hughes together with their team at The Royal Free Hospital in London organised the first ever joint LSD Patient Meeting for adults with Gaucher, Fabry and Pompe Disease.

Dr Mehta and Dr Hughes organised the programme to ensure that the day focused on the patients and the welcome was given by UK Gaucher Association Chairman, Jeremy Manuel, who praised the organisers on this initiative and reminded those present of the benefit of the designation of the seven National centres which ensures that all patients with Lysosomal Storage Diseases are seen by experts.

Dr Derralynn Hughes presented to the audience of over 100 an introduction and overview to LSDs explaining their causes and effects and detailing both existing and emerging therapies and the way they operate. Although the manifestations of each of the diseases are quite different they are all diseases which result in the storage of material leading to the physical symptom.

Moving presentations were made by patients with each of the disorders and for many present this was the first occasion when they had heard about the experiences of a patient living with one of the other conditions. The presentations were deeply personal and in many ways were most uplifting. One participant said, "I did not know much about the other conditions but the challenge of coming to terms with a rare inherited long term disease that may have a treatment but no cure is very similar for all of us even though our symptoms may be quite different."

Dr Rob Anderson of the Health Technology Agency spoke about the National Collaborative Study of the Lysosomal Storage Diseases that is shortly to be starting. He explained the rationale behind what is to be a longitudinal study to capture information about the various diseases (the study is to start with collecting data on Gaucher Patients, MPS I Patients and Pompe Patients with Fabry Patients to follow) and a lively discussion ensued over the benefits of such study and the possible risks to patients.

After a well-earned lunch generously supplied by Marks and Spencer, participants broke into separate disease specific workshops.

Dr Ramaswami and Dr Hughes opened the Fabry workshop with two brief presentations on Fabry disease in children and adults respectively. This was followed by a discussion and question-and-answer session on a wide range of topics from genetic counselling and testing to Enzyme Replacement Therapy and Chaperone Treatment.

After tea a patient from each of the three workshops gave a short presentation of what had been discussed and what they had gained from the day. The meeting concluded with a vote of thanks given by Christine Lavery, Chief Executive of the MPS Society.

New Adult Inherited Metabolic Disease Service

An adult inherited metabolic disease (IMD) service has recently been established at the University Hospital of Birmingham (UHB) NHS Foundation Trust. The UHB is one of the leading NHS Foundation Trusts in the UK caring for half a million patients a year and a £521 million state of the art new hospital currently under construction due to open in 2010. The metabolic unit based in the Endocrine department consists of an adult consultant in inherited metabolic disorders (Dr T Hiwot), metabolic dietician (Louise Ryder) and specialist nurse pending an appointment.

HUB has a long tradition of providing metabolic service for adult patients under the care of various specialities such as liver, neurology, kidney and heart. The metabolic team is building on the experience of the aforementioned specialities and works closely with them as multidisciplinary team.

The adult metabolic service is also closely linked to Birmingham Children's Hospital (BCH) metabolic unit, who currently care for more than 1200 patients. A seamless transfer of care of adult patients from BCH to UHB is currently underway. We have also jointly established a transitional care for children between the age of 16 and 18 years old. BCH is one of seven centres nationwide to care for patients with lysosomal storage diseases and as a natural extension of the children's service, the UHB will be seeking national specialist commissioning funding to be a designated centre for enzyme replacement therapy for adults. This service will incorporate transitional care and will allow us to bring care closer to patients home.

Referrals are received from the Midland's area for adult patients with inherited metabolic conditions, including those with biochemical defect in carbohydrate, amino acid and fat metabolism, lysosomal storage diseases, mitochondrial disease, organic acidemia, urea cycle disorders, lipid disorders, etc.

Our contact details:

Dr T G Hiwot, Consultant Tarekegn.Hiwot@uhb.nhs.uk
Phone : 0121 627 1627, Ext. 51592 or 53546

Miss Louise Ryder, Dietician Louise.Ryder@uhb.nhs.uk
Phone : 07799038656

University Hospital of Birmingham, Raddle Barn Road, Birmingham, B29 JD, Fax: 0121 627 8589

Latest News

As will see from the articles and overviews that follow, there have been a series of new initiatives over the years that have led to approved treatments. Now we are in the very fortunate position that the increasing interest by the pharmaceutical companies to date has resulted in more pre-clinical research and increasing numbers of clinical trials.

To start with it might be helpful to members to understand a little of the drug development process.

The process for developing, testing and gaining approval for new medical therapies involves many steps. Each step has a different purpose and helps answer different questions. In Europe the drug development process is regulated by the EMEA (European Medicines Evaluation Agency). In the USA there is a similar process for developing new medicines through the FDA (Food and Drug Administration). Although it varies, the drug development process often lasts several years or more.

Each stage in the drug development process has a specific purpose. The process starts with laboratory and animal experiments, usually referred to as preclinical stage, and then moves in human studies known as the clinical phase. The clinical phase may last over four stages that includes safety studies, early efficacy studies in limited numbers of patients and finally a safety and efficacy study in larger numbers of patients.

MPS I

Aldurazyme™, administered once-weekly, has been approved in the United States and in 15 countries of the European Union for long-term enzyme replacement therapy (ERT) in patients with a confirmed diagnosis of MPS I, to treat the non-neurological manifestations of the disease. Aldurazyme was developed by BioMarin and Genzyme under a joint venture agreement that assigns commercial manufacturing responsibilities to BioMarin, and worldwide sales and marketing responsibilities to Genzyme.

Additional information can be obtained at **www.aldurazyme.com**.

MPS I Intrathecal ERT for Spinal Cord Compression
One-Year Extension Study Approved
Enzyme Replacement Therapy (ERT) has been developed for MPS I. ERT helps many physical ailments due to the disease, but does not treat the central nervous system, due to the inability to cross the blood brain barrier. The purpose of this study is to test delivery of ERT to the spinal fluid via intrathecal injection in patients with MPS I. In this pilot study, recombinant human alpha-L-iduronidase will be administered intrathecally once per month for four months to individuals aged 8 and older with the Hurler-Scheie and Scheie forms of MPS I and spinal cord compression. If successful, intrathecal delivery could represent a practical, straightforward method of treating central nervous system disease due to lysosomal storage.

Primary Outcomes: Safety of intrathecal enzyme treatment by blood and spinal fluid tests each month; improvement in neurologic signs related to spinal cord compression, by neurologic examination and Japanese Orthopaedic Scale each month; improvement in neurologic symptoms related to spinal cord compression, by subjective assessments and independence of functioning scale each month; improvement in mobility, by six-minute walk test each month; improvement in spinal cord compression by MRI imaging and somatosensory evoked potentials at baseline and four months; improvement in lysosomal storage by spinal fluid glycosaminoglycan levels at each treatment.

Secondary Outcomes: Improvement in spinal fluid pressure, by opening pressure measurements at each intrathecal treatment; improvement in hydrocephalus and other brain lesions by MRI at baseline and four months.

Expected Total Enrolment: 10

Additional Information can be obtained at www.clinicaltrials.gov/ct/show/NCT00215527?order=1

MPS I Intrathecal ERT for Children Being Considered for Transplantation
The University of Minnesota has recently obtained US Food and Drug Administration approval for the delivery of Laronidase into the spinal fluid of children with Hurler syndrome being considered for marrow/cord blood transplantation. The goal of these studies is to decrease the neuropsychologic decline that has been observed in children with Hurler from the time the patients are initially evaluated to the time they are one year from transplantation. The hypothesis is that there is a significant delay in achieving sufficient enzyme levels in the brain following transplantation, and that this may be overcome by giving enzyme into the spinal fluid until this occurs. Patients with Hurler syndrome who are between 8 and 36 months of age who have not previously received enzyme therapy and are being considered for transplantation at the University of Minnesota are eligible. Patients receiving Laronidase in the spinal fluid

will also be on intravenous Laronidase prior to transplant. The study will involve four doses of Laronidase given during a lumbar puncture (spinal tap) approximately three months before transplantation, at the time of admission to the hospital for the transplant, three months after the transplant and six months after the transplant.

MPS II

Elaprase™ is a long-term ERT for patients with a confirmed diagnosis of MPS II, Hunter Disease, which has been approved for use in the United States, Canada, and many countries in Europe. Elaprase was developed and is produced by Shire Human Genetic Therapies (formerly TKT), and is given as weekly infusions to replace the missing enzyme that Hunter syndrome patients fail to produce in sufficient quantities.

Additional information can be obtained at **www.shire.com**.

Shire Human Genetic Therapies is committed to conducting a clinical trial in individuals with MPS II who have neurological involvement. Currently this study is projected to be at the University of North Carolina.

MPS III

Shire Pharmaceuticals group, as part of their research to evaluate new approaches to the problem of treatment of the central nervous system, is hoping to move its MPS IIIA, Sanfilippo Disease, program forward. If the trial to directly administer the enzyme into the central nervous system of individuals with MPS II is successful, Shire hopes to expand their research initiatives to include MPS IIIA. The Shire website is www.shire.com.

Use of Miglustat in Sanfilippo Disease (MPS III)
In early 2007, a clinical trial with Miglustat was initiated by the Hospices Civils de Lyon (HCL), France, in patients with Sanfilippo Disease. A majority of the participants were from France with two patients enrolled from England. HCL acted as the sponsor and Dr Nathalie Guffon took the role of principal investigator. This placebo-controlled, double-blind trial enrolled 25 patients for 12 months. A short extension of the trial with the two arms was planned until the end of July 2008 to allow HCL to compile, review and analyse the data.

In June 2008, the results of the trial were evaluated by an independent committee of experts. In July, an individual patient analysis was also performed. The committee concluded without any ambiguity that the results could not support any kind of clinical efficacy for the use of Miglustat in patients with Sanfilippo disease. The safety and tolerability data were similar to that which has previously been seen in other lysosomal storage disorders, with diarrhoea presenting as the main adverse reaction. Six serious adverse events were recorded during the trial, none of them drug-related. One patient discontinued. The results of the trial will be placed on the websites of the Hospices Civils de Lyon (HCL) and Orphanet by the end of September 2008.

In conclusion, therefore, there is no evidence at all that Miglustat may be of any benefit to patients with Sanfilippo Disease. Consequently, Actelion UK will not consider any application for the compassionate use supply of Miglustat in Sanfilippo patients.

MPS IVA

Enzyme Replacement Therapy for MPS IVA
Two companies have announced Natural History Studies and Clinical Trials for MPS IV, Morquio disease. Vivendy will carry out a natural history study of 65 patients aged 7-15 years running for 12 months from September 2008. Participants must be able to walk a minimum of 5 metres. The clinical trial is planned to start in February 2010.

BioMarin will carry out a natural history survey (MorCAP). Recruitment of 300 patients will start in October 2008. Clinical trial phase 1 / 2 for ERT in MPS IVA will start in March 2009. This will be followed up by a larger pivotal clinical trial. See press release page 39.

MPS VI

Naglazyme™ is the ERT for individuals with a confirmed diagnosis of MPS VI, Maroteaux Lamy disease and has been approved for used in the United States and in many European countries. Developed and produced by BioMarin Pharmaceutical, Inc, Naglazyme has been shown to improve walking and stair-climbing capacity.

Additional information can be obtained at **www.biomarinpharm.com.**

ML II/III

The following update is kindly provided by Jenny Noble, Secretary of Lysosomal Diseases New Zealand (LDNZ).

2007 - First ever Natural History began for ML II and ML III patients at the Greenwood Genetics Clinic. 50 Families from USA, England, Australia, Canada and New Zealand took part. Families supplied blood samples for analysis and Gene mutation diagnosis. X-Rays and original diagnosis information was also supplied.

It was always thought that ML III was on a different sub unit from ML II. The diagnosis of the 50 patients showed that they were all on the Alpha Beta sub unit, and are classified as either an A or B mutation. However, 9 ML III children in Israel have also been through diagnosis via a separate program, their mutation is on the Gamma sub unit and are classified as ML IIIC.

Of the 50 children tested, 5 children have a stop codon in the gene sequence and 7 children have come back

with an unusual mutation. These children are classified as ML II/III. Dr Leroy and Dr Sarah Cathey are continuing their research with this unusual mutation.

1. Drs Braulke and Tiede researchers in Germany have developed knock out mouse models of the alpha/beta and gamma subunits.

2. Drs Steet (husband and wife team) from Georgia USA have developed zebra fish to study the skeletal complications in ML and other LSD's.

3. Dr Kakkis/BioMarin Labs are treating ML II and ML III cell lines with Aldurazyme. What they found was that there was no change to the ML II cell lines. The ML III cell lines showed some changes. Dr Thomas Braulke (Germany) was asked to repeat the tests on the cell lines and confirms the changes in the ML III cell lines. The data to date would suggest that it is possible to have some effect but it is not clear how much. Dr Kakkis is now considering treating some ML III patients with Aldurazyme.

This is a very exciting step forward. Since 2004 myself, Dr Burton and Lynda Nagy (ISMRD board members) have been suggesting to Dr Kakkis and others that perhaps ML might benefit from a cocktail of ERT as ML III in particular has a number of enzymes that have the targeting messages missing but still function at a low level.

4. The 1st International consensus meeting on Bisphosphonate Therapy in Oligosaccharidoses is being held in New Zealand on 19th - 20th November 2008. This meeting is bringing together more than 12 International experts in this field along with many observers, to discuss the issues and treatment of bone disease in ML and other LSD's. LDNZ wishes to acknowledge the support of the UK MPS Society, without their help this very important meeting would not be moving forward.

The goals and focus for this meeting are:

1. To understand the musculoskeletal complications in Oligosaccharidoses.
2. The clinical benefits of Cyclic Intravenous Pamidronate and related Bisphosphonates.
3. Bone Pathology in Mucolipidosis and the Mucolysaccharidoses.
4. Results of Bisphosphonate therapy in Fucosidosis, Mannosidosis, MPS VI, Galactosialidoses.
5. Consensus statement on treatment with Bisphosphonate therapy in ML II and ML III
6. Clinical trials for skeletal and Lysosomal disorders.

LDNZ and ISMRD (the International Advocate for Glycoprotein Storage Diseases) are thrilled to be involved in helping to push these research initiatives forward. We have learnt that even though we are a small voice we are playing a rather large role in getting some movement for the super orphan diseases within the Lysosomal group.

I can also add that the first ever Natural History Study has been started for Fucosidosis and is being compiled by Prof John Hopwood, Adelaide and Dr Michael Beck and Carolyn Paisley-Dew, one of ISMRD's board members.

Update appears courtesy of Jenny Noble, Secretary, Lysosomal Diseases New Zealand

Fabry Disease

Amicus Clinical Program Update: AT1001 for Fabry

Amicus has released results from the initial treatment phase of four Phase 2 studies of AT1001, a pharmacological chaperone under investigation for Fabry disease. A fifth extension study is ongoing to evaluate different doses and dose regimens.

The phase 2 studies included either 12 or 24 weeks of treatment with AT1001 in men and women. Key findings include:

AT1001 was generally well-tolerated. The studies are ongoing and data continue to be collected.

A majority of males and females showed an increase in alpha-Gal A levels after treatment with AT1001 in key tissues including white blood cells, kidney and skin.

Reduction in kidney substrate accumulation was assessed by measuring GL-3 levels in urine. The majority of male and female subjects who showed increases in alpha-GAL A levels also showed decreases in urine GL-3.

A majority of subjects who showed increases in alpha-Gal A levels self-reported an improvement of their Fabry symptoms.

Amicus plans to meet with regulatory authorities this year to discuss the design of a Phase 3 trial for AT1001.

Amicus and Shire HGT Collaborate

Amicus has entered into a strategic collaboration with Shire Human Genetic Therapies Inc. to jointly develop Amicus' three lead pharmacological chaperone compounds for lysosomal storage disorders (AT 1001 for Fabry Disease, AT 2101 for Gaucher Disease, and AT 2220 for Pompe Disease). Shire has received rights to commercialise these products outside of the United States. Amicus will retain all rights to commercialise these products in the US.

John Crowley, CEO of Amicus, stated: 'We are immensely pleased to enter into this partnership with Shire, which leverages both companies' unique experience and expertise in developing therapies for lysosomal storage disorders.

The combination of Amicus' strong science foundation in pharmacological chaperones and Shire's proven track record in drug development and commercialisation will greatly enhance our efforts to bring these novel therapies to patients.'

Matthew Emmens, Shire's CEO said: 'Amicus' pharmacological chaperone products have the potential to be an excellent addition to our current enzyme replacement therapy business. This technology should provide significant benefit to patients with these serious genetic diseases.'

Article reproduced from Amicus Connection, Volume 2, Issue 1, Spring Summer 2008

BioMarin announces two clinical studies for Morquio patients

BioMarin Pharmaceutical is focused on the development of therapies for areas of high unmet medical need, particularly orphan diseases. In just 10 years BioMarin Pharmaceutical Inc. has made available three breakthrough products to patients around the world. Two of these products are enzyme replacement therapy treatments for lysosomal storage diseases: Aldurazyme® (laronidase) for Mucopolysaccharidosis I (MPS I), and Naglazyme® (galsulfase) for MPS VI.

Now, BioMarin is investigating a potential treatment that may benefit patients diagnosed with the lysosomal storage disease, Mucopolysaccharidosis IVA (MPS IVA), which is also known as Morquio syndrome. BioMarin plans to initiate two clinical studies: MorCAP (a clinical assessment program) and a Phase 1 / 2 clinical trial.

Individuals who have participated in the International Morquio Organisation (IMO) survey are welcome and encouraged to participate in MorCAP and/or BioMarin's Phase 1 clinical trial.

MorCAP, the Morquio Clinical Assessment Program, has been designed to provide a fuller understanding of Morquio syndrome by measuring endurance and respiratory function in affected patients among other important aspects of Morquio syndrome. These insights will help BioMarin design future clinical trials as well as understand the medical needs of Morquio patients. Participating in MorCAP will require one or more visits to a clinic or hospital. This study is scheduled to begin in September 2008.

'We hope to learn the extent and depth of disease in this program and we want to include as many patients as possible.' says Dr Emil Kakkis, Chief Medical Officer, and Senior Vice President of BioMarin.

In early 2009, BioMarin plans to begin enrolling a small number of patients in a Phase 1 / 2 clinical trial investigating a potential treatment for Morquio syndrome. The objective of this clinical study is to establish dose response to an enzyme replacement therapy for Morquio syndrome based on pharmacokinetic and pharmacodynamic parameters.

Pending results of the Phase 1 / 2 study, BioMarin expects to conduct a Phase 3 double-blind, placebo-controlled study enrolling up to 100 patients. This study will likely to be conducted in many centers.

Preliminary experiments were designed to demonstrate that the native enzyme being investigated by BioMarin can reach the bones and other tissues that are affected in Morquio patients, says Dr Kakkis. 'We have verified that it is taken up well and that it can penetrate growth cartilage in animal models. It appears to naturally bind bone in in vitro studies. Data published to date suggests the native enzyme can clear storage in many cell types including bone cells.'

To learn more about participating in MorCAP or the phase 1 / 2 study, visit **www.morquioBRMN.com**. Individuals may also register at the website to receive updates on trial developments.

About Morquio syndrome: Morquio syndrome is an inherited disease caused by the lack of a specific enzyme that is essential in breaking down glycosaminoglycans, or GAGs. GAGs are composed of long chains of sugar molecules used in building bones, cartilage, skin, tendons and many other tissues in the body. Individuals with Morquio syndrome are missing an enzyme used to break down a specific GAG called keratan sulfate. Incompletely broken down keratan sulphate remains stored inside the Morquio patient's cells and begins to build up, causing progressive damage. Morquio syndrome is estimated to occur in 1 in 200,000 to 300,000 live births.

New DVD resource available

The Journey from Diagnosis to Therapy

An educational DVD for further understanding of Mucopolysaccharidosis VI, **Maroteaux Lamy disease**

Includes sections on:

Diagnosis

Management and therapy

The patient and family

Summary of product characteristics

For a free copy please contact the MPS Society by phone on 0845 389 9901 or email **mps@mpssociety.co.uk**

WANTED!

Adventurous people throughout the UK to do something like this...

for

FREE!

Minimum sponsorship required

Help us raise funds by making an exhilarating 10,000 feet freefall parachute jump. No experience is required and if you raise the minimum amount of sponsorship you will get to jump for FREE!

Phone **0845 389 9901**
or email **fundraising@mpssociety.co.uk**