MPS

Care Today, Hope Tomorrow

Winter 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: Josiah Oyawale (MPS IV, Morquio Disease) with his new laptop, one of the wishes from the Ollie G Ball 2008

MPS Society

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Registered Charity No. 287034

Magazine Deadlines

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

Become a Friend of MPS

CONTENTS

CHIEF EXECUTIVE'S REPORT

Winter 2008

The usual New Year wishes are this year tempered with recognition that not only are our members and their families dealing with the day to day challenges of MPS, Fabry or a related lysosomal disease, but for many they face an uncertain year in the job market and higher bills. We don't underestimate the additional burdens this economic downturn may have and want you to know the advocacy team are here to listen and wherever possible provide practical support.

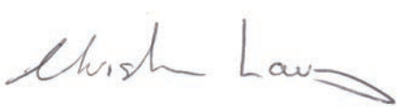
In December the Society's Board of Trustees presided over one of the most challenging budget meetings in recent years because it is difficult to predict how Trust and corporate donations as well as fundraising will be affected by the credit crunch. I am pleased to say that we have recruited to the two vacant posts of Advocacy Officer and Sarah Irvine and Jolanta Turz start in February. At times like this it is hard to ask you to do more in fundraising for the Society but the more funds raised, the more we can do for our members and their families throughout the United Kingdom.

This Autumn has again been a continual round of collaborative meetings and specialist conferences providing unique opportunities to promote Mucopolysaccharide, Fabry and related lysosomal diseases. An important new development in November was the launch of a dedicated Lysosomal Storage Disease Service for Wales. We welcome this initiative of Dr Graham Shortland, Dr Mike Badminton and Dr Geoffrey Carroll and you can read more on this further on in the MPS Magazine.

Plans for the BioMarin MPS IVA natural history study (MorCAP) and clinical trial for Enzyme Replacement Therapy are progressing and the Society welcomes the news that three paediatric national specialist commissioning sites, Great Ormond Street, Birmingham and Manchester will be participating. Regular updates on MPS IVA and other MPS diseases from a clinical and research perspective will be given in the MPS Magazine and MPS website, www.mpssociety.co.uk.

During 2008 we engaged with a record number of people who matter most to the MPS Society - children and adults with MPS and Fabry Disease, parents, partners, caregivers and volunteers; doctors, nurses, teachers, social workers and scientists; the pharma industry, politicians, fundraisers and donors.

We wish every one of you a Happy New Year and thank you all for your support over the past year.



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 Meetings held on 3-4 October and 6 December 2008

Personnel

Trustees agreed to the appointment of an external HR company, Peninsula, who will provide the Society with 24 hour support and advice as well as giving a critique of the Society's policies and procedures. With the ever-increasing changes to employment law this was the best way to manage risk in this area. In December, Trustees were advised that one Advocacy Officer had been recruited and a second appointment was under consideration. *[The appointment of the second was confirmed subsequently and both start in February 2009].*

Fundraising

At the October meeting two MPS members and a businessman presented their ideas for helping to generate income for the Society. Mindful of the need to generate more long term income to underpin the services offered by the Society it was agreed to seek a Fundraising Officer in 2009.

Brains for Brain (B4B)

Trustees were appraised by the CEO of the B4B meeting she attended in Frankfurt to help compile the application for the EU 7th Framework application to fund vital research into MPS diseases. In December Trustees were advised that the B4B application had been submitted to the EU. The outcome is expected in March 2009.

Treasurer's Report

In October, having presented the current financial position the Treasurer advised Trustees that the Society had received £40,000 from the proceeds of Ann Canton's home (MPS member since 1982) following her death. In December the Trustees unanimously agreed a new staffing structure and approved the income and expenditure budgets for y/e 31 October 2009.

Jeans for Genes

Trustees were advised that the MPS Society provided a Fabry family to take part in the Fabry video for Jeans for Genes funded by Genzyme and Shire Pharmaceuticals. In December, Trustees were advised that MPS, based on information provided by the CEO of Jeans for Genes, should receive around £180,000 from the proceeds of the 2008 Jeans for Genes campaign.

Support to Members

Trustees were updated on all aspects of the Society's events and very encouraging feedback was received on the Morquio conference. It was proposed and agreed that if funding can be found a similar conference be held in 2010 for Sanfilippo disease.

Research

Tipping the Lens - The CEO advised Trustees that there are plans to undertake a Phase 2 of the Tipping the Lens Project. It was agreed that this was a great initiative which mobilised the Society's MPS IVA members especially at the Morquio conference.

MPS I Patient Survey - The CEO tabled a paper on the International Patient Survey of MPS I Disease Management and informed Trustees that the UK scored highly.

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.

WHAT'S ON!

2009

MPS CLINICS

16 January	Manchester BMT clinic (under 6 yrs)
23 January	Manchester BMT clinic (over 6 yrs)
3 February	Newcastle MPS Clinic
27 February	Birmingham MPS Clinic
24 April	Manchester BMT clinic (under 6 yrs)
1 May	Manchester BMT clinic (over 6 yrs)
14 May	Northern Ireland MPS Clinic
12 June	Birmingham MPS Clinic
17 July	Manchester BMT Clinic (under 6 yrs)
24 July	BMT Clinic (over 6 yrs)
16 October	Manchester BMT Clinic (under 6 yrs)
23 October	Manchester BMT Clinic (over 6 yrs)
19 November	Northern Ireland MPS Clinic
20 November	Birmingham MPS Clinic

CONFERENCE EVENTS

26 - 28 June	MPS National Weekend Conference
17 - 21 December	MPS Disney Conference Orlando

REGIONAL EVENTS

15 May	MPS Awareness Day
27 June	Childhood Wood Remembrance Day
2 October	Jeans for Genes Day
23 October	Childhood Wood Planting

Would you like to volunteer for MPS?

Volunteering is fun and rewarding. It could also help you learn new skills and gain valuable work experience. The MPS Society relies on volunteers for our events and conferences to assist in the care needed for children and young adults affected by MPS and Related Diseases. All of our volunteers undertake training in moving and handling and are fully briefed prior to the event. Volunteers should be 18 years or over, will need to provide two references and undergo a Criminal Records Bureau check and attend a training day in Amersham. Those volunteering for our conferences will receive accommodation and all meals throughout the weekend. ***Contact us now to register your interest and availability.***

MPS National Conference, 26 - 28 June 2009, Northampton Hilton Hotel

New Members

Imtiaz and Farhat Bhatti have recently been in contact with the Society. Their daughter has a diagnosis of Sanfilippo Disease. Anya is three years old. The family live in the Midlands area.

David and Mette Fisher have recently been in contact with the Society. Thomas is four years old and has a diagnosis of Hunter Disease. Thomas has a six year old brother and a nine year old sister. The family live in the South East.

Michael Gough has recently been in contact with the Society. He has a diagnosis of Fabry Disease. Mr Gough lives in the Midlands area.

Mr and Mrs Thompson have recently been in contact with the Society. Tommy has a diagnosis of Sanfilippo Disease. Tommy is three years old. The family live in the South East.

Deaths

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

David Haskell who died on 22 August 2008 aged 37 years and who suffered from Fabry disease.

Matthew Home who died on 14 October 2008 aged 14 years and who suffered from Hunter disease.

Suhayb Khan who died on 9 November 2008 aged 1 year and who suffered from ML II.

Suhayl Khan who died on 9 November 2008 aged 1 year and who suffered from ML II.

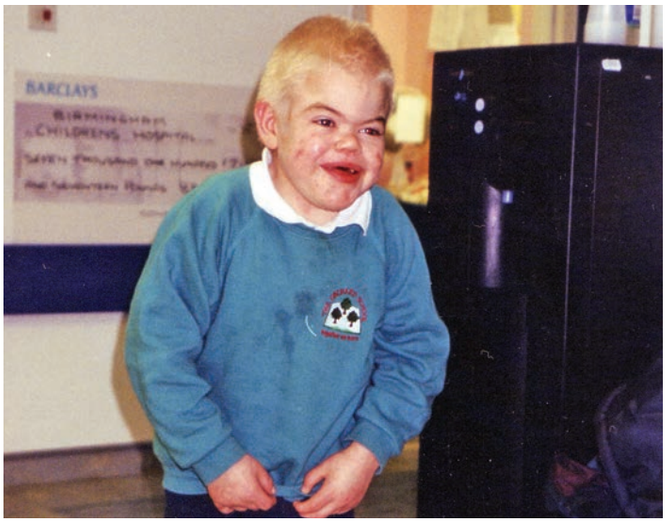
Benjamin Williams who died on 12 November 2008 aged 13 years and who suffered from Hunter disease.

Wajid Sadiq who died on 17 November 2008 aged 10 years and who suffered from Multiple Sulfatase Deficiency.

Claire Rowland who died on 13 December 2008 aged 38 years and who suffered from Fabry disease.

Births

Many congratulations from all at the MPS Society to Miriam Blowers, our Volunteer and Event Co-ordinator and her husband Duncan. Miriam and Duncan became the proud parents of Solomon Zach Blowers on 23 November 2008. Solomon weighed 7lb 5oz.



Benjamin Williams (MPS II)

Sadly missed...

Nigel Frost sadly passed away on 7 February 2008. Nigel was a civil servant for nearly 19 years. He started his career in the Employment Service in Watford and in 1994 he became a member of the Department for Education.

From 1997 he worked in the Teachers' Pay Team where he stayed throughout his time here. He remained resolutely cheerful and committed to his work whilst suffering many health problems.

He was an inspiration and it was fantastic to see his zest and enthusiasm for life. Colleagues in the Department collected more than £200 for the MPS Society which championed the condition that Nigel suffered from.

Article appears courtesy of Feedback; Department for Children, Schools and Families, Issue 42.

ANNOUNCEMENTS

Matthew Home suffered from Hunter Disease and died on 14th October 2008 aged 14.

My Brother

He may not have learned to read or write, or to run and play

But he carried on with his life in his own special way

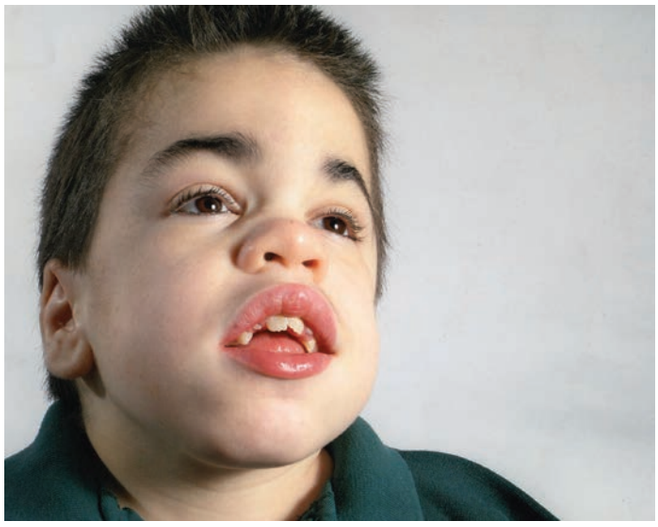
He enjoyed school and loved his TV

That he laughed at with much glee

From his special bed to his wheelchair

He couldn't have lived a happier life anywhere

by Becci Home



Heaven's very special child

A meeting was held quite far from Earth!
It's time again for another birth.
Said the Angels to the LORD above,
This Special Child will need much love.

His progress may be very slow,
Accomplishments he may not show.
And he'll require extra care
From the folks he meets down there.

He may not run or laugh or play,
His thoughts may seem quite far away,
In many ways he won't adapt,
And he'll be known as handicapped.

So let's be careful where he's sent,
We want his life to be content.
Please LORD, find the parents who
Will do a special job for you.

They will not realise right away
The leading role they're asked to play,
But with this child sent from above
Comes stronger faith and richer love.

And soon they'll know the privilege given
In caring for their gift from Heaven.
Their precious charge, so meek and mild,
Is HEAVEN'S VERY SPECIAL CHILD.

by Edna Massionilla
December 1981
The Optomist - newsletter for PROUD
Parents Regional Outreach for Understanding Down's Inc.

Mazda London Triathlon

The Society is delighted to have secured some Bond places in the Mazda London Triathlon in August 2009.

To enter, please contact the MPS Society for an entry form: **fundraising@mpssociety.co.uk**, or phone 0845 389 9901.

For more information on the event visit **www.thelondontriathlon.com**

There are a number of race entry options available to you so please make sure you specify which race you would like to do. There is a swim, bike and run with varying distances depending on your ability and fitness level.

We are asking for a deposit of £75 to secure your place with a minimum possible pledge of £650.

Just Giving
Just Giving are an official partner of the event, and last year The London Triathlon was the second largest online fundraising event after the marathon, so it really is a great way to get your sponsorship.

Training days
Training days will be available and more information will be available closer to the event on www.thelondontriathlon.com

Celebrating
Jeans for Genes Day 2008

Rickmansworth School fundraising

Rickmansworth School in Hertfordshire raised funds for Jeans for Genes Day by holding a variety of events involving their pupils from Year 7 to Sixth Form. Everyone got busy baking for three cake sales held during the week leading up to Jeans for Genes Day, and the school also ran a short story competition and a debate on genetic engineering. A photo competition on the theme of "My Jeans Are More Interesting Than Me" produced many thought-provoking entries, with photos of pupils wearing their jeans in interesting situations!

Pupils learnt more about Jeans for Genes and The MPS Society with assemblies from Sophie Thomas (MPS Society Senior Advocacy Officer), and the week rounded off with pupils and staff donating £1 to wear their jeans as part of a non-uniform day.

Many thanks to all the pupils and staff (some of whom are regular MPS childcare volunteers) who took part in these events and who helped to organise them, especially Mrs Caroline Wilkes who co-ordinated the fundraising effort.

JEANS FOR GENES APPEAL

2 October 2009
www.jeansforgenes.com

MPS Staff participation

Several MPS Staff members gave talks on Jeans for Genes and The MPS Society at various school assemblies in Buckinghamshire and Hertfordshire during the week leading up to Jeans for Genes Day.

Christine Lavery, Chief Executive, gave talks during two assemblies at Dr Challoner's High School in Little Chalfont. Sophie Thomas, Senior Advocacy Officer, spoke at assemblies at the Berkhamsted Collegiate School, at Rickmansworth School, and at Woodside Junior School in Amersham. Sue Cotterell, Office Manager/PA to Chief Executive, drew on her previous experience as a teacher to give two primary school assemblies, at Bell Lane School in Little Chalfont, and St George's Infant School in Amersham.

The talks were designed to be age-appropriate for the varying audiences, and included a slideshow of photos, facts and figures to enable the pupils to gain a greater understanding of genetic diseases and what Jeans for Genes Day raises money for.

Ollie G Big Top

On Saturday 14th June 2008 the Ollie G Big Top Ball was held at Whithorn Farm in Surrey.

Guests were welcomed with a glass of champagne and treated to a fantastic aerial display by the Royal Navy Heritage display team.

Dinner followed in the Big Top which was lavishly decorated in a circus theme with festoons of silk, lights and a dazzling pink and purple colour scheme.

There were a number of wonderful acts on show that evening, including cabaret performances from the Globe Girls, trapeze acts and fire eating. Jo Wilson, herself an MPS sufferer, beautifully sang Wind Beneath My Wings.

The Main Auction, hosted by Nick Bonham, was held next with some stunning prizes on offer for those who bid highest.

The evening ended with live music from a number of distinguished musicians including Gary Brooker, and Eric Clapton who closed the evening's entertainment.

The MPS Society would like to thank David Gosling in particular and all those who participated in the organisation of the Ollie G Big Top Ball. We would also like to pass on our enormous appreciation to those who so kindly donated money. We were delighted that the Ball raised £196,464 for the MPS Society.

David and Simone Cawkhill attended the Ollie G Ball in June 2008 and bid on a silent auction item which was "Dinner at the Crown Inn in Chiddingfold for 36 Guests", very generously donated by Dan and Hannah Hall.

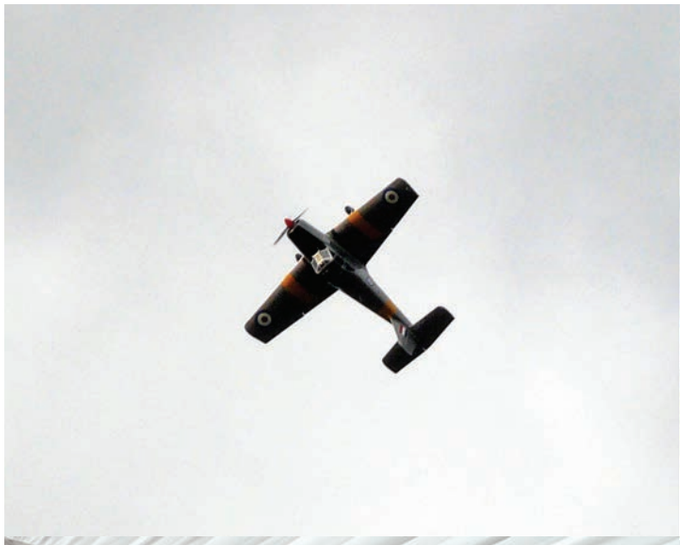
David Cawkhill wrote to the MPS Society in November as follows:
One of the auction prizes at the Ollie G Ball this summer was dinner for 36 at, and donated by, the owner of, the Crown Inn in Chiddingfold, Surrey. We used that as an opportunity to raise some more money for the MPS Society.

Everyone who attended the dinner had an excellent evening. David Gosling had a couple of prizes left over from the Ollie G Ball which we also managed to auction. He will collect £5,000 proceeds from two of the guests and so in total including gift aid the evening raised a little over £9,000 for the MPS Society.

It is nice to have combined an opportunity to raise money with an evening of such pleasure.

Our grateful thanks go to David and Simone Cawkhill for their kindness and generosity, and to Dan and Hannah Hall for providing the guests with wonderful food, free flowing wine and their amazing hospitality.

Ball 2008



Ollie G Wishes 2008

Here are some of the lucky recipients of the Ollie G Wishes 2008. Each of the families would like to convey their appreciation and gratitude to those who organised the Ollie G Ball 2008 and all the guests on the tables who pledged their support. A big thankyou!

Georgia, Sanfilippo Disease, 12 years old

Georgia is a beautiful 12 year old little girl with Sanfilippo Disease. This is a progressive disease associated with physical and mental disability which worsens over time. She has deteriorated a great deal, her mobility is very limited and she is no longer able to talk. Throughout everything that she has to deal with, she always manages a smile and her spirit shines through. Even though Georgia cannot talk and despite everything, she is after all, still a little girl, I'm sure this would be her wish…

"If I could, I would wish to have a magical 'make over' for my new bedroom, to make it extra special and fit for a princess (that's what my dad says I am)."

"At the moment, my new bedroom is empty and bare. Mummy and Daddy were waiting for my bathroom to get finished. That's finished now, but my bedroom looks quite scary. There's equipment in there and a huge monster right across the ceiling, that I'm really frightened of. Mum says it's a hoist, I don't know what that means, but I know I don't like it!"

"My new bedroom is bigger and if it was fit for a princess I could spend time in here with some of the special people who help to look after me. They are nurses and they love to play, read and sing to me. I'm not sure what it's all about, but if it keeps them happy, I'll go along with that!"

"In my new bedroom, when I'm resting, I could look out of the window at the trees, which I love to do. I like watching the birds and squirrels too. I'm waiting for my new bed; this will go in my new bedroom. It's a special cot bed that will stop my arms and legs getting stuck down the sides. Mum says I'm a 'wriggly worm' in bed, but how can I be a worm, when I'm a princess?"

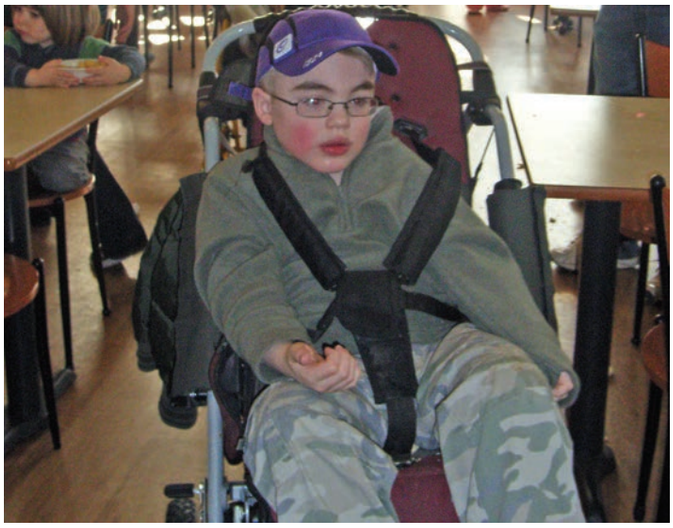
"Mum has tried to put me in the monster on the ceiling, for me to get used to it, but I get very scared and frightened. I don't want to 'get used to it', but I don't think it's going to go away."

"Maybe if I had a magical princess's bedroom I could ignore the monster, pretend it wasn't there. Maybe one day it will become my friend and I'll wonder why I was so scared."

Georgia's new bedroom is currently empty, a blank canvas. We would like some flooring, furniture plus some 'magical princess' accessories. **Georgia's Mum**

Below are two photos of Georgia receiving her gift of bedroom accessories, sensory lights and TV.

Ben, Sanfilippo Disease, 9 years old



Ben is 9 years old and has Sanfilippo Disease. This is a progressive disease associated with physical and mental disability that worsens over time. Ben has become less active and falls over much more easily. Even with all the problems and pain caused through this condition Ben remains a very happy friendly child. I would love for some help to be able to make Ben's room into a more stimulating environment which is also relaxing for him. I feel that Ben would really benefit from having some lights and sensory equipment. **Ben's Mum**

Ben's wish for sensory lights has been granted and he and Ben's mum were delighted.

Josiah (9 years), Morquio Disease

Morquio Disease is associated with very severe physical disabilities. Josiah would love to have a lap top computer. This will enable Josiah to do his homework and keep up with school work. Because of their disease, children with Morquio often find it very difficult to write with a pen or pencil. They often have very lax joints which can be very stiff so it makes gripping a pen difficult for any length of time. If he was lucky enough to get a laptop Josiah would be able to do his homework on the lap top which would be much easier for him as he goes through the next important stages of his education.

Josiah's Mum wrote to thank MPS for informing that Josiah's dream of owning a laptop was to be fulfilled thanks to the generosity of the Ollie G Ball. She wrote that the timing could not have been more perfect as Josiah had just returned from the hospital having been told that he would probably need an operation on both legs. The news about the laptop changed his countenance.

The photo below shows Josiah with his new laptop surrounded by this sister and brothers.

News from Great Ormond Street

I am a Clinical Psychologist who has recently joined the Metabolic team at GOSH. I work mostly with children that have a lysosomal storage disorder. Overall, my aim is to work together with families to enhance their well-being and quality of life. Often this involves me linking in with local services and making sure families can access help close to home. I can also be available to attend school reviews and professional meetings when necessary.

I can provide 1:1 work with children or young people, family appointments and can also support siblings. I am working with a colleague to set up some group work in the future as we know that children, young people and families often benefit from meeting others and sharing their own experiences, as well as, expertise.

Dr Jenny Jim
Principal Clinical Psychologist

Update from the Enzyme Lab at GOSH

The Enzyme Laboratory is a highly specialised, fully nationally accredited laboratory involved in the diagnosis and monitoring of a wide range of lysosomal storage disorders (LSDs) and other metabolic disorders at an international level.

We have 11 members of staff including a Principal and Senior Biochemist, a Chief Biomedical Scientist and two Senior Biomedical Scientists (one of which is funded by NCG). We are currently in the process of recruiting a Professor to head the laboratory, expand our test repertoire and develop our research interests in the field of LSDs. We work very closely with the Regional Molecular Genetics Laboratory and the Histopathology Laboratory to provide a comprehensive on site diagnostic service for LSDs.

Although the laboratory is currently located in the Institute of Child Health it will soon be moving to join the rest of Pathology in a modern bright laboratory building on the main GOS hospital site.

The Enzyme Laboratory is regularly represented at GOS multidisciplinary meetings and forms an integral part of the LSD clinical service offered at Great Ormond Street.

Physiotherapy for MPS at GOSH

My name is Michelle Wood (pictured above) and I am the physiotherapist for the metabolic team at GOSH and I see all the MPS children. I have been working with this group for over 10 years and it has been great to see new treatments, for some conditions, emerge and implement our therapy accordingly.

I see all MPS patients either as in-patients or out-patients and then feedback my findings at our weekly multi-disciplinary meeting. We therefore have an excellent team approach ensuring a holistic view to the condition. My role in the team is to assess physical abilities. I measure range of movement, muscle power and look at various functional abilities such as dressing, rising to standing, walking ability etc.

We have a relatively new piece of equipment - the Gaitrite which is a mat embedded with several thousand sensors. As children walk along it we can monitor very closely their walking pattern. It assesses each footfall to see if both feet adopt the same posture, how big a stride is taken, how much weight is taken through each foot as well as many other useful parameters. All this information is very valuable for monitoring physical abilities and measuring any response to treatment. It is easy to repeat and everyone enjoys the assessment.

I am always available for telephone discussion with parents and local physiotherapy teams for any physiotherapy related problems.

I have been working in Leicester Royal Infirmiry as a Consultant paediatrician with a special interest in metabolic medicine and I am really excited to be joining the metabolic team at Great Ormond Street for the next two years whilst Dr Cleary is in Singapore. I have previously worked in the metabolic team here and it is obviously a great pleasure to return. I look forward to meeting Dr Cleary's patients over the next few months. **Lara Abulhoul**

A new Clinical Research Facility for GOSH

On Monday 8th December a new purpose built, children's research facility opened on level one of the Frontage building in Great Ormond Street Hospital. The Somers Clinical Research Facility (CRF) offers for the first time, a dedicated environment for children, young people and staff taking part in research studies and clinical trials. Prior to the unit opening, it was necessary to use outpatient space or inpatient beds.

The CRF is located in the heart of the hospital and has access to all of the facilities you would usually find in a clinical area. It therefore provides a high quality and safe clinical environment in which research programmes can safely and effectively be undertaken, according to robust and ethically approved protocols. This may be an ideal area for any future Enzyme Replacement clinical trials.

My name is **Sonia Lozano** and I joined GOSH as a Speech and Language Therapist in June 2008. I work as part of the metabolic team and see children with a range of lysosomal storage disorders. My role is to assess and advise on feeding, swallowing and communication, in conjunction with the child's local Speech Therapist. I'll see children on the ward or in outpatients as necessary and I take part in the multidisciplinary MPS III clinic. I can also be contacted by telephone if parents or other people involved with children are concerned about their feeding. It's been an exciting new development for me to work as part of the team here, and I look forward to working with them over the next year.

Around the UK in 90 days...

GREAT ORMOND STREET HOSPITAL: 2 October 2008

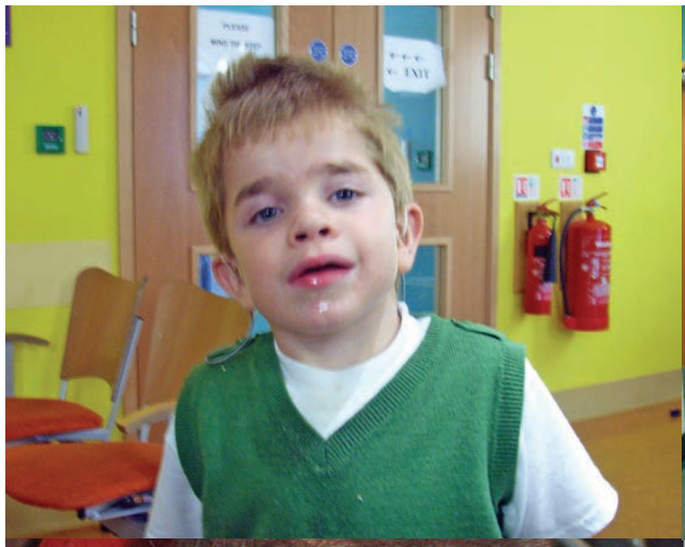
As usual the clinic was extremely lively and this kept the team of Dr Vellodi, Dr Cleary, Naimh, Lynn, Sonia and Michelle very busy! Ellen was on hand too. Ollie and Ben Illingworth enjoyed the play area particularly the cars and books and even found time to make a few new friends from another clinic! It was lovely to see Rajev Levy and Harrison King, they had both travelled a long distance to the clinic but still managed to raise a smile despite being stuck in traffic!

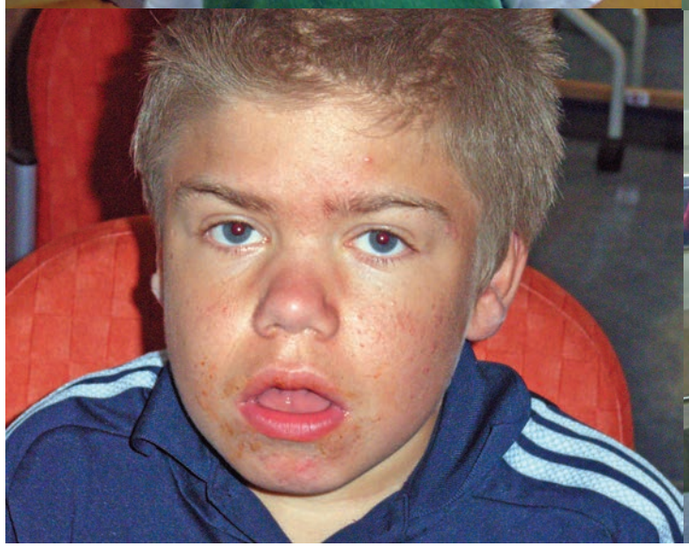
ROYAL MANCHESTER CHILDREN'S HOSPITAL: 10 Oct '08

Having survived the very early morning train journey, this busy under 5 years old clinic soon brought me back to life. It was so lovely to see all of the children and hear how many of them have recently started school. Stories were shared about new school friends and what everyone likes to do, playtime sounded the favourite! It also gave the parents an excuse to reminisce of their tales of school days too. Even some of the much younger children are enjoying playschool and they too have been making some new friends. With 10 MPS children attending the clinic, it almost ran to time.

ROYAL MANCHESTER CHILDREN'S HOSPITAL: 17 Oct '08

How lovely it was so see everyone today and watching all the boys playing together, giving their parents an opportunity to have a catch up and chat. Mr Loughlin managed to entertain his son Matthew as well as Steven O'Reilly thanks to the help of a football! The boys had quite a long rally going at one time and were really enjoying themselves in between their hospital appointments. Everyone was so pleased to see Isaac Turner who has very recently undergone surgery on his back, but we wouldn't have known as he was so busy socialising and swapping his mum's shopping secrets! We all now know where Louise does her shopping and what is in her trolley too! Isaac was busy talking to Jacob and occasionally in between listening Jacob was able to get a few words in! Bradley and Callum exchanged views on school and particularly uniforms. Callum was in high spirits even though he is due to have an operation on his hip the following week. Finally the clinic came to an end and all the boys left, the only evidence they had been there was the empty box which had earlier in the day had snacks and drinks in it for everyone.





BRISTOL MPS CLINIC: 4 November 2008

Another busy clinic with families arriving throughout the morning. It was lovely to see so many new faces. There were snacks and drinks available for the children, unfortunately we have not yet found a suitable space to offer teas and coffees for the parents! It was Nurse Sally's second clinic although she seems to have got a good understanding of the children and she was kept very busy throughout the morning. We look forward to the next Bristol Clinic.

BIRMINGHAM MPS CLINIC: 21 November 2008

What a full clinic list! Catherine and Satnam were both kept very busy throughout the day. This was my first Birmingham clinic and I was so pleased they were both available. During the day I saw so many children, some just wanted to play, some were hungry and one or two wanted to just go home! But a huge thank you to everyone I did see because you made me feel very welcome. Despite being a very full clinic it only over ran by 30 minutes!

NORTHERN IRELAND MPS CLINIC: 28 November 2008

Just rounding off this year's clinics throughout the UK, was Northern Ireland. It was a pleasure to finally meet all the families, as I have spoken to some frequently. Despite the bad weather I was able to arrive at the clinic only 15 minutes late. Food and beverages were laid on for everyone at the clinic, and there were some very hungry children and parents! With a full list of families arriving, the clinic even managed to run to time!

Thank you all again for making it such a lovely experience for me as this was my first visit to the Northern Ireland Clinic.

Linda Warner, Advocacy Officer

Here are a selection of photos taken at various clinics. Thank you to all who participated.

Childhood Wood Planting Day

On Friday 24th October 2008, the Society held its annual Tree Planting Day at the Childhood Wood. Six families attended the day to plant trees and release balloons with their own special messages.

We were very lucky with the weather as the sun was shining throughout and not a rain cloud in sight!

Some families had joined us earlier for a buffet lunch at the Clumber Park Hotel, whilst others decided to meet us in the Woods.

County Councillor Elle Loziak read a beautiful poem, which was followed by Paddy Tipping MP, reading the names of the family members and finally balloons were released in memory of their loved ones. Families were then able to take their time to plant the sapling trees and enjoy the tranquillity of Childhood Wood.

For my part it was a privilege to be allowed to share in this very special day. Here are a selection of photos from the day. **Linda Warner**

CHILDHOOD WOOD

MPS Adult Weekend

The MPS Adult Weekend took place on Saturday 29 and Sunday 30 November 2008.

The weekend kicked off on Saturday lunchtime as MPS individuals, volunteers and MPS staff met at the London Hilton Tower Bridge. Having had time to catch up with old friends, meet new ones and generally get to know each other, we headed off to Covent Garden to hit the shops! It was a little cold and it did rain slightly but this didn't dampen our spirits.

We headed to Brown's Restaurant in Covent Garden for an early dinner and then girls and boys split into two groups for the evening's entertainment. The boys went to see Avenue Q at a nearby theatre, whilst the girls intrepidly ventured across London to see Wicked. We then met back at the hotel for a nightcap and swapped stories about our evening.

The next morning was a morning of leisure, it was Sunday after all! We all met up for pizza at lunchtime then took a spin on the London Eye in the chilly, dusky afternoon before heading back to the warmth of the hotel to say our goodbyes.

The weekend was an enormous success and we are delighted to have received so many positive responses and words of thanks from so many of you. Here are a selection of photos from the weekend.
Antonia Anderson

'I am writing to thank you for inviting me on the adult weekend trip to London. I had a fantastic time and have to admit that seeing the theatre production 'Wicked' and travelling on the London Eye have been the highlights of my year. Going on the trip has given me the confidence to venture out on my own without the support of my family.' Lois Pack

Newcastle Christmas Party

On 7th December the MPS Society held a Christmas party at Newcastle United's football ground. We were lucky to be given a box which gave a great view of the pitch and I was reliably informed that Alan Shearer's box was only a few up from ours, so we must have been given a good spot.

The room was decorated with hats and crackers all sporting the Newcastle logo and gifts and we had our own personal bar and waiting staff. Once everyone had arrived and taken in the view we sat for lunch which was a Carvery style meal with a festive twist. While everyone was taking coffee Mr Lido set up his box of tricks ready to entertain the children as well as the adults. Mr Lido was fantastic and kept us all entertained with magic tricks which included a customary rabbit in a hat and two white doves.

The day was coming to an end but we still had one visitor to arrive. With a knock at the door and a loud Ho! Ho! Ho! Father Christmas entered the room bearing gifts for all the children, even the big ones.

The day ended on a high and a good day was had by all.
Sophie Thomas, Senior Advocacy Officer

EVENTS

Glasgow Christmas Gathering

It was cold, dark and foggy when I left home very early on Sunday 14 December to make the trip to Glasgow for the MPS Christmas gathering at the Hilton Hotel. Whilst Terminal 5 at Heathrow appeared to be functioning normally the weather unfortunately wasn't and we were grounded at the airport for two hours due to the fog! I finally managed to get to the Hilton with 10 minutes to spare, but I needn't have worried as Gary our entertainer for the afternoon was busy looking after the families.

After a hearty Christmas lunch of turkey with all the trimmings and a huge selection of desserts, we settled down to learn how to make a dog out of a balloon. According to Gary, the dog was to be a poodle, his certainly was but the ones our families were trying to make, left a lot to the imagination! The children did really well, following Gary's instruction in every detail, such a shame about some of the grown-ups understanding of how to make a poodle dog.

In between all the loud laughter I was finally able to judge the best balloon poodle. It was a tough decision as there were some excellent contenders and some rather unusual dogs with very short bodies, extra long legs and wobbly tails but after much deliberation we had a winner, Dean Mount. Gary went on to make him the most amazing colourful spider with huge eyes, long legs and a big head all out of balloons. I did wonder how the family were going to get the spider home without bursting any of the balloons.

Gary then entertained everyone with various magic tricks, from disappearing eggs to a Christmas tree inside a hat; it was truly a magical show. We were even treated to watching Liz McDowall having a hole drilled in her back and tummy and watched Gary pouring water through, just to prove there really was a hole there!

Although Gary did tell the children not to try this at home, in case they made a mess with the water on the floor! As the children helped Gary with his magic tricks they were given their own magic wands to keep. Who knows we may even have some future entertainers in the making?

No Christmas gathering would have been complete without a visit from Santa Claus and we heard he may have been nearby, so thought if we all sang Jingle Bells very loud he may just come and see us. We were very lucky as he must have heard our singing and sure enough he appeared. He spoke to all the children and gave them surprise presents and before saying his final Ho, Ho, Ho! Santa Claus asked all the children to remember to leave a carrot out for Rudolph on Christmas Eve, explaining that all the travelling makes him very hungry.

Before Gary left he presented all the children with a balloon-making kit, which may also give the grown ups a chance to improve on their model-making skills!

As the afternoon drew to a close and children were leaving with magic wands, poodles, balloon-making kits and also their present from Santa Claus, everyone said what a lovely time they had today. For my part, although a very busy and a long day, it was well worth it, as I reflected on my journey home. This time luckily there were no delays in Glasgow and no fog at Heathrow either!

I would like to say a huge thank you to all those who attended the Christmas gathering and made me feel so welcome. It was lovely to meet you all and spend time talking with you, it was a shame the day went so quickly.
Linda Warner Advocacy Support Officer

First Asia - Pacific Lysosomal Diseases Conference
12th National Australian MPS Society Conference
20 - 23 November 2008
Christchurch, New Zealand

This Conference marked the 25 years of the Australian MPS Society's operation throughout Australia and New Zealand and was hosted jointly with the Lysosomal Diseases Association New Zealand. This was the first time that the Australian National MPS Conference had been held outside of Australia, and is indicative of the global village that we all now operate within. David Oliver, President of the Australian MPS Society, in opening the Conference reiterated what I think we all agree: that even whilst the electronic era has brought with it an unparalleled ability to communicate quickly and easily throughout our countries, our continents and indeed around the world, there is still little substitute for the energy and connection that can be found in a physical gathering of like-minded people.

Of course Adelaide 2010 and the International Symposium on Mucopolysaccharide and related diseases is next year now. Momentum towards building another very successful International MPS meeting really took off in Christchurch not least because of Jenny Noble and Wendy Boon's ability to demonstrate through this meeting that they are capable of challenging the best of the best in organising International MPS meetings. The proof was there from beginning to end; the welcome ceremony with the Maori cultural experience, the three day conference programme and the childcare programme that even had a 'Fantastic Weather Programme' and wait for it, an alternative 'Wet Weather Programme'. I am pleased to say the sun shone and the 'Fantastic Weather Programme' went ahead as those of us from the UK including Dr Ed Wraith, Professor Bryan Winchester, Dr Jonathon Cooper, Barry Wilson and I could only imagine the pleasure of even having the need to plan a 'Fantastic Weather Programme' for our 2009 MPS Conference in the middle of June!

There was something for everyone in the Conference programme: the Brain and Central Nervous System; Existing Therapies; Emerging Therapies; Clinical Management; Newborn Screening and Access to Medicines.

The programme entry for the Gala Dinner stated 'Be prepared to have a fantastic evening!' We were not disappointed. Ko Tane, the Maori group, returned to entertain us with some wonderful singing and dancing. They even had the gala dinner guests on their feet participating. Some even took to the stage! As is becoming the practice on these occasions two presentations were made to acknowledge the work of two outstanding scientists - Prof John Hopwood from Adelaide Women and Children's Hospital and Prof Bob Jolly an eminent internationally recognised New Zealand veterinarian in the field of MPS animal models.

On behalf of the UK delegation I would like to offer our appreciation to the Australian MPS Society and Lysosomal Diseases Association New Zealand for their generous hospitality and wonderful meeting.
Christine Lavery, Chief Executive

Ladies and Gentlemen, good morning to you all. My name is Barry Wilson and I am Chairman of the UK MPS Society and have been so for almost eight years now. I am also the proud father of my wonderful 22 year old daughter Joanna, who has MPS I Scheie.

It is my immense pleasure to be here in Christchurch, indeed New Zealand. I last visited this wonderful country in 1962 whilst serving in the Royal Navy on board HMS Tiger. At that time I am sure a number of you were not even born. I was amazed by the generosity and affability of the people of New Zealand then, as indeed I am now. So, thank you to the Board of Directors of LDNZ and to Prof. David Sillence for inviting me here to take part in this International Consensus Meeting.

The UK MPS Society is now in its 26th year. It was founded by a lady that I know most of you know very well, Christine Lavery. Today, she is still 110% committed to the work that the Society carries out and pursues that dedication in the role of Chief Executive of the Society.

We Support over 1200 individuals and their families affected with a Mucopolysaccharide or related lysosomal storage disease. Over the last 24 years, we have funded in excess of £7 million of scientific and clinical research and I know there are a number of people here that could vouch for the benefits that that has brought.

And it is in the context of the MPS Society's commitment to research that it is my great pleasure to be here for this important expert meeting, of which the UK MPS Society has been pleased to be a major funder.

One of the key reasons for supporting this initiative is because the MPS Society considers it very important that advances in clinical management are peer reviewed, and it is the view of my Board of Trustees that this expert meeting will help to build a global understanding of the clinical benefits and limitations of the use of Bisphosphonates in the Mucolipidoses and other related lysosomal storage diseases to the benefit of those affected.

We, that is the world of lysosomal storage disease contributors and benefactors, have come a long way in the last twenty years or so. So much so, that through research and application of that research, so much is known about the various diseases that we can indulge in individual conferences and expert meetings targeting specific diseases or specific aspects of the diseases, like this one today, the 1st International Consensus Meeting for Bone Disease in Lysosomal Diseases.

I am going to let the experts get on with it now, so please enjoy the meeting and I hope we all take away from it a little more knowledge that will allow our global community to defeat these crippling and debilitating diseases. And on the way to that goal, facilitate the means to give all the patients the best possible quality of life that we can. Thank you.
Barry Wilson, Chairman of Trustees

Picture top left: Ron Law, Denise Law (past president), David Oliver (President of the Australian MPS Society), Christine Lavery and Barry Wilson.

1st International Consensus Meeting on Bisphosphonate Therapy in Oligosaccharidoses 19 - 20 November 2008 Chateau on the Park, Christchurch, New Zealand

This very important meeting, convened and hosted by Lysosomal Storage Diseases New Zealand with a grant from the UK MPS Society, was an exciting step forward for our families whose children, young adults and adults are affected by the Oligosaccharides, principally MLII and MLIII.

The first Mucolipidosis patients to be treated with IV Pamidronate started in New Zealand in 2000. This began the first ever study on bone disease in ML. A year later 6 Australian patients joined the study, and since then more than 30 patients worldwide are gaining benefits from Bisphosphonates. The successes have been varying with varying protocols being used. It is for this reason that this important meeting was called with the key objective of working to develop internationally accepted protocols and endpoints for the use of Bisphosphonates as a treatment for the Oligosaccharide group of diseases. The meeting also set out to look at what other options might be available to help manage the severe bone disease seen in this and other groups of Lysosomal Storage Diseases. A longer term hope is that ultimately perhaps we will see significant research taking place which will enable a better understanding of the bone pathology and musculoskeletal complications in LSD's.

Over 50 clinicians and scientists from the world over, experts in the field of LSD's and degenerative bone disease, presented their findings and were joined by a handful of expert parents to share their personal experiences of Pamidronate and enter into the debate.

Dr Ed Wraith told how the bone dysplasia seen in MLII and MLIII is not typical of classical dysostosis multiplex and advised that in severely affected patients bony lesions can be detected before birth with severe osteopenia, fractures and rickets-like changes characteristic of severe hyperparathyroidism can be seen. In MLIII the ball and socket joints of the shoulders and hips are usually severely affected.

Dr Grace Vizcarra-David gave the results of an Australian and New Zealand multicentre review of 25 cases of Mucolipidosis diagnosed over th past 30 years. The patients' phenotypes could be broadly grouped into MLII (15), MLII/III (5) and MLIII (5). A detailed analysis of clinical, radiographic and histologic features concluded that the skeletal pathology of ML was characterised by two forms of bone disease, Dysostosis Multiplex and Osteodystrophy. Six patients received treatment with Cyclic Intravenous Pamidronate 30mg/M^2 for periods of at least 12 months. After 12 months of therapy with Cyclic Intravenous Pamidronate, all the patients had decreased bone and joint pain, increased mobility and an increase on bone mineral density at several sites. One patient (MLII) experienced medication-related vomiting. Two patients continuing on therapy after 12 months had pathological fractures of the hip requiring cessation of bisphosphonate therapy and surgical fixation. This speaker concluded that Cyclic Intravenous Pamidronate reduces bone and joint pain, increases mobility and improves quality and bone density in patients with ML. She further stated that the issues surrounding the safety of long term use of bisphosphonates have yet to be resolved and continuing therapy is likely to result in Osteosclerosis.

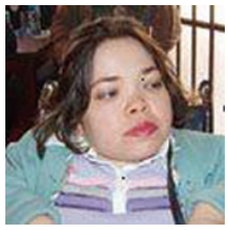
Sally Motomura, mother of Tetsuya (1984 - 2007) gave a summary of the disease course of MLII/III in Tetsuya whilst sharing with those present Tetsuya's enormous drive for life, determination and courage. She said that Teysuya was always very short stature, but didn't ever need surgeries and didn't complain much about pain. Tetsuya was intellectually very able, used his computer to most effect to communicate with others with ML and graduated from High School. At the age of 21 years Tetsuya started with Pamidronate therapy, which was discontinued after 2 infusions, due to marked deterioration in his cardiac function. It isn't clear whether this was caused by a misunderstanding about protocols, or simply because Teysuya's physical condition was already too far advanced.

Jenny Noble the mother of two adult children with MLIII shared her experiences of Pamidronate therapy. Jenny concluded that the use of sustained intravenous bisphosphonate therapy is important for the control of bone pain and secondary metabolic bone disease in patients with MLIII, but in the longer term suggested there is a greater need to work towards accepted protocols for its use, develop end points for treatment and look at other drugs that may give similar results to Bisphosphonates for the long term maintenance of secondary metabolic bone disease in Mucolipodosis.

This is just a brief summary of the meeting. Over the coming month all the clinicians, scientists and patient organisations involved will be collaborating to produce an internationally agreed Concensus Statement on the 'Use of Bisphosphonate Therapy in Oligosaccharidoses'.

Christine Lavery
Chief Executive

The Victory of Hope



I am Ayse Nihan Uguz. I was born on 10 February 1976 in Adana. This is a city in the southeast of Turkey, North of Cyprus and I still live there.

At the age of 6 I was diagnosed with Morquio Disease, MPS IVA. At that time there was no research into this condition. The doctors told us that they were just finding out about this disease and there was nothing that they could do for me. As a family we were in great shock as we did not know anything about it.

But I decided that things could not continue the way they were. Starting with the philosophy that life is beautiful regardless of everything else; I rolled up my sleeves and took up the path that brought my present day success. I became strong and determined. I had my family and friends who loved me. I thank my mother and father again for their support and encouragement.

I loved painting. During my high school years my interest for arts grew even stronger. After finishing my A levels, I attended Cukurova University Fine Arts Education Centre.

Now I am carrying on my work at the studio I jointly opened with three of my Art teacher friends.
Up until now I have had several personal exhibitions in several cities including Mainz in Germany.

One of my works was successful in a competition 'How do you look at life?' run by Genzyme, the pharmaceutical company and travelled around the world. I have also had an offer to exhibit at the Art Fabric Hotel in Wuppertal, Germany in September 2009. I am very happy and excited about this.

I wish all my MPS friends to be strong, to see the beautiful side of life and never to lose hope. We have so many wonderful and beautiful things around us. Please try to seek and see them. **Ayse Nihan** Uguz - MPS artist

Editor's Note: Fer Pidden, a UK MPS Society member, has sent in this article. Fer writes: I learnt about this young lady at the Morquio Conference in Northampton from an Irish family. I got in touch with them and we have been e-mailing and talking on the phone since then. I asked her to write a piece for the MPS magazine and translated it myself.

Natural History Study for Fucosidosis

A Natural History Study for Fucosidosis is being undertaken which will vastly contribute to the knowledge available about Fucosidosis, will be invaluable for newly-diagnosed families and their physicians, will provide information to assist with symptom management and is a necessary step towards an effective therapy.

The Natural History Study for Fucosidosis is an international collaboration between Professor John Hopwood at the Women's and Children's Hospital in Adelaide, South Australia; Dr Sara Cathey at the Greenwood Genetic Centre in South Carolina, USA; and Professor Micheal Beck at the Children's Hospital in Mainz, Germany.

The Study will enable families to contribute to better understanding of the progression and effects of Fucosidosis. Because it is so rare, little or no information is available about Fucosidosis, and compiling information about its course from infancy to adulthood is vital.

This study will include a gene mutation analysis, which can be conducted on already-harvested fibroblast cultures (skin samples which may already have been taken for diagnostic purposes and which may still be in storage) and where possible, urine samples.

Fucoside storage will be measured from these samples using mass spectroscopy. This biochemical information will be put together with any clinical information that may be available.

If you have an affected family member, or know of anyone with Fucosidosis, please pass this information on to them.

This study is an excellent opportunity to contribute to the information available concerning Fucosidosis. If you are interested in taking part please email Professor John Hopwood at john.hopwood@adelaide.edu.au

Three weeks ago I was awarded the New Zealand AMP Insurance company Premium award Scholarship for 2008, for my work in bone disease for Mucolipidosis and other LSD's. For more information visit www.amp.co.nz under 2008 Scholorship Recipients. The second part of this is that I also received funding to be able to include families in New Zealand and Australia to be entered into the Natural History study for Glycoprotein Storage Disease. I will be working with Dr Sara Cathy and Prof David Sillence on this initiative.

It is very exciting to see that in New Zealand Lysosomal Diseases are finally being acknowledged and having the ability to raise the focus for these very rare diseases through AMP is extremely humbling. I would just like to say thank you for all your support, guidance, encouragement, knowledge and patience that has helped this happen. Paul and I are thrilled for all LDNZ families in Australasia.

Jenny Noble
Secretary/Treasurer
Lysosomal Diseases New Zealand
www.ldnz.org.nz

WORLD* Symposium 2009

Presented by Lysosomal Disease Network

(*We're Organising Research on Lysosomal Diseases)

18 - 20 February 2009

The Westin Gaslamp Hotel, San Diego, CA

The Lysosomal Disease Network is a consortium of scientists, laboratories, healthcare professionals and clinics working as networked centres of excellence to improve basic knowledge and understanding of lysosomal disorders, improve diagnosis and advance therapeutic options for individuals affected by these disorders.

For more information:

www.lysosomaldiseasenetwork.org or
www.cmecourses.umn.edu

BioMarin Announces the Planned Retirement of Dr. Emil Kakkis, Chief Medical Officer

Novato, Calif., November 21, 2008 – BioMarin Pharmaceutical Inc. (Nasdaq: BMRN) today announced that Dr. Emil Kakkis, its Chief Medical Officer will retire from the company on February 27, 2009. Dr. Kakkis has decided to leave his position with the company to allow him to be able to devote more of his time to personal endeavors related to rare diseases. Dr. Kakkis will continue to be a consultant for the company for the foreseeable future.

"I am extremely grateful to have been part of this organisation over the last ten years and to have been part of developing three novel treatments for rare genetic diseases. I would like to thank all of the patients, doctors and, particularly, the BioMarin employees that have helped me in those efforts. I am looking forward to the opportunity to devote additional time to developing an institute for rare diseases and legislation to support ultra-orphan drug development," said Dr. Kakkis.

"Emil has been an invaluable part of the leadership team here at BioMarin and we are sorry to see him leave. He has been instrumental in the development of all of BioMarin's products, dating back to the very early work that he did on Aldurazyme while he was on the faculty of UCLA and before he was an employee of BioMarin. We are very fortunate to have had his service and fortunate that he has agreed to continue helping the company on a consulting basis. Emil has done an excellent job building up the R&D organisation, particularly in the past three years, and we have outstanding personnel in place to continue our innovation track and move forward with our current development plan," said Jean-Jacques Bienaimé, Chief Executive Officer of BioMarin.

Dr. Kakkis joined BioMarin in September 1998. Prior to joining BioMarin, he was an assistant professor at the Harbor-UCLA Medical Center, Division of Genetics, Department of Pediatrics. Together with his colleague Elizabeth F. Neufeld, Ph.D., of the University of California at Los Angeles (UCLA), Dr. Kakkis discovered how to produce a recombinant form of alpha-L-iduronidase (later to become known as Aldurazyme® [laronidase]), the enzyme which people with Mucopolysaccharidosis I (MPS I) are lacking. While at BioMarin, Dr. Kakkis was instrumental in guiding Aldurazyme, Naglazyme and Kuvan through development and regulatory approval. He also drove initial phases of development of PEG-PAL for PKU, BH4 for sickle cell disease, BH4 prodrug, and intrathecal enzyme therapy. He started with the company as a vice president in charge of the development of products for genetic diseases and since that time has come to lead the company's pre-clinical, clinical, regulatory and research organisations.

The company is initiating a search for a new Chief Medical Officer. It hopes to have a new Chief Medical Officer by the end of the first quarter of 2009.

BioMarin Announces Initiation of Clinical Assessment Program for Morquio A Syndrome

Novato, Calif, November 3, 2008 – BioMarin Pharmaceutical Inc. (Nasdaq: BMRN) announced today the initiation of the Morquio Clinical Assessment Program (MorCAP) for patients with the lysosomal storage disease Mucopolysaccharidosis Type IVA (MPS IVA), or Morquio A Syndrome. MorCAP is designed to augment available data on the disease by measuring endurance and respiratory function and other parameters in affected patients. BioMarin expects to follow the MorCAP program with a Phase 1b clinical trial of enzyme replacement therapy beginning in the first quarter of 2009. The primary objectives of the Phase 1b study will be to evaluate safety and to establish the optimal dose of enzyme based on pharmacokinetic and pharmacodynamic parameters.

"After successfully advancing two enzyme replacement therapies in approximately five years each from IND filing to FDA approval, we plan to leverage our clinical, manufacturing and regulatory expertise to develop a treatment for Morquio A syndrome," said Emil Kakkis, M.D., Ph.D., Chief Medical Officer of BioMarin. "Preliminary studies are promising and indicate that our drug candidate binds naturally to bone matrix and can adequately reach the growth cartilage after IV infusion. We recently have also shown that GALNS can reduce keratan sulfate storage in Morquio chondrocytes. This is important as the skeletal system is a primary concern in the treatment of this disease."

"We are excited to be the first site to enroll patients in the BioMarin Clinical Assessment Program for MPS IVA patients. This study is crucial to developing a deeper understanding of the clinical outcomes for this rare disorder, which will help lead to better disease management and therapy options, said Barbara Burton, M.D., Director, MPS/ML Treatment Program, Children's Memorial Hospital."

BioMarin has developed and manufactures two FDA-approved enzyme replacement therapies, one for the treatment of MPS I and one for the treatment of MPS VI. Naglazyme® (galsulfase) for MPS VI is wholly developed and commercialised by BioMarin.

Aldurazyme® (laronidase) for MPS I is manufactured by BioMarin and marketed by Genzyme Corporation.

About MPS IVA

Mucopolysaccharidosis IVA (MPS IVA, also known as Morquio A Syndrome) is a disease characterised by deficient activity of N-acetylgalactosamine-6-sulfatase (GALNS) causing excessive lysosomal storage of keratan sulfate (KS). This excessive storage causes a systemic skeletal dysplasia, short stature, and joint abnormalities, which limit mobility and endurance. Malformation of the thorax as well as macrophage storage in the lung likely impairs respiratory function and contributes to sinopulmonary infections. Odontoid hypoplasia and ligamentous laxity can commonly cause cervical spinal instability and potentially spinal cord compression. Other symptoms may include recurrent infections, hearing loss, corneal clouding, and heart valvular disease. Initial symptoms often become evident in the first five years of life. Depending on severity of the disease, age of diagnosis will vary. Many patients end up wheelchair dependent in their second decade of life and undergo numerous surgeries to manage their disease.

The rate of incidence of MPS IVA is as yet unconfirmed, but estimates vary between 1 in 200,000 live births to 1 in 300,000 live births. Approximately 400 patients worldwide are currently registered in a public registry, based on their publications. The prevalence of patients with MPS IVA appears substantially higher than that with MPS VI based on published reports.

Enzyme Replacement Therapy for MPS IVA in the UK

Three sites in the United Kingdom - Great Ormond Street Hospital, Royal Manchester Children's Hospital and Birmingham Children's Hospital have been recruited to enrol UK post MPS IVA patients into the BioMarin Clinical Assessment Programme (MORCAP).

Rare disease registries - a global community

Emma James DPhil (Oxon)
UK Registry Co-ordinator, Genzyme

Suyash Prasad MBBS MRCP MRCPCH MFPM
Director, Global Medical Affairs, Genzyme

The lysosomal storage disorders (LSDs) are progressive and often severe metabolic disorders that require therapy from various different healthcare professionals. In general, LSDs are very rare, occurring in around 1 in 40,000 individuals for Fabry disease,1 and 1 in 100,000 individuals for MPS I. (2,3) As a result, it can be difficult to identify enough patients in which to study the diseases and to test new treatments in clinical trials. Some medicines in these rare (or 'orphan') diseases are given licences based on results from few patients; however, to properly investigate the treatments, licensing authorities may request that disease registries are set-up to collect more information on the disease and its treatment over the longer term.

Registries are large databases that collect information on a wide population of individuals and evaluate care in the 'real life' setting. The aims are usually to determine the progression of a disease; establish if treatments are effective; monitor safety; and measure quality of life. There are increasing numbers of registries in existence, which investigate a broad spectrum of diseases, including neurological disorders, cardiology, and oncology, amongst others. Genzyme currently oversees four LSD Registries: one each for Fabry disease, MPS I, Gaucher disease, and Pompe disease. The data in the LSD Registries are governed by an independent group of physicians who have extensive scientific and clinical expertise in these diseases. They evaluate and report aggregated data to the medical and scientific communities through publications and presentations, and develop recommendations for monitoring patients to make a contribution to the understanding of the disease and optimise care. Additionally, data can be summarised to provide 'patient case reports' that can help doctors review the progress of individuals over time. As such, these databases of information can be used to directly benefit both the people who contribute their data and the wider medical community.

The participation of individuals affected by such conditions in these registries - from agreeing to their data being collected to completion of the quality-of-life questionnaires, which help determine the effects of diseases and treatments - is crucial. For registries to be effective, they need to include all eligible participants to avoid biases in the results, and certain information is required to be able to adjust outcomes for different risk and management factors. (4) There are, however, strict consent and privacy issues to ensure confidentiality, and data are anonymised by removal of obvious identifiers such as name or address when entered into the registries.

Registries collect longer-term data on a larger population of individuals than is possible in a clinical trial. In the LSDs, these data are crucial to establish disease characteristics, management, and outcomes with or without treatment. The Fabry Registry, (5) which was established in 2001, now has nearly 3000 enrolled patients globally, with over 250 of these from the UK. Meanwhile, the MPS I Registry, which has been running since 2003, has well over 700 patients globally, with an impressive proportion - over 130 - from the UK. Data from both patients who are and are not receiving treatment are equally important to enable the study of disease both with and without therapy and to maximise the benefits from the registry programme.

With the co-operation of consenting individuals, the registries are already contributing substantially to the knowledge about these rare disorders. Information that has been captured includes the characteristics of patients at diagnosis; (5,6) disease symptoms and progression; (5,7,8) and the long-term effects of different doses of enzyme replacement therapy. (9,10) For instance, although females with Fabry disease were previously thought to be 'carriers' and develop only minor symptoms of the disease, data from over 1000 women in the Fabry Registry supported studies demonstrating that female carriers of the disease may be severely affected with a substantially reduced quality-of-life. (7) Meanwhile, data over an 8-year period from the Gaucher Registry suggested that it may take several years of treatment

before any improvements in the bone symptoms of Gaucher disease are seen. (10) This type of information helps doctors to identify the disease earlier and to better manage patients, to create standards for disease monitoring, and to develop guidelines to ensure that patients get the best treatment possible.

Registries can contribute hugely across all disease areas, but they are particularly important for rare diseases such as Fabry and MPS I, where they provide a way to gather evidence and increase the growing body of information. The consent of individuals to have their data collected, however, underpins the registries - without their help and co-operation, this partnership between patients and the medical community will not succeed. Participation in these programmes means joining a collaborative global community that is fundamental to establishing the causes, signs, and symptoms of disease, and improving treatment for all.

References

1. Desnick RJ. Enzyme replacement therapy for Fabry disease: lessons from two alpha-galactosidase A orphan products and one FDA approval. Expert Opin Biol Ther 2004; 4: 1167-1176.
2. Wraith JE, Clarke LA, Beck M et al. Enzyme replacement therapy for mucopolysaccharidosis I: a randomized, double-blinded, placebo-controlled, multinational study of recombinant human alpha-L-iduronidase (Laronidase). J Pediatr 2004; 144: 581-588.
3. Moore D, Connock MJ, Wraith E, Lavery C. The prevalence of and survival in Mucopolysaccharidosis I: Hurler, Hurler-Scheie and Scheie syndromes in the UK. Orphanet J Rare Dis 2008; 3: 24.
4. Williamson OD, Cameron PA, McNeil JJ. Medical registry governance and patient privacy. MJA 2004; 181: 125-126.
5. Eng CM, Fletcher J, Wilcox WR et al. Fabry disease: baseline medical characteristics of a cohort of 1765 males and females in the Fabry Registry. J Inherit Metab Dis 2007; 30: 184-192.
6. Pastores GM, Arn P, Beck M et al. The MPS I Registry: design, methodology, and early findings of a global disease registry for monitoring patients with mucopolysaccharidosis Type I. Mol Genet Metab 2007; 91: 37-47.
7. Wilcox WR, Oliveira JP, Hopkin RJ et al. Females with Fabry disease frequently have major organ involvement: lessons from the Fabry Registry. Mol Genet Metab 2008; 93: 112-128.
8. Bodamer. Reporting on the natural history and treatment of MPS I through the MPS I Registry. WORLD Symposium, 13-15 February 2008.
9. Weinreb NJ, Charrow J, Andersson HC et al. Effectiveness of enzyme replacement therapy in 1028 patients with Type 1 Gaucher disease after 2 to 5 years of treatment: a report from the Gaucher registry. Am J Med 2002; 113: 112-119.
10. Wenstrup RJ, Kacena KA, Kaplan P et al. Effect of enzyme replacement therapy with imiglucerase on BMD in Type 1 Gaucher disease. J Bone Miner Res 2007; 22: 119-126.

Declaration of interest:
The authors are employed by Genzyme Therapeutics, Oxford, UK.

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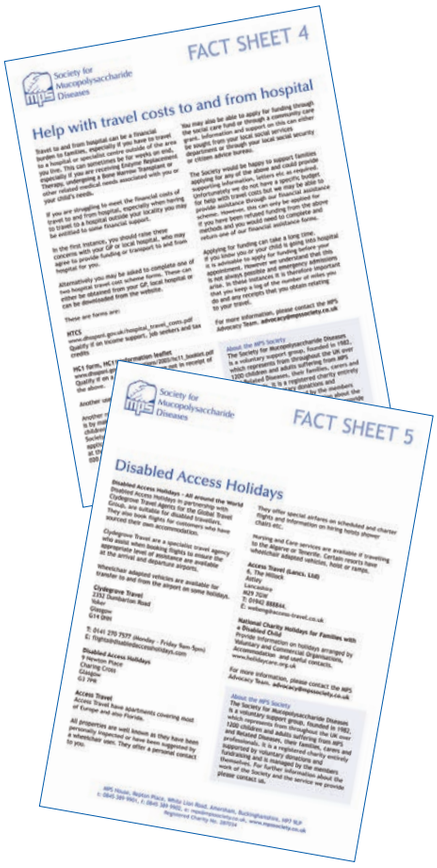
News from the EU

On 11 November 2008 the European Commission adopted a communication and a proposal for a European Council Recommendation on rare disease, setting out an overall community strategy to support member states in diagnosing, treating and caring for EU citizens with rare diseases.

European action will help patients, their families and professionals to collaborate across member states in order to share and co-ordinate expertise and information. Please visit **www.eurordis.org/article.php3?idarticle=1860**



MPS Advocacy Fact Sheets

The MPS Society produces a series of fact sheets giving further information on particular areas of support provided by the Advocacy Team. These are all downloadable in pdf format from our website **www.mpssociety.co.uk** from the Downloads page.

You may like to have a look at these if you haven't done so already as they could answer a number of questions or raise points that you hadn't previously considered.

Factsheet 1 Disability Living Allowance
Factsheet 2 Life Insurance
Factsheet 3 Travel Insurance
Factsheet 4 Hospital Travel Costs
Factsheet 5 Disabled Access Holidays
Factsheet 6 Carers Legal Rights
Factsheet 7 Carers Allowance
Factsheet 8 Education Maintenance Allowance
Factsheet 9 Wheelchairs Flights
Factsheet 10 Clinical Waste

For further information and to ask for support, please phone the advocacy team on 0845 389 9901 or email advocacy@mpssociety.co.uk.

International Alliance of Patients Organisations (IAPO) Launches Patient Safety Tools

IAPO has launched an advocacy toolkit for patient groups to inform and support their activities, advocating for improved patient safety. The Toolkit addressing patient safety is the first multi-issue resource for patient groups, providing the means for patients and patient's organisations to engage in the provision of healthcare and contribute to a quality and safe healthcare system.

The IAPO Advocacy Toolkit contains background information, facts, and figures on key patient safety issues identified by IAPO's 200 patient members as of immediate concern to them. These include medical error, taking medicines correctly, hospital acquired infections, Quality and Safety of Medicines and Clinical Trials.

The toolkit also provides detailed advice and tips on how to advocate and build partnerships to achieve patient safety goals and on communicating messages to patients and other health stakeholders.

For further information contact:
www.patientsorganizations.org
jeremiah@patientsorganizations.org

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

All Wales LSD Service

On 4 November the All Wales Inherited Metabolic and Lysosomal Disease Service was launched at the University of Wales Hospital, Cardiff. Christine Lavery, Chief Executive of the MPS Society attended the launch and reports on the new service for Welsh patients.

The launch was attended by clinicians, nurses, representatives from the Pharmaceutical Industry, representatives from patient organsiations, laboratory staff and healthcare commissioners. Dr Graham Shortland opened the service by outlining the services and facilities on offer at the University of Wales Hospital and gave examples of their experience in treating patients with lysosomal storage disorders. Dr Jeffery Carroll, Medical Director for Health Commission Wales gave an overview of the financial commitment that the Welsh Assembly has made to the development of this new service. This was followed by Christine Lavery, Chief Executive of the MPS Society who spoke on behalf of LSD patients outlining what patients and their families expect to see from specialist service. Dr Chris Hendrikz, Consultant Metabolic Physician at Birmingham Children's Hospital gave a comprehensive presentation on infantile and adult Pompe disease and Dr Atul Mehta, Clinical Director of the LSD Unit at the Royal Free Hospital, London spoke about Fabry disease.

The establishment of a new commissioned clinical service for the diagnosis, assessment and treatment of adult and paediatric patients with inherited metabolic and lysosomal storage disorders will consist of a Lead Clinician (Dr Graham Shortland), Consultant Biochemist (Dr Mike Badminton), Nurse Specialist (Andrew Dobson), Specialist Pharmacist (Zoe Taylor), Dieticians (Angharad Banner and Kath Singleton), Physiotherapist (Ann Baldwin), Clinical Psychologist (Bethan Phillips) and support services.

Currently most Welsh Lysosomal Storage Disorder (LSD) patients are seen at one of the seven LSD centres of excellence in the England. With the launch of this new service all Welsh patients will be offered the opportunity to transfer their care to the new service. Patients who wish to continue being seen at one of the seven centres in England will be able to do this on a shared care basis, where Dr Shortland and his team will be integral in their clinical management and treatment decisions.

The All Wales service will use the current National Commissioning Group (NCG) guidelines used at the seven LSD centres of excellence in England when assessing patients for treatment.

New UK LSD Collaborative Group Established

Representatives from UK Lysosomal Storage Disorder Patient Organisations have come together to join forces to become a strong lobbying and action group for LSD patients and their families in the UK. The group is made up of patient representatives from the Gauchers Association, The Society for Mucopolysaccharide Diseases (the MPS Society), Battens Disease Family Association, Niemann-Pick Group (UK) and the Pompe Association, reports Tanya Collin-Histed, Executive Director of the UK Gauchers Association;

The group first met in January 2007 to discuss common issues such as working with the Pharmaceutical Industry, the development of homecare services for patients, new born screening, the development of metabolic networks in the UK, the need for research into the brain and representation on the Health Technology Assessment longitudinal study into enzyme replacement therapy for LSDs.

The group meet 3 times a year and are in the process of developing 'Terms of Reference' and a work programme for the next 12 months. Future activities for the group will include; Transition Planning for LSD patients, the development of a patient group for MLD, a stand at the 2009 Party conferences to lobby for ongoing specialised service status for LSDs, a meeting with the Shadow Health Secretary pre 2012 when the current set up for LSDs in England will come to an end and ongoing representation on the HTA study into ERT for LSDs.

We are keen to open membership of the group up to other LSDs group, if you would like more information on the group please contact; Tanya Collin-Histed on: 00 44 1453 549231 or e-mail: ga@gaucher.org.uk

At the Childhood Wood planting in October 2008, Jenny Mellors who was there representing the Lord Lieutenant of Nottingham's office, told me about a very special book she had been made aware of concerning bereavement, especially for younger children. The book is ***The Next Place*** by Warren Hanson. The author describes the book as an inspirational journey of light and hope to a place where earthly hurts are left behind.

'The next place that I go will be as peaceful and familiar as a sleepy summer Sunday and a sweet untroubled mind. And yet... it won't be anything like any place I've ever been... or seen... or even dreamed of in the place I leave behind."

The book is colourful and beautifully illustrated and I found it very moving in the simplicity of the telling of its story. **Wilma Robins, MPS Trustee**

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