MPS

Society for Mucopolysaccharide Diseases

Summer/Autumn 2009

What's Inside...

Photos and feedback from our recent events including MPS Weekend Conference 2009 and MPS Sibling Weekend

... plus the LATEST in research, treatment, news and views

The MPS Society

Founded in 1982, the Society for Mucopolysaccharide Diseases (the MPS Society) is the only national charity specialising in MPS and Related Diseases in the UK, representing and supporting over 1200 affected children and adults, their families, carers and professionals. The MPS Society:

Acts as a **support network** for those affected by MPS and Related Diseases

Brings about more **public awareness** of MPS and Related Diseases

Promotes and supports **research** into MPS and Related Diseases

MPS & Related Diseases

Mucopolysaccharide (MPS) and Related Diseases affect 1:25,000 live births in the United Kingdom. One baby born every eight days in the UK is 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.

At present there is no cure for these devastating diseases, only treatment for the symptoms as they arise.

Where does your money go?

A donation of *£2 per month* could help us to offer so much more support in so many ways:

Access to clinical management and palliative care

MPS Regional Specialist clinics

Support with disability benefits

Paving a child's way in accessing education

Upholding rights in employment

Advising on home adaptations

Bereavement support

Please donate to
www.mpssociety.co.uk,
phone 0845 389 9901
or post your donation to our office,
MPS House.

Front cover photo:
MPS Sibling Weekend

Society for Mucopolysaccharide Diseases

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Magazine Deadlines

Winter	1 Dec 2009	Spring	1 Mar 2010
Summer	1 Jun 2010	Autumn	1 Sep 2010

Friend of MPS

Become a Friend of MPS to receive the Society's magazine and fundraising newsletter plus a range of other benefits. Contact us for more information.

IN THIS ISSUE...

Summer/Autumn2009

Welcome to a bumper edition of the MPS Magazine. This is a combined Summer/Autumn edition as the editor has recently been on maternity leave. The deadline for the Winter 2009 edition is 1st December 2009.
newsletter@mpssociety.co.uk

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CHIEF EXECUTIVE'S REPORT

It is one of the most rewarding parts of my role to report to our members upcoming clinical trials particularly when these are for diseases where there has been a void of therapeutic treatment since the diseases were first described.

As you will read in this magazine, the first Enzyme Replacement Therapy for MPS IVA, Morquio disease, was given at Birmingham Children's Hospital to Sultan Ali as part of the first clinical trial of its kind for this disease. For Victoria Vickery, the second child to receive ERT for Morquio disease, it was a double celebration. It was Victoria's birthday and as she sat through her first infusion at Great Ormond Street Children's Hospital she had the distraction of eating her way through a chocolate birthday cake provided by the nurses. The third UK clinical trial centre for Morquio A Disease is the Royal Manchester Children's Hospital who have also now finished recruiting patients.

It is also hoped that a one year natural history study will also soon be underway for MPS IIIA, Sanfilippo disease at the Royal Manchester Children's Hospital and more information on this study will be given in future MPS Magazines.

In this difficult economic climate, I am even more aware of demonstrating the value to our members and the global MPS community of the Society's presence at overseas meetings. These meetings provide a unique opportunity to hear the perspectives of Fabry, MPS and related disease specialists from different parts of the world, network with the pharmaceutical industry, learn from them and share our experiences. I would like to take this opportunity to thank the industry and global patient organisations who have supported my attendance at overseas meetings and invited me to speak.

In the last magazine the Society's Chairman sent out a message encouraging our members to 'THINK MPS' when it comes to fundraising or donations. On behalf of the Board of Trustees I would like to take this opportunity to thank all our members, their families and friends, who have supported the MPS Society these past few months. Without a doubt, achieving the income needed to maintain the current level of services is going to be an ongoing challenge well into 2010 and beyond. Not only are our members rallying but so are the staff team. On 9 August 2009, our Finance Officer, Gina Page; Senior Advocacy Officer, Sophie Thomas and Advocacy Officer, Jolanta Turz, jumped out of an aeroplane for MPS. This was no mean task and I would like to thank all our members who recognised their efforts by making a donation.

All we can ask is that through these challenging times if a fundraising or donation opportunity presents itself that you 'THINK MPS'. In the meantime, the whole team at MPS are here to support you.

Christine Lavery
Chief Executive

Become a Friend of MPS

Would you like to show your support by becoming a Friend of MPS? We would welcome relatives, friends, overseas MPS families, professionals or indeed anyone interested in the work of the Society or the field of MPS and Related Diseases.

This would encourage us, help us plan for the future and bring about more public awareness for this group of rare, genetic, life-limiting diseases. You can also keep up to date with the latest information, news and stories.

Visit **www.mpssociety.co.uk** to download the application or phone us now on **0845 389 9901**.

Highlights from the Management Committee

The Society's Board of Trustees meet regularly. Here is a summary of the key issues that were discussed and agreed at the Management Committee Meetings held 28 - 29 March and 22 - 23 May 2009.

Personnel

Following two rounds of interviews Trustees were informed that Kate Barker has been appointed Fundraising Officer with responsibility for Charitable Trust and Corporate Donations.

Governance

In May the Trustees reviewed 43 of the Society's 63 policies making changes as appropriate. Sue Cotterell's Health and Safety report was presented to the Trustees. The risk registered was tabled. Trustees considered eight items where work is continuing to mitigate these risks.

Treasurer's Report

Judith Evans presented the figures from the MPS bank accounts. The Treasurer stated that the financial situation was challenging as receipts were down and despite streamlining there are regular payments still to be made. Trustees noted that the income received by MPS from the 2008 Jeans for Genes day was £100,000 compared with over £400,000 three years ago.

Generating Income

The Trustees acknowledged the considerable success of Monica Hartwell, Fundraising Consultant over the last three years. As Monica moved on to make way for our full time Charitable Trust and Corporate Fundraiser, Kate Barker, she went out on a high securing a three year Department of Health grant and a one year BBC Children in Need grant.

Support to Members

Sue Cotterell continues to support Sarah Long with the Tipping the Lens project for Morquio A disease.

Trustees were advised of the MorCAP study; ERT for MPS IVA underway in London, Manchester and Birmingham. MPS has made at least one visit to each site and on 21 April 2009 Christine Lavery and Jolanta Turz were present when the first ERT infusion for MPS IVA was administered to Sultan Ali.

MPS Research Grants

At their March meeting the Trustees agreed a grant of $NZ 7000 to Prof. Bob Jolly for the purpose of identifying the physiological principles underlying intrathecal enzyme replacement therapy in LSDs. This project is funded jointly by the MPS Society and Lysosomal Storage Diseases New Zealand.

ANNUAL GENERAL MEETING 2009

The annual general meeting of the Society for Mucopolysaccharide Diseases took place at the Northampton Hilton Hotel at 9 a.m. on Sunday 28th June 2009. 8 Trustees and and another 47 members were present at the AGM.

The Chairman, Barry Wilson, opened the meeting and welcomed those present. The Chairman stated that apologies had been received from Judy Holroyd, Faith Parrott and Peter Conlin.

The minutes of the Annual General Meeting held on Saturday 3rd May 2008 were published in the Summer 2008 edition of the MPS Magazine, and were distributed in advance to those members present at the meeting. The minutes were accepted as true and accurate.

The Chairman presented the Trustees' Report. This is published in the MPS Annual Report and Accounts for the year ending 31 October 2008.

The Treasurer, Judith Evans, presented the Statement of Accounts for the financial year ending 31 October 2008, the details of which are also to be found in the Society's latest annual report. It was proposed and seconded that the auditors McLintocks, Chartered Accountants, Chester, be appointed the Society's auditors for the financial year ending 31 October 2009.

Four Trustees had been nominated for re-election. As there were four places vacant, a vote was not necessary and Bob Devine, Faith Parrott, Bob Stevens and Tim Summerton were elected. This was proposed and seconded by members present.

Under AOB, Dr Fiona Stewart thanked the MPS staff team for the organisation of the conference, and thanked the volunteers for their work with the children during the conference. The Chairman reiterated the need to raise funds and said that the Society appreciated the efforts of those who had fundraised. He also stated that the UK MPS Society is one of the most respected MPS Societies in the global community.

The Chairman thanked everyone for attending and the meeting closed at 9.21 a.m.

Barry Wilson, Chairman of Trustees
On behalf of the Management Committee
28 June 2009

How your money helps...

More professional support for more MPS Families

MPS Advocacy Workers offer a whole range of services to help children and adults living with Fabry, Mucopolysaccharide and related diseases and support their families. We are there at the time of diagnosis and offer support for as long as we are needed. A donation of £2 per month could help us to offer so much more support in so many ways.

Access to expert clinical management & palliative care
MPS Regional Specialist clinics
Support with disability benefits
Paving a child's way in accessing education
Upholding rights in employment
Advising on home adaptations
Bereavement support

More MPS advocacy workers

You'll be helping to fund more advocacy workers that are so crucial to empowering children and adults living with MPS and related diseases and their families through the information, advice and advocacy they provide.

More vital information

Your donation could help us to have more trained advisors running our MPS Helpline at the MPS Society's national resource centre. One child born every eight days in the United Kingdom will be diagnosed with an MPS or related disease.

More help to cope with the isolation of a rare disease

The chances are you have never heard of Mucopolysaccharde diseases, Mucolipidosis or Fabry disease. The truth is most of the families we support have never heard of these diseases either. That is why they need your help to enable MPS to provide national and regional family conferences, activity weekends for siblings, young adult weekends for those affected and run the MPS befriending scheme.

More noise to force through change

The MPS Society is already recognised for punching above its weight to achieve improved clinical care for all those affected, over half of whom will lose their lives in childhood. We campaign for change, we fight to eradicate discrimination and we aim to ensure that all affected children and adults get the health and social care whoever and wherever they are.

More help

Even if you don't know anyone living or dying with Fabry disease, a Mucopolysaccharide or a related Lysosomal disease your help is vital and enables us to help over 1200 affected families in the United Kingdom.

For more information, to seek support and advice from our advocacy team, or to help raise funds so we can continue our work, contact us now!

0845 389 9901 **mps@mpssociety.co.uk**

Meet our new Trust and Corporate Fundraiser...

Hi! My name is Kate Barker and I joined the MPS Society's team at the beginning of April 2009 as the charity's first full-time fundraiser.

My background is in journalism and fundraising/communications, and I have five years' experience of successfully raising funds for another small charity.

I'm a bit of a workaholic but, when I can be dragged away from the office, I love salsa dancing (and anything Latin, including the music, the food and the sunshine). I also enjoy hiking and photography, and have just taken up belly dancing!

I have been really inspired by all the moving stories of families living with MPS and am enjoying the challenge of making grant applications that will hopefully keep the money coming in to fund all the Society's vital work.

Kate Barker
k.barker@mpssociety.co.uk

The 13,000 foot MPS staff skydive!

We decided it was time we did something to raise some money for the Society instead of keep asking you. So the only thing we could agree on was a Tandem Parachute Jump.

9th August arrived, Sophie, Jolanta and I set out at 6.45am to RAF Weston-on-Green, near Bicester, it was a lovely bright warm morning. We had to register at 8am, they suggested we lined our stomach with some food, but of course we knew best, no food. The three of us were quiet, not sure what we were all thinking. We were all calm, the instructors said "this was the worst type, they normally freak at the aeroplane door."

Family and friends arrived to offer their support, we had to wait to be called. There was a small café on site with hot drinks and the smell of bacon cooking! We spotted the plane that would take us up, it was smaller than their usual plane as it only held four jumpers and the pilot. Then our names were called, we had a twenty minute brief and had to wait again till our names were called on the loud speaker.

We were relieved to see our names on the second flight. No time to get nervous. We disappeared into the hanger and there we were met by our instructors, Russ, Ed and Dunc. We got suited up and then our harnesses were put on. It was rather uncomfortable and not very glamorous. We then walked to the bus that would take us to the plane.

We entered the plane and it took off, as we were climbing the instructors were showing us how high we were. All I could see were clouds and we saw a plane full of holiday travellers. As we were climbing we had to arrange ourselves ready to go, we were asked to put our hands on the instructors knees, lift ourselves up and basically sit on their laps. It was all very up close and personal.

We reached our target height of 13,000ft, I am not sure where the other 3,000ft come from. The door opened and you heard the sound of the engines, wind and you felt the cold air. The adrenalin had taken over by then and you could not feel the cold. Sophie was first to go, I was asked to twist my legs towards the door, and Sophie had gone, it was like she was sucked up by a monster. I cannot remember how I got to the door, the next thing I remember is hanging, not in the plane, not out the plane. We had to tuck our legs back under the plane, arch our back and head back on to the instructor, all you could see was clouds. Then we threw ourselves out of the plane. We were falling at 120mph, falling through the clouds. The noise was unbelievable, it was amazing and indescribable. We fell for around 6000ft and then the parachute went up. What a relief! The silence was immense, the view was amazing. Once things were settled down we did some acrobatic turns, you could hear a pin drop. On landing we had to lift our legs up and let the instructor land. Three perfect landings.

Would I do it again, same time same place next year.
Gina Page

You can still sponsor Gina, Sophie and Jolanta by logging on to http://www.justgiving.co.uk/sophie-gina-jolanta/

Members' Announcements

New Members

Mr and Mrs Malcolm have recently been in contact with the Society. Their son Jack had a diagnosis of Hurler Disease. Sadly Jack passed away after a successful bone marrow transplant on 25 November 2008 aged 19 months. The family live in Scotland.

Ms Amanda Hughes has recently been in contact with the Society. Her son Zack has a diagnosis of Hunter Disease. Zack is two years old. The family are living in East Anglia.

Teresa and Shane have recently been in contact with the Society. Their son, Corey, has been diagnosed with Hurler Disease. Corey is 9 months old. The family live in South Devon.

Mr Darrin Minett has recently been in contact with the Society. Darrin has a diagnosis of Fabry disease and is 43 years old. He lives in the Midlands with his family.

Mr A Gillan has recently been in contact with the Society. Andrew has a diagnosis of Fabry disease. Andrew lives in Northern Ireland.

Mr Martin Beecroft has recently been in contact with the Society. Martin has a diagnosis of Fabry Disease. Martin is 49 years old and lives in East Anglia.

Caroline Shuttleworth has recently been in contact with the Society. Her son George has been diagnosed with Hunter Disease. The family live in Manchester. George was 2 years old in March and has received ERT since September 2008.

Mrs Angela Watson has recently been in contact with the Society. Angela has a diagnosis of Fabry Disease. Angela is 51 years old and lives in the East Midlands.

Edgard and Rachel Zaldua have recently been in contact with the Society. Daniel has a diagnosis of Sanfilippo Disease. Daniel is four years old and the family live in the South West.

Deaths

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

Stephen Young who died on 24 January 2009 aged 13 years. Stephen suffered from Hunter disease.

Tariq Mahmood who died on 7 June 2009 aged 14 years. Tariq suffered from Morquio disease.

Pritika Silhi who died on 9 July 2009 aged 28 years. Pritika suffered from Scheie disease.

Robyn Watterson who died on 18 July 2009 aged 12 years. Robyn suffered from Hurler disease.

Jibram Shoukat who died on 1 September 2009 aged 23 years. Jibram suffered from Morquio disease.

Do you have a story to share?
Please email
newsletter@mpssociety.co.uk
or phone **0845 389 9901**

Changing the world for children with genetic disorders

Friday 2 October 2009
Jeans for Genes Day

Congratulations to Ken and Pam Ballard who celebrated their 60th Wedding Anniversary on 17 September 2009.

Pam and Ken have been volunteering and fundraising for the MPS Society since it was founded in 1982. Their first grandchild Simon Lavery had MPS II, Hunter disease and died that year aged 7 years.

For the last 27 years Pam and Ken have given their time to pack the MPS Magazine. They have asked for donations to the MPS Society in lieu of gifts for their 60th Anniversary.

Mark your calendars and hop down for the
11th International Symposium on Mucopolysaccharide and Related Diseases
Adelaide, South Australia, 23 – 27 June 2010
www.mps2010.com.au

Mucopolysaccharide and Related Diseases Society Aust. Ltd., Lysosomal Diseases Australia and Lysosomal Diseases New Zealand warmly invite you to join them in Adelaide,
23 – 27 June 2010 for the 11th International Symposium on MPS and Related Diseases.

The scientific and family programmes will be exciting and relevant with a focus on the areas of newborn screening, prognostics, understanding pathology and therapeutic options. Genuine opportunities for thorough discussion and debate will be a feature of the program.

Adelaide is a city surrounded by parklands, sports fields, a top class golf course, walking and cycling tracks and beautiful gardens.

We hope you will hop on down under and join us for five exciting days of cutting edge science, exciting family experiences and an enjoyable cultural experience.

Daryll celebrates his 40th birthday!

Hi Everyone, my name is Carol Westland and on Saturday 17 January 2009 we proudly celebrated the 40th birthday of my eldest son Daryll who suffers from Sanfilippo Disease, MPS III.

Despite a horrid windy and rainy evening, over 50 guests came to his party held in a local hall just across the road from his childhood home where he had enjoyed his early life and the freedom of the garden. Relatives, carers and friends of many years helped to make the evening a great success.

We were entertained by a disco and a folk trio - who were friends of his late Dad. They sang 'Happy Birthday' and played other music close to him which caught his attention.

Although it was a very special evening the fact that he couldn't enjoy his birthday cake as he had a PEG fitted 8 years ago made me feel sad as he enjoyed his food so much when he was younger, but through the evening he managed a few smiles and that meant so much to us all. Daryl will never know how much he is loved or how proud so many people are to know him. **Carol Westland**

Many congratulations to Daryll from all at the MPS Office!

IN MEMORY

Kym and Dwain Caines

On 27 May 2006, Kym Caines, aged 22 years, passed away peacefully. Kym was one of eight children to Pauline and Simon and was sadly missed by her brothers and sisters, Donna, Aaron, Dwain, Ben, Sadie, Cye and T-jay.

On 4 April 2009 Kym's brother Dwain Caines, aged 23 years, passed away. Both had Sanfilippo A.

Due to the complicated symptoms of their disease, our mother Pauline made the brave choice for Dwain to undergo a post mortem to enable genetic specialists to research the disease, the possible related illnesses to the disease and ways of enhancing the quality of life for others with Sanfilippo.

Whilst most people may not want to talk about this and may not even consider this as its their child, I feel it is important to both Kym and Dwain and our mother Pauline for making such a brave decision that will help medical and research purposes and find future ways of treating children and young adults with Sanfilippo.

Kym and Dwain touched the hearts and left a lasting legacy to all whom knew them.

Our family will never be the same again but we are thankful our angels have each other in their little garden of heaven. So until we are together again as a family, I would like to dedicate this poem to Dwain and send love, hugs and kisses, and will always be thinking of you.

Mum, Dad, myself Donna, and all your brothers and sister. We miss you and love you both.

The photos below are of Kym (left) and Dwain (right).

Our Angel Dwain

God sent down from heaven
An angel from above
He blessed our days with happiness
He filled our hearts with love.

He gave us this child
But then he did say
We could only have him for a while
He must return to heaven some day.

This angel was so special
He was warmer than the sun
He shined more than the stars
He brought us lots of fun.

This boy shared lots of kisses
And lots of cuddles too
He relished in the good times
He braved right through the blue.

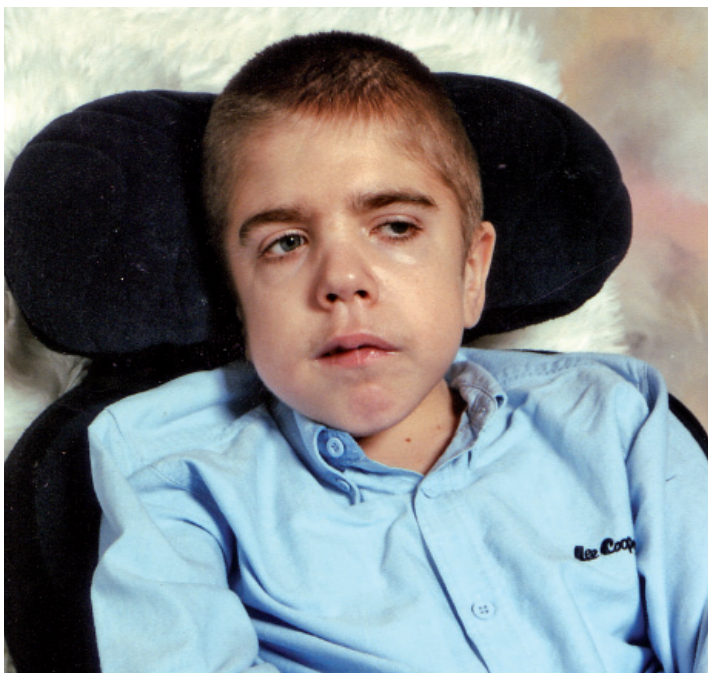
Then a little unaware
And a little by surprise
God came down one day
And closed the angel's eyes.

Our angel had a calling
From his sister in the clouds
To say that he could join her
And he had done her proud.

This angel was our world
This angel blessed our heart
This angel's time had ended on earth
This angel must depart.

It's hard to let you go
It's hard to say goodbye
But angels - they live forever
Angels never really die.

So thank God for this angel
Who we will meet again
And who is loved so very much
Our special boy - Dwain.



In Memory of Sarah Cupitt
by her Mum, Diane Reynolds

My daughter was born on 16 November 1996 and she passed away on 27 January 2009. My daughter Sarah was a precious little princess who touched the hearts of everyone who met her. My daughter Sarah's life was cut short by a terrible illness called Sanfilippo Disease.

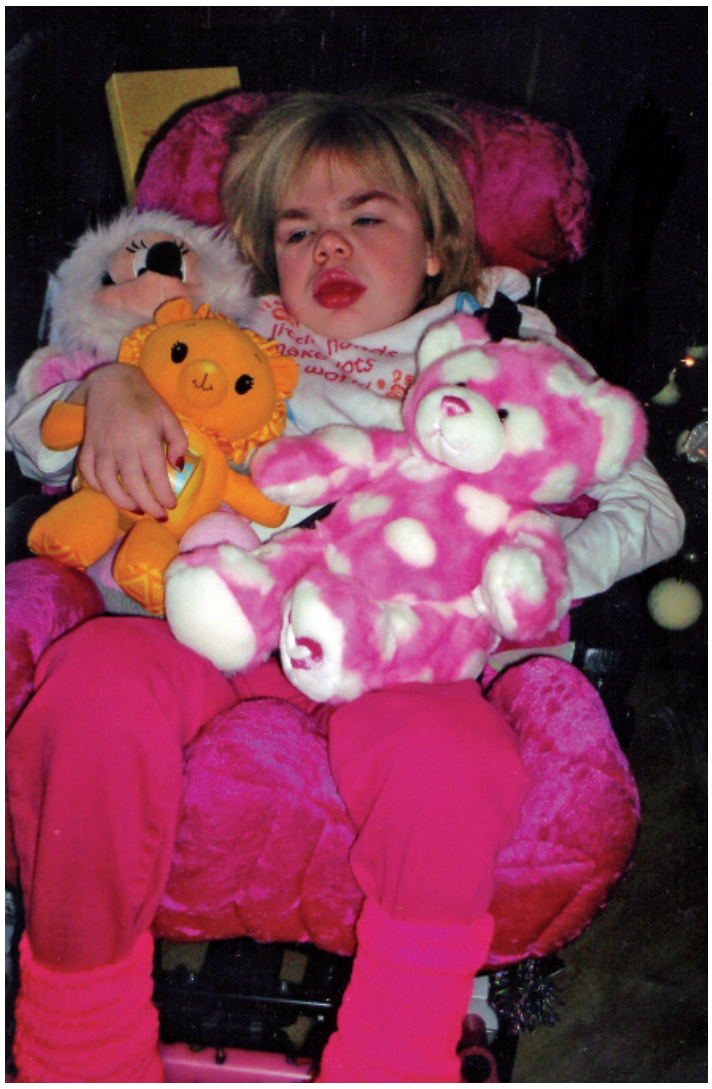
She was taken from me so suddenly. Sarah enjoyed her life to the full. Myself and her step dad Anthony took her everywhere we could. It became a regular family outing. She loved dancing with her sister Lisa. Her brother Luke loved her so much and he misses her deeply. He used to watch DVDs with her on her bed. We took Sarah and the other children to the seaside. She really enjoyed spending the day at Woburn Safari Park where she stroked the kangaroos. She loved it there. We all went to Sherwood Forest Farm where my daughter loved stroking the rabbits.

When my daughter was happy, we all were. I would always read to her and sing to her. I would rub her hands and feet as sometimes they were very cold with her condition. She always watched videos such as Tweenies, Fimbles and Shrek. As her condition got worse I would feed her, dress her and shower her. Everything we did revolved around my daughter. Happiness from the moment she woke in the morning.

My life without Sarah is very hard. We all miss her so deeply. She will always be in our hearts forever. Her stepdad was much loved by her. She loved her Mummy very much and her brothers, Luke, Ashley and sister Lisa too. Her grandad Norman used to play with her too and look after her when my husband and I went out for a while. My brother Andy, Sarah's uncle, used to play with her and feed her. She loved her uncle to bits. Her auntie Hayley used to feed her too. Her stepdad was a brilliant father to her. He would carry her everywhere. We all loved Sarah. She was our darling princess.

Sarah also used to go out with a linkworker called Marlin. She was a special lady. Marlin took to Sarah and Sarah loved her so much. Marlin took Sarah for meals out in the town. Sarah was very happy when she went out in the town. When she was little we took her to Great Yarmouth in the caravan. Her stepdad cooked tea and gave Sarah chips whilst her stepdad and I had pie and chips. Sarah made me laugh because she took the pie off her stepdad's plate and started to eat it. It was so funny the way she did it.

We all miss her to bits and just wish she was still here. She will always be my little girl, Sarah, and she's my special princess. Love Mummy Diane, Stepdad Anthony, brothers Luke and Ashley and sister Lisa.



WHAT'S ON 2009 -2010!

MPS CLINICS

16 Oct '09	Manchester BMT Clinic (under 6)
23 Oct '09	Manchester BMT Clinic (over 6)
10 Nov '09	Bristol MPS Clinic
20 Nov '09	Birmingham MPS Clinic
11 Dec '09	Northern Ireland MPS Clinic
TBC	Cardiff MPS Clinic

REGIONAL EVENTS

9-12 Oct '09	Scottish Short Break
23 Oct '09	Childhood Wood Planting
29 Nov '09	Glasgow Christmas Party
4 Dec '09	Birmingham Christmas Party (TBC)
March '10	Young Adult Weekend (TBC)
12-16 April '10	Sibling Weekend (TBC)

CONFERENCE EVENTS

27-29 Nov '09	Fabry Patient Meeting, Vienna
17-20 Dec '09	MPS Disney Conference
14-16 May '10	N. Ireland Conference (TBC)
29-31 May '10	Family Weekend & AGM, Camelot
23-27 June '10	International MPS Conference Adelaide

Swine Flu

The MPS Society has been closely monitoring the developments across the nation and the world regarding the spread of and containment of Swine Influenza. At the end of the MPS Conference one of the children was medically suspected of being affected by Swine Flu. This was subsequently confirmed by swabbing and up to eight others are believed to have also contracted Swine Flu. Thankfully all those affected have made an uneventful recovery.

However this is a timely reminder that individuals with MPS, Fabry and related diseases need to be extra careful as they may be at a higher risk for complications from this or any other infection. I would like to remind families about the importance of prevention of infection, including hand-washing. It is important to remember that individuals with MPS, Fabry and related diseases may have compromised respiratory systems, and it is more difficult for these individuals to fight infections such as Swine Flu.

Christine Lavery ***Chief Executive***
c.lavery@mpsssociety.co.uk

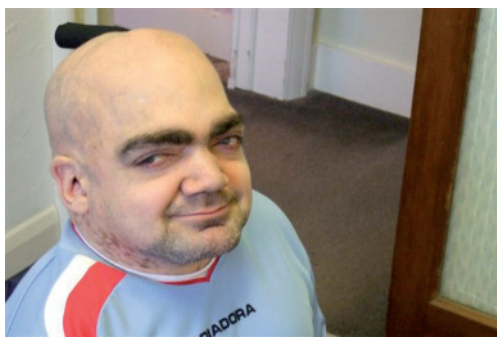
Disney MPS Family Conference

17th – 20th December 2009

The National MPS Society in the United States will celebrate its 25th Anniversary during the opening evening of the Disney Conference on 17th December 2009. A wide range of topics and speakers are planned for the conference on 18th December, and 11 UK families will be there to hear about MPS American-style as well as enjoying the parks. The lucky families whose names were drawn at the MPS Conference Gala Dinner are:

- Callum Pollock (MPS I) and family
- Oliver Robinson (MPS III) and family
- Lucinda Mickleburgh (Fabry) and family
- Jack Ferguson (MPS II) and family
- Ali Anwar Khan (MPS IVA) and family
- Sophie O'Connor (MPS I) and family
- Lewis Williams (ML III) and family
- Hannah Cooper (MPS I) and family
- Diane Hughes (Fabry), Laura Hughes (Fabry), Thomas Hughes (Fabry), Nicola Hughes (Fabry)
- Chloe McCauley (MPS I) and family
- Thomas Coney (MPS VI) and family

Paul is 30!



Paul was born on 9th July 1979 weighing in at 10lbs 6oz. (ouch!). It was obvious that he had some problems, loud breathing etc. and after lots of hospital visits and tests, Paul was diagnosed with Hurler's syndrome as it was called in those days in January 1980. An early diagnosis was achieved because of a very good and caring G.P.

By coincidence a friend had fostered a little girl who had Hurler's syndrome and was under Dr. Hugh-Jones and Professor Hobbs at the Westminster Children's Hospital. Paul was referred to them and underwent a Bone Marrow Transplant in June 1980. He was the first in the world to have a transplant for an "inborn error". The help and support that Ray and I received from family and friends was enormous.

Paul has had a few problems over the years but has overcome them with his usual cheerfulness and good humour. Over the last thirty years family life has been as normal as we can possibly make it. Paul has an older sister Emma and after he came out of hospital he had to fit in with all the things that we did before he came along. He was a cub and a scout and an altar boy at our church when he was younger. He was awarded the John Cornwell medal (a recipient of the Victoria Cross during the first world) by the scout movement, for overcoming his difficulties.

Paul lives at home with us but likes to be as independent as he can possibly be. (As long as dad ferries him back and forth but dad has retired now and so has nothing better to do). Paul uses a wheelchair (his wheels as he calls them) and gets about as much as possible, going to the cinema and snooker with his mates. Paul loves films and could go on Mastermind with his knowledge of who directed and who starred in most films of his era. He likes going out to eat and amongst his favourites are Chinese and steak and chips. He volunteers at an after school club one afternoon a week and works in the local McDonalds on Fridays. He has been there for about eight years and after his first job, clearing tables (in his chair) he was promoted to putting together the "happy meals" and children's party boxes.

Paul also loves to follow most sports, in particular our local team Crystal Palace. His chair is blue with red wheels, Crystal Palace colours. Like his films his knowledge of other teams, who the players are and who they have previously played for is extensive.

Emma, Paul's sister, is married now and has a little girl, Katie born in October 2007. Paul is thrilled to be an uncle and she loves her uncle Paul. Emma has been a wonderful sister to Paul and she and her husband James look after him for us when we need a break. **Anne and Ray Franklin**

Do you have a story to share?
Please email
newsletter@mpssociety.co.uk
or phone **0845 389 9901**

Helen's Big Day

Last Saturday was a big day for Helen Skidmore, of Monmouth, because it was the day she celebrated her thirtieth birthday.

What's so special about that, you are probably asking yourselves? Well, within months of her birth, Helen was found to be suffering from a rare genetic condition called Hurler Syndrome and it was likely she would not survive more than five years. The condition had only been discovered in the mid-sixties and any treatment was purely experimental and open to failure.

The story of how Helen survived the condition and of the refusal by her family to give up the fight on her behalf is a remarkable one.

Proud Mum, Pat said 'Helen is the oldest surviving Hurler's patient, the treatment was so experimental at the time that only two boys had been treated. Helen was the first girl.'

Pat and husband Michael, who are in business in the town, were delighted when Pat had a little girl, who they named Helen. They already had a son, Richard.

Within months the joy had turned to despair when doctors discovered she was suffering from Hurler's Syndrome, a genetic condition which would eventually leave Helen blind, deaf and possibly even suffering from brain damage.

Medical experts admitted her to Westminster Children's Hospital, where two boys had already been treated for the condition. It was agreed Helen would receive a bone marrow transplant, using her brother Richard who was five at the time, as the donor.

'He was not a good match, but he was at least a sibling' said Pat Skidmore. The operation failed and a decision had to be taken on the way forward. Finally it was agreed the second donor would be Michael.

Before the procedure and for many weeks after, Helen was contained in a sterile bubble, protecting her from infections in the outside world.

For weeks Pat kept a lonely vigil, watching and waiting for the signs that her beloved daughter was slowly recovering. Finally, at the end of October, months after Helen had first been admitted to hospital, Helen and Pat returned home 'after what seemed a lifetime of having our family separated,' said Pat.

Since then, the daily lives of the Skidmore family has been a series of milestones.

Helen started attending the local infant school, which she quickly adapted to and then adored. There was the proud and happy moment when Pat, Michael and Richard were able to see Helen perform as Mary in the Nativity play in the end of term concert with all the other normal children.

'The treatment for Hurler's and associated conditions has progressed to astonishing levels over the years, but I do not forget the heartache and sacrifice of so many parents who agreed to treatment for their precious son or daughter in the hope of removing the pain and suffering their child was destined to endure,' said Pat.

She said the last 30 years had been, at times, frightening, worrying and many tears had been shed. 'But then I have heard Helen giggle over something quite simple and I count my blessings and plod on through the quagmire called life.'

Do you have a story to share?
Please email
newsletter@mpssociety.co.uk
or phone **0845 389 9901**

Pat said, in making the arrangements for the birthday celebrations, she had come across numerous press cuttings and pictures which had reminded her of what Helen and the family had gone through all those years ago.

'I remembered the fact that Helen was the last person to use the sterile bubble,' said Pat. 'I found pictures of Helen horse-riding, of her on her roller skates, so she did all the normal things in her childhood.'

She added 'It has, in some respects, been thirty years of hell, but you look at what we have and you realise it has all been worthwhile.'

Pat said their son, Richard, who is now 33 and works for EMI, had always thought the world of his sister.

'It was Richard who gave Helen the bone marrow for the first transplant and he has been absolutely wonderful'.

Saturday's big day was attended by around one hundred people, many having been involved in various ways with Helen, her treatment and even helping to raise cash for the Helen Skidmore Appeal Fund, all those years ago.

The birthday party raised £635. As Helen had said she did not want presents, the cash goes to the MPS charity.

Article written by Robert Williams appears courtesy of the Monmouthshire Beacon.
www.monmouth-today.co.uk

Isaac's Scouting Award

Isaac, who has MPS I, Hurler Disease, has been a keen Cub for 5 years, and after spending 6 weeks in hospital last year with meningitis and numerous complications, getting back to Cubs was something he really wanted to do. At times he seemed too tired to go but he insisted.

In January this year rehearsals for the annual gang show began, and Isaac wanted to take part. For several consecutive Sundays Isaac was practicing scenes from Bugsy Malone and High School Musical, eventually getting costumes and make-up. In February the Gang Show started, 5 nights and 2 matinee performances. Throughout his time at Cubs, Isaac has been encouraged and supported by Kaa (Sarah Goodstadt), and it was her help that really made sure Isaac went to cubs and took part in all the different events.

At the Saturday afternoon matinee, the audience was packed with Isaac's family and friends. Bugsy Malone and High School Musical scenes came and went, with Isaac on stage performing. Just as everyone was getting ready to leave their seats for the interval and refreshments, the Chief Scout for the North-West took the stage and began to tell the audience about the bravery of John Cornwell, a 15 year old scout who was killed during the First World War, and stayed with his gun during the Battle of Jutland despite terrible injuries, and that since then the Scouting Association has created the Cornwell Award in recognition of Scouts who show considerable bravery in the face of great odds and challenges.

It suddenly dawned on us that Isaac was going to receive this rare award, so rare the Chief Scout told us that most scout leaders would never see one in a lifetime of scouting. Isaac was called to the front of the stage to shake hands and be photographed, hardly a dry eye in the house. On being presented with the certificate and badges Isaac responded, 'I've already got one'. 'Not like this one' he was told.

We are incredibly proud of Isaac's achievement, as was Isaac, and he and us got used to seeing him in the local papers over the next few weeks. Isaac's cub leader Sarah had made the application for the Cornwell Award following the terrible year he had and the dedication he's shown to being a scout despite the many challenges Isaac faces and lives with, and in many ways it's her award as well. **Adam Turner**

Sarah's Story

My name is Sarah McKnight and I was born with MPS I, Hurler Disease. When I was a year old I went to see Dr Ed Wraith and he got me a bone marrow transplant. The first one didn't work but I had a second one soon after and that was successful, so Dr Ed, there's hope for Newcastle United.

Mum and Dad have given me a pretty normal life and I have been in local schools since I was five years old. In September I go back into the sixth form of Dyffrn Taf where I have been for the last five years. I really enjoy school and will be taking the computing foundation course (Facebook and BEBO have made me a bit of an expert), beauty therepy and ASDAN which is designed to teach me how society works. I also go horse riding and swimming every Friday which is cool.

In the picture here, I am on my trike (I love that bike) and in the basket is my Dad's dog called Jude. She is really cute and one of my best friends.

I often ride down to the village which is called Pendine and onto the beach which is six miles long.

I've just been on holiday with my Dad and sister Rhoswen. We went to Rhodes which was good but I don't really like the very hot weather and only got my hands and feet tanned. Also I'm very picky about my food and lived on spag bol for a fortnight! I hope you enjoy my story. **Sarah McKnight**

Ian's Story

Hello everybody. As some of you already know me I will try not to bore you too much but for those who don't know me, here is my story.

I am a 43 year old male who has Fabry disease. My family was diagnosed with Fabry Disease 8 years ago, my uncle Robert was the first diagnosed and unfortunately he was extremely poorly when the diagnosis came through.
I was the next to be tested as my medical history had mirrored uncle Rob's, and you can probably guess, it was positive. So from then on the dam had broken and the flood waters were in rapid flow. My mum Kath was tested along with my sister Diane and you guessed it again, they were both positive.

Now this was fine as I had always had problems with my health, but nothing was ever conclusive and I was branded a hypochondriac despite many fevers, pains and many periods in hospital. So I was excited that at last I was somebody who had been told 'yes, we know what's wrong with you' so therefore all I wanted was my medication and life would get better.

I had recently had to change my career due to the fact that I was struggling with my work. I was a highly qualified driving instructor and at one point was the youngest instructor in Wales, but now I was working as a consultant with some big organizations and had a speciality in motorbikes and trucks. But I would get home from doing advanced training on the bike and could not get off it. To me this was normal for me to experience, however I was struggling more and I found it hard to put my bike on its stand. I had just trained a doctor and he noticed I was having problems and suggested I go to my GP. This I did and arthritis was the diagnosis, it was decided I would have a change in direction and went into sales, and ended up working for Vodafone as a retail manager. So, after 12 months of working for them, I get my diagnosis. I thought I WOULD GET MEDICATION and it would be gone and life would be great, or so I thought.

I was 35 when I was diagnosed with Fabry Disease and have thought many times that 'I wish I had found out earlier in life' but I have come to the conclusion that I was better off not knowing what was wrong. How can you say that, I hear you ask? Well, I knew nothing else, to me life WAS normal and if I had known sooner, could I honestly say I would have experienced the things I did, so apart from not having the condition I can say I would not have changed a thing.

It is only 8 years on that I can say that, I am who I am partly because of the sum of my experiences. So where was I now? And what happened between diagnosis and now? All I can say is that it has been the biggest roller coaster I have ever been on. Oh and I don't like roller coasters.

But it has not been all bad. Yes I have had lots of investigative work done, which was sometimes hard to go through and also there is the medication that is needed. However there have been many positive things, I have learned a lot about myself and how I cope with things. I will be honest in saying that I have not always dealt with things well and have suffered with depression and have needed time in hospital because of it and counselling. Although this has helped, I realised the only person who can change anything in my life is ME!

And this is what I have tried to do, one of the biggest things was to realise I have limits, (the tag from growing up of 'your lazy' has been hard to overcome) real limits although I guess like most people today finding them is hard. If I need to have a sleep in the afternoon then I will have one, bearing in mind that it can become a habit.

I have also got myself a little job on the permitted work scheme and this has been very good for me mentally and physically. I am now a productive member of the company I work in, they know fully about the Fabry disease even if they don't understand it. If I am not well though there is no problem and I just don't go to work. It is important for me to know that I do not need the job but it is something I want to do, and as a result of that, there is little pressure on me. That takes pressure off me to earn a living which psychologically is very important.

The other thing that is important is the fact that I will only do work that I can manage, i.e. I would not go and work on a building site but maybe working in the office on a building site is possible. And that is what I have done, although I find it difficult to work as an instructor again, I can work in the office and help with training matters from a different angle.

NEWS ON THE OLLIE G WISHES 2008

So finding something that is possible to do is a key to building coping strategies, for instance I have spent a lot of time learning to use a computer which again has helped keep my mind active. Also I have started to read and currently I am working through the books of Robert Ludlum. Our mind is a very strong force within us and it can work to help us or it can destroy us, so trying to learn to look at the positives is a definite challenge for me. Maybe I have not mastered it fully so shall we say this part of me is a work in progress.

So for the future I have made plans. I had not done this for a long time but feeling better has given me the will to look to the future. I am restoring my old BMW R80RT and of course, as many men will understand, you have to have the proper place to 'play' so I have started painting the inside of my garage and friends are helping me convert it into a work shop.

Also, I still plan to continue with my little job, just being around a normal working environment helps keep my feet on the ground. Also I see the problems other people have who don't have an underlying critical condition and, you know, some have it even harder than I do.

I would like to give you an update in the future, until then keep strong, keep positive and keep your mind busy.
Take care **Ian Hedgecock**

Editor's Note: We hope that Ian will continue to write a regular column for us about his personal experience of living with Fabry Disease.

News on the Ollie G Wishes 2008

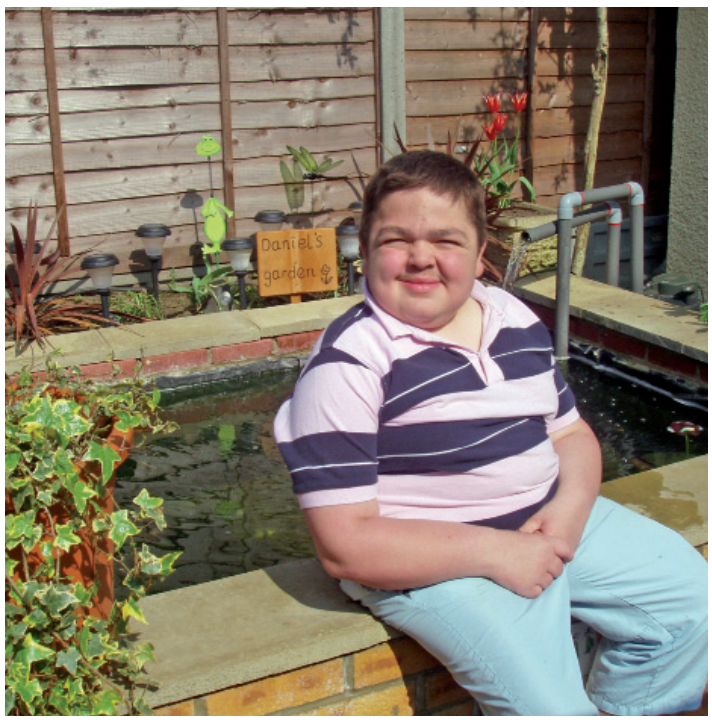
Daniel Cooke

It was a very sunny morning when Sarah and I arrived at the Koi Carpe Fisheries near Hitchin. We had previously spoken to Karl Cooke and been advised of the colours which Daniel likes and now armed with this information we wanted to ensure that we selected the correct fish. I never realised how difficult it was to catch a fish, I thought you simply placed the net over the one chosen and that was it! How wrong I was, the process took the assistant ages as the ones we had chosen just did not want to be caught! But the assistant persevered and 30 minutes later, united with the fish in colours which Daniel likes, placed inside a huge plastic bag, we were heading towards Daniel's house.

Well, he was so pleased with his gift, the photograph here best describes just how happy he was. Daniel loves fish and was really happy to show Sarah all the fish in his pond explaining much about them and how they ended up in his garden. It was a truly memorable morning for us and we would like to take this opportunity to add a big thank you to his parents for making us so welcome.

Linda Warner *l.warner@mpssociety.co.uk*
and **Sarah Irvine** *s.irvine@mpssociety.co.uk*

Daniel and his family 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!**



MPS Regional Specialist Clinics

Newcastle Clinic

5 May 2009

Due to bad weather the clinic usually planned for February was cancelled and a new date was finally agreed for 5th May 2009. All those who had an appointment for the February clinic were invited to attend the May clinic and everyone bar one person attended.

As always the clinic went well with only minor delays. Our thanks go to Dr Ed Wraith, Dr Rylance, Dr Leech and their team of nurses in outpatients for all their dedication, help and support

Sophie Thomas ***Senior Advocacy Officer***
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Northern Ireland Clinic

14 May 2009

The first of two Northern Ireland MPS clinics for this year took place on 14th May. Although it was raining the attendance was high and almost everybody managed to be there on time. We met with 7 children and 4 adults who are MPS members and their families and we welcomed a new Morquio member to the MPS Society. It was an opportunity for some of you to meet up as for some, it had been a long time since you last saw each other. Some gossip, jokes and contact details were exchanged. The clinic was very busy but it managed to finish on time, but unfortunately the tea supply finished before that. We are really sorry about that!

On the behalf of the Society, Sophie and I would like to thank Dr Fiona Stewart, her team and Dr Ed Wraith for another successful clinic.

Sophie Thomas and
Jolanta Turz ***Advocacy Support Officer***
j.turz@mpssociety.co.uk

Manchester BMT Clinics

1 and 22 May 2009

These clinics were the last two to take place in Royal Manchester Children's Hospital in Pendelbury. At the end of June the whole hospital is moving to the centre of Manchester. Although all families have many memories and are used to the Pendelbury I am sure children can't wait to explore new places to play in a new hospital.

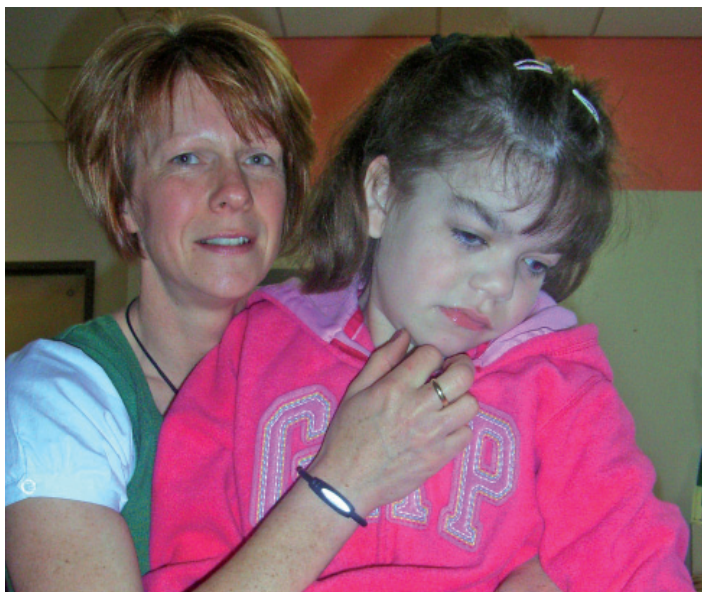
The clinic for under 6 years old on 1 May was busy but went really smoothly. It was lovely to meet all the children and their families who were all new to me but made me feel very welcome. The most popular colour among girls was pink and the most favourite thing to do at school for all children was… reading. To my surprise all the children said that they liked school but they made it obvious to everybody that they did not forget how to play.

The BMT Clinic for over 6 years old on 22 May was very exciting. There was a film crew and all the families turned up almost at the same time. It was a great opportunity to get together so all parents were chatting and all children were playing together making the room at the Willink very lively. The views on football teams and soon arriving summer holidays were exchanged. Although all the families were new to me I was made to feel very welcome.

I think we couldn't have dreamed of nicer clinics to say 'good bye' to the old hospital but I am already looking forward to seeing you again in a new place.

Linda Warner ***Roald Dahl Advocacy Officer for Progressive Neurological MPS Diseases***
l.warner@mpssociety.co.uk and
Jolanta Turz

Left: NI Clinic, Jade McAfee (MPS III). Right: Manchester BMT Clinic, Leighton Barker (MPS I BMT)



Great Ormond Street MPS III Clinic

21 May 2009

We had a very pleasant journey to GOSH, despite the rush hour traffic, and both Sarah and I thought with only five children today, it should be a fairly quiet and relaxed clinic. Well, the lesson we learnt was very clear, don't assume anything! Although only five families attended, what a lively clinic we had. Everyone was wide awake and there was heaps of energy in the waiting area.

Sarah and I were kept very busy not just making rockets from cardboard, drawing, colouring and reading stories but we also managed to rescue the receptionist's glasses several times and did our best to avoid a potential flood in the toilets, when the taps were turned on - fully!

The clinic offered a chance for families to chat, both parents and their children were kept very busy. Even Jamie managed to stop building his rocket long enough to read a story with Sonia, the Speech and Language Therapist at GOSH.

As the clinic came to an end and everyone had gone home there was little evidence left of just how busy the day had been. Although the receptionist is now talking about investing in some contact lenses!

As Sarah and I left for our journey home we reflected on what an enjoyable clinic it had been, albeit exhausting!

We would like to thank Dr Vellodi and his team for all their support and making us welcome.
Linda Warner and
Sarah Irvine ***Advocacy Support Officer***
s.irvine@mpssociety.co.uk

Bristol Clinic

26 May 2009

The clinic was a busy one with seven families attending from across the region.

As with all best laid plans the visits did not quite run to time due to unforeseen delays and I'd like to take this opportunity to thank everyone for their patience. From my point of view it gave me a chance to speak to the majority of the children, their siblings, mums, dads and carers and this was a great pleasure.

There were also two new families, one of which we visited on the ward as he was having his ERT.

Many thanks to Dr Jardine, Dr Wraith and Sally Melson 'Alice Rose' Metabolic Nurse Specialist for their help and expertise and to the staff at Bristol Children's Hospital for making me feel welcome.
Sarah Irvine

Cardiff Clinic

5 June 2009

Jolanta and I set off very early for our first visit to meet families at the Cardiff Clinic. Not only was the traffic and weather kind to us but we even found a parking space right outside the out-patients department.

The main waiting area was very busy, but with the help of Dr. Shortland and the hospital staff, we were able to meet with many of the families as they arrived. It was great to hear that Alicia was enjoying school and that Megan was learning to drive. It was lovely to meet Caroline who has just finished her degree and has promised an article on her work for a future magazine. Jolanta and I also spent a lovely time playing with Summer and Mabon while mum caught up with Dr. Shortland. Summer particularly liked the play house while Mabon enjoyed drawing with pencils and playing with the toy cars.

Many thanks to the families and staff at Cardiff hospital for making us feel welcome and the day so enjoyable.
Sarah Irvine and **Jolanta Turz**

Left: GOSH MPS III Clinic, Sophie Shields. Right: Top photo - Birmingham Clinic, Fahim Hussain (MPS III), botton photo - Bristol Clinic, from left Dr Germaine Pierre, Sally Melson ('Alice Rose' metabolic nurse specialist) and Dr Philip Jardine

Birmingham Clinic

12 June 2009

It was my first visit to Birmingham Children's Hospital and I was impressed by the warm atmosphere generated by the staff and families. Linda and I met Ansam and her mum, together with Fahim and his mum after their appointments with Dr. Hendriksz. It was also great to meet Natasha with her mum and dad and little sister to catch up on how things were going.

Linda and I went out to meet families in the waiting room, and while Linda chatted to Faye's mum and dad I sat with Pavan and his parents. Both families were looking forward to attending the conference.
Before the clinic ended Linda introduced me to Jasmine and Caitlin and their respective parents, and we were able to catch up with Louise and say thank you for all her help during the clinic. Once again I would like to say thank you to all the families and professionals for making it such an enjoyable experience.
Linda Warner and **Sarah Irvine**

Manchester BMT Clinics

17 and 24 July 2009

Jolanta and I attended both clinics and were looking forward to seeing the new Willink Clinic at St. Mary's Hospital. We were very impressed with the play area for the children and the range of toys available. At the under 6 clinic, while the weather outside was a little overcast, Shantelle, Luke and Ethan brought their energy and lovely smiles to brighten up the day. Shantelle looked great in her very stylish pink trainers and enjoyed playing with her sister and brothers while waiting for Dr. Wraith. Ethan and his brother had great fun racing each other round the waiting room pushing a buggy each, while Ethan took a shine to the activity walker.

At the over 6 clinic it was great to see Emma and Sarah and talk about their further education plans starting in September. Emma was looking forward to going on holiday later that evening, while Sarah told us about her adventures on her trike. Sarah's sister showed us a great photograph of Sarah on her trike and Sarah has written an article to go with it for this magazine.

We would like to extend our thanks to Jean, Ed and the team at Manchester for looking after us and the families and for making us feel so welcome.
Sarah Irvine and **Jolanta Turz**

Editor's Note:
Linda Warner is the *Roald Dahl Advocacy Officer for Progressive Neurological MPS Diseases*

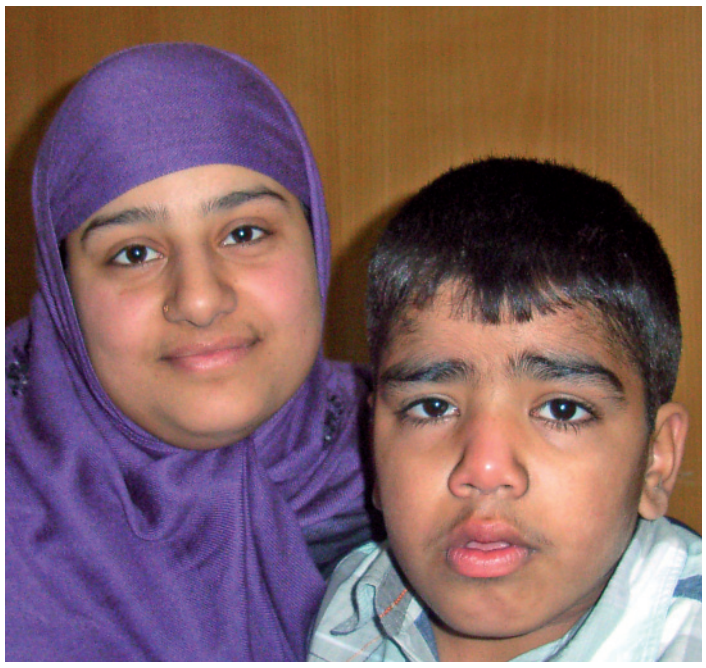
Providing practical support for children with brain, blood and literacy problems

Bristol Clinic

25 August 2009

It was a very busy clinic this month. It was great to see Archie and Faye again and to meet Andrew, Edward, Reece and Farhaan for the first time.

Jolanta and I, on behalf of The MPS Society, would like to welcome Dr Germaine Pierre as the new consultant at the Bristol Clinic. We wish her well in her new role and look forward to working with her and the rest of the team in the future.
Sarah Irvine



Sibling Weekend

'This year's Sibling Weekend was filled with loads of fun and continous laughs, from canoeing to archery and zip wire to giant swing (a swing that really was giant). The weekend was one certainly to remember. It was an action-packed experience, which also included a magnet search between floor boards. Thank you MPS and keep up the great work.' **Vishal**

The sibling weekend was held at Boreatton Park from 14th to 17th August. I travelled by train to the PGL camp along with the staff Sophie and Jolanta. Dale and Vish were the volunteers. I went with two other siblings Abraham and his sister Rachel. We got to the camp where we met our group leader Rachel and she took us to our cabins. At around 5 o'clock we went for dinner which was an Indian night. The evening activity was 50/50 where we were split into 2 teams and we had to do different challenges such as peel an orange with your mouth or create clothing from 3 bin bags. After that Rachael left us and the boys played football until it was time to get into our rooms.

The following day we were woken up and went to breakfast at half-past eight then we had our first activity which was a challenge course which included climbing a flat 3 metre wall, balancing along beams, climbing through a tyre tunnel and a slanted climbing wall. We then swapped with the other group and went on to the big swing in pairs which was great. We went back to the canteen to have our lunch. After lunch we went canoeing along a river with four in the boat. We played a game where you had to swap seats with someone else and if you were last you had to do a forfeit. We had dinner and then did our final activity of the day which was passport to the world where we had to find a flag and answer the questions to get points.

Our first activity on the Sunday was archery where we had to hit different parts of the board to get a particular item to eat with that lunch. This was one of my favourite activities along with fencing. The rest of the day we did abseiling where we had to walk down a wall, fencing and giant zipwire. After dinner we played a game called robot wars. We were put into teams and had to dress someone up like a robot then they were put in an arena and were blindfolded. The rest of the team had to direct them to pick up flour or water to throw at the opposition.

On the last day we had two activities which were quadbiking and leap of faith (trapeze). After we had finished our activities we went back to the white marquee to collect our packed lunches.

To conclude, it was a great weekend and I think everyone really enjoyed it as well.

Will Summerton

"When I first arrived at Boreatton Park I met Rachel who was going to be our team leader. Later on I met my new friend Georgia and I shared a room with her. We did lots of briliant activities that weekend and they included: big swing, challenge course, canoeing, fencing, zipwire, quad biking, absailing, trampeeze and archery. My favourite was the zipwire.

I really enjoyed the weekend as we had lots of fun. Thank you to all the MPS leaders, and me and my brother Mikkel are looking forward to going again next year." **Mia Fisher**

London Zoo Family Day

Meeting friends, old and new at London Zoo

On Saturday 1 July, we got up early and prepared for a trip into London for a day out at the London Zoo with the MPS Society.

We met the MPS team outside the gates at 10 am, quite a feat to be up, ready and in London by that time – especially on a Saturday. We all met up and had drinks in the zoo before splitting up and going off in various groups.

Meeting up at the beginning of the day was excellent as it gave us the opportunity to meet up with all the other families and get to know them. This was great as we were then able to stop and chat as we bumped into various families during the day. At this point it looked like the sun was going to shine for us.

We started with the Aquarium, as did most people, and spent quite some time viewing the fish and chatting with the other families. By the time we had completed this it was time to retire for coffee, we settled down outside the café with the Gremo family and rearranged the tables to accommodate ourselves. It was great to see another pair of families arrive and rearrange the remaining tables, the take over of the outdoor seating was complete! With a collection of buggies and wheelchairs, siblings and parents there was no room for anyone else. The staff were great and didn't mind at all, once they had got over the initial shock!

After a quick pit stop to change the children we moved on to the Carousel. The attendant was most helpful and his enthusiasm persuaded Bob and I to take Nathan on the ride (pictured below right). It was quite an experience for us but we all agreed that Nathan thoroughly enjoyed it. Getting him back down was quite a challenge.

We realised that we had spent far too long talking and we went off in search of the big cats. We detoured to view the butterfly tunnel, such amazing creatures, it kept both Sophie and Nathan interested.

We met two new families both with Sanfilippo children and spent some time chatting and comparing notes before moving on to finally see the tigers! Stopping en route for a very late lunch.

As time was running out we again bumped into the the families and spent a while discussing the joys and problems of living with Sanfilippo, as I have said before it is great to meet families at these gatherings and realise you are not alone and the problems you face are not only yours.

As we drove home we all commented how few animals we had seen and all decided that next time we would go back to see the animals! **Tim Summerton**, MPS Trustee and father of Sophie (MPS III) The Summertons are pictured below left.

MPS Conference 2009

The National MPS Conference was held at the Hilton Hotel, Northampton on Friday 26th – Sunday 28th June this year, and was a great success with nearly 400 people attending for all or part of the weekend. Seventy UK MPS Society families were represented, along with 6 Irish MPS Society families and 4 Spanish MPS Society adults. Also present were Fabry family representatives from Ireland, Australia and the USA, and two MPS family representatives from Saudi Arabia. Nearly a hundred UK professionals from the fields of Healthcare, Science and the Pharmaceutical industry attended, along with nine international professionals from Poland, France, the Netherlands, Ireland, and the USA.

Conference Programme

On the Friday evening the MPS families and the professional delegates arrived to register and enjoy a buffet dinner and a chance to catch up with old friends.

On Saturday three symposia ran simultaneously. The theme for Conference A was "Thinking Ahead – From Paediatrics to Independent Living". The speakers included healthcare and scientific professionals from Manchester, Birmingham, Belfast and London. It included presentations from MPS family members Samantha Brockie (MPS I) on "My Story of Spinal Intervention in MPS I"; Sandra Bates (mother of Jamie, MPS I) on "Bone Marrow Transplant from a Patient's Perspective"; Faye Longley (MPS IVA) on "Pathways to Living Independently with MPS IVA"; and Paul Moody (father of Oliver, MPS VI) on "Enzyme Replacement Therapy – the Highs and Lows".

The focus for Conference B was "MPS Neurological Diseases". The speakers from the fields of healthcare and science came from Bristol, London, Birmingham, Northampton, Belfast and Manchester. Presentations were also given by MPS family members Janet Gremo (mother of Nathan, MPS III), on "Managing Challenging Behaviour from a Parent's Perspective"; Fer Pidden (mother of Natalie, who had MPS III) on "Support for our daughter with MPS III in an NHS setting"; Nita Tailor (mother of Pavan, Multiple Sulphatase Deficiency) on "Palliative Care Management and Hospice Care from a Parent's Perspective"; and Gordon & Anne Hill, parents of Louise (who had MPS III) on "We led our Daughter's palliative care – Our Experiences".

Conference C was the Fabry programme and featured presentations from healthcare and scientific professionals from Cardiff, Manchester, Amsterdam, Cambridge, London and New York. It included presentations from Fabry family members Ian Hedgecock and Diane Hughes on "Three Generations of Fabry disease in our Family"; and a presentation given by Sarah Irvine (Advocacy Support Officer) on behalf of Fabry member Victoria Excell on "My experience of pregnancy whilst on ERT treatment for Fabry". The programme also featured Special Guest Family Speakers from the United States, Dan Kuusisto and his daughter Sandra Stenersen, who come from an extended family of Fabry sufferers in the USA. They spoke on "Families Living with Fabry Disease" and presented a video about Fabry disease in their family.

An exciting new addition to the programme was the introduction of a Great Debates in each symposium. In Conference A the motion was, "This house believes that the level of disability benefit should be reviewed after two years on Enzyme Replacement Therapy". In Conference B the motion was "This house believes that children with progressive MPS Neurological Diseases who are in the Palliative Care stage should have their care concentrated locally through the Palliative Care Team rather than at a National Specialist LSD Centre". In Conference C the motion was "This house believes that children who are asymptomatic for Fabry disease should receive Enzyme Replacement Therapy in order to avoid disease progression in their future life."

These topics led to lively arguments For and Against each motion, with well-argued cases being made throughout. The debates encouraged audience participation using the Audience Response Technology, which included a keypad for every audience member to give their responses. It was very interesting to see the percentage of the audience who agreed or disagreed with the motion at the beginning of each debate, and then see whether the percentages changed once the topic had been debated. See page 27 for results. We look forward to using the Audience Response Technology at future conferences to gain more audience feedback and involvement. Many audience members have said how much they enjoyed being able to give their opinions in this way!

On the Sunday morning the Society's AGM was held, and then there was a full Research programme featuring science and healthcare professionals from London, Paris, Manchester, Cambridge, Gdansk and Birmingham. The first part of the morning focused on "Research – Looking to the Future", while the second part focused on reports on research projects currently being funded by the MPS Society.

Meanwhile the Fabry members had a discussion group to share their thoughts on life with Fabry in the UK, with our international Fabry visitors giving their views on living with Fabry in their respective countries.

We would like to thank all our speakers for giving such excellent and informative presentations, and especially those speakers who took part in the Great Debates as it was such a new venture and proved to be a great success.

Bereaved Programme

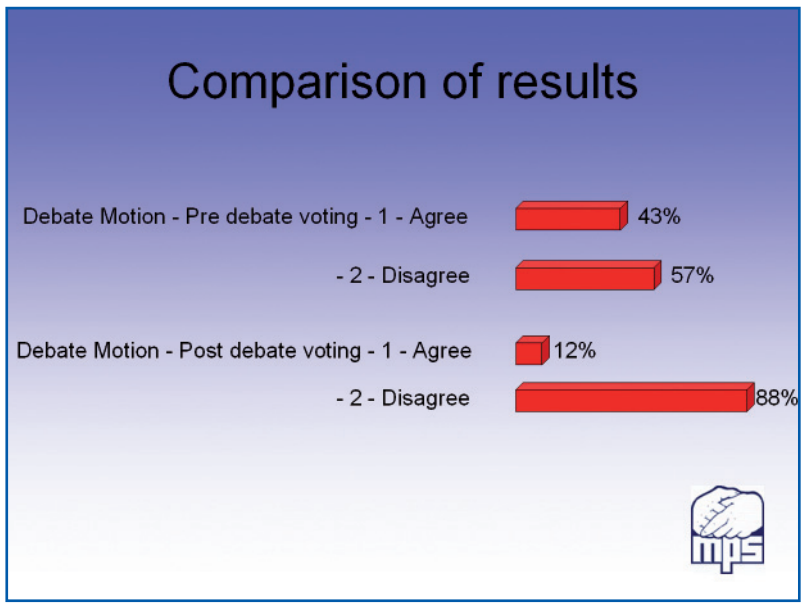
On the Saturday seven MPS family adults travelled from the hotel to Nottinghamshire, to meet with three other families for the Childhood Wood Remembrance Day. The day began in the beautiful surroundings of Rufford Country Park in Ollerton, with a chance to enjoy the countryside as well as to have a look at the stalls and activities which were part of a ceramics fair being held that weekend. The families then enjoyed lunch in the Saville Restaurant. Afterwards the families travelled to the Childhood Wood at Sherwood Pines for a time of remembrance and a balloon release to remember twenty-three children and adults who had lost their lives to MPS, Fabry or a related disease. We give our thanks to Sue and Dave Peach who ensured that the day ran smoothly and enabled the families who attended to have a time of peacefulness to remember the loved ones they had lost.

Gala Dinner

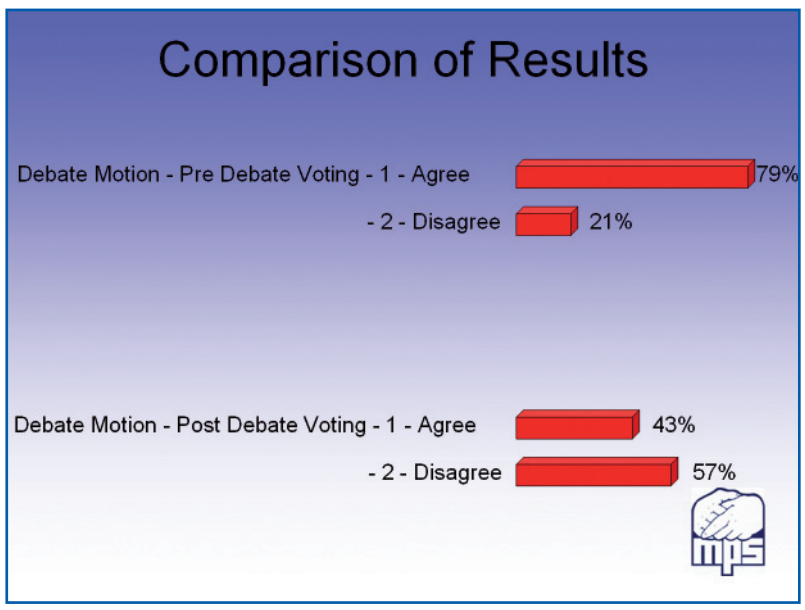
On Saturday evening, the adults were treated to an evening of fine dining and entertainment in the beautifully decorated Marquee. After a lovely meal, a raffle was held with many people winning great prizes, and raising over £700 for the work of the MPS Society in the process. The draw for the Disney Conference was undertaken by Barry Wilson, Chairman of Trustees, with 11 lucky families being drawn out of the hat from all the families who had entered. After this, the crowd was wonderfully entertained by 'The Dreamettes' and then a disco with all the hits to get everybody on their feet. The dance floor was full to bursting with people showing their best moves while the "stars" in the ceiling twinkled above. Outside, many people were delighted to take a ride or two (or more, in some cases) on the Simulator (just to check that the children would enjoy it the next day, of course).

Debate Results

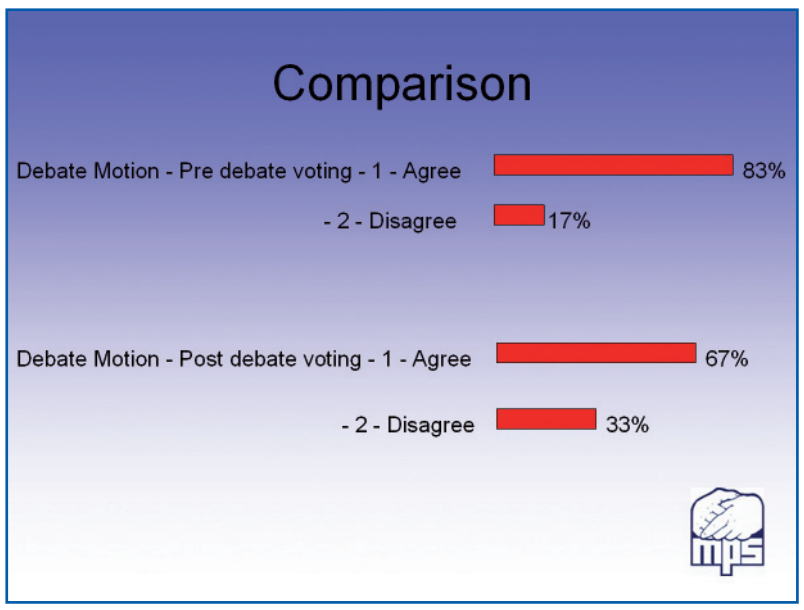
Debate A
“This house believes that the level of disability benefit should be reviewed after two years on Enzyme Replacement Therapy”



Debate B
“This house believes that children with progressive MPS Neurological Diseases who are in the Palliative Care stage should have their care concentrated locally through the Palliative Care Team rather than at a National Specialist LSD Centre”



Debate C
“This house believes that children who are asymptomatic for Fabry disease should receive Enzyme Replacement Therapy in order to avoid disease progression in their future life.”



MPS Conference 2009

Children and Young People's Programme

Over eighty children enjoyed the Children and Young People's Programme this year, supported by 43 volunteers.

On the Saturday the children headed off to Drayton Manor Park for a fun day out with their volunteers. A great time was had by all and luckily the weather was kind to us this year! On Saturday evening the children had childcare in their rooms watching a DVD of "Bolt" and eating all the bags of sweeties they had bought at the Tuck Shop. The teenagers enjoyed an evening out bowling.

On the Sunday morning a whole host of activities had been organised on-site. In the marquee the children could build a "Be My Bear", have their face painted, make dinosaurs or create some stylish jewellery. There was also balloon modelling and canvas painting and printing. Just outside the marquee there was a bouncy castle, a very exciting Laser Quest, and a 4-car Racing Challenge for the kids (and some of the volunteers!) to compete to be the fastest. There was also a Simulator which could take up to 17 people at a time on a variety of whirlwind rides such as rollercoasters, racing cars and a journey into outer space.

In the hotel some of the children had fun in the Soft Play Room, while next door the children could make friends with a small zoo of animals including a tortoise, snakes, rabbits and a skunk (who had been de-scented, much to everyone's relief!)

The children gathered in the marquee round an enormous cake to sing Happy Birthday to Daniel Muers who turned 13 on the Saturday, to Christian Huntley who had his 10th birthday on the Friday, and to Amy Donegani whose 20th birthday was also on the Friday. And to top everything off, there was a visit from two well-loved television characters, Peppa Pig and Fifi from the Flowertots, who went around the marquee to say hello and have a hug or a handshake with many of the children.

All the children and young people had a lovely time, and we would like to thank all our volunteers who do such a wonderful job at every MPS event, and without whom the childcare programme would not be possible. We know how much the parents appreciate the hard work and enthusiasm of the volunteers, how much the volunteers enjoy spending time with the children, and of course how much the children love the activities and the support of their volunteers at each event.

Close

By Sunday afternoon it was time for everyone to depart, feeling perhaps a little tired but hopefully having learnt more and having had an enjoyable time in each other's company once again. We look forward to the next National MPS Conference in two years' time, Friday 24th – Sunday 26th June 2011. We hope to see you there!
Sue Cotterell ***PA to CEO*** *sue.cotterell@mpssociety.co.uk*

"Our son had an excellent volunteer who listened to us and responded well to his needs."

" Our volunteer was superb. She took on all the MPS issues without any problem. She was quick to learn and gave me a high level of confidence."

"A very conscientious and helpful young man. My son enjoyed himself thoroughly. Many thanks."

"The carers were fantastic and our children really enjoyed being with them."

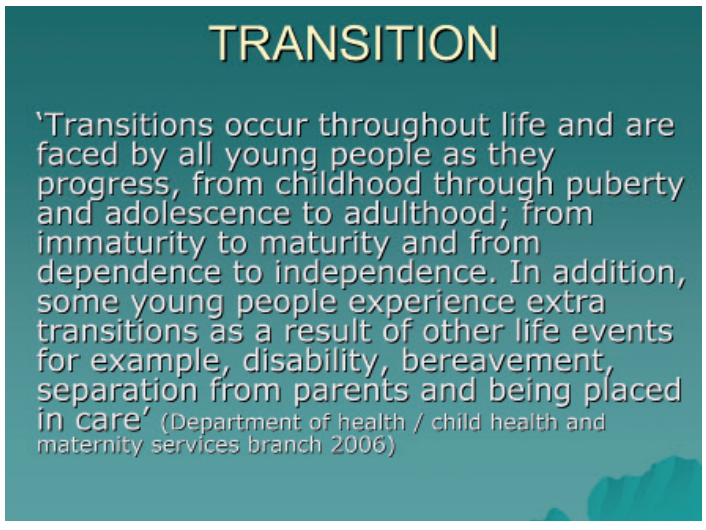
"Lovely rapport with the children - very caring. We felt very relaxed leaving the children with her."

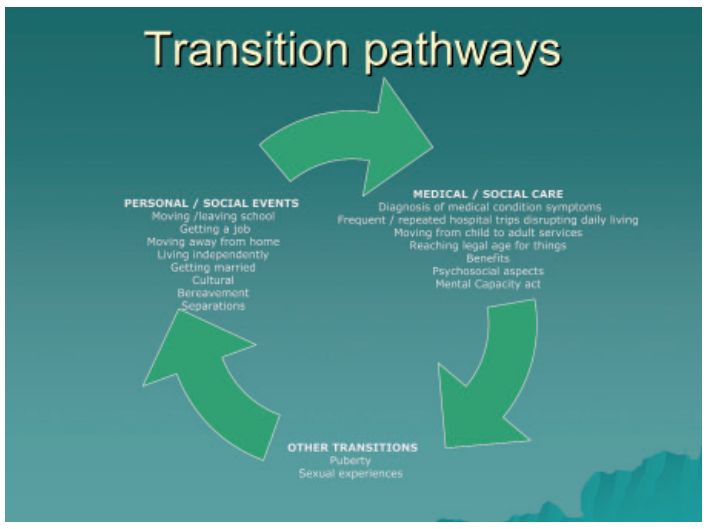
Conference Presentation

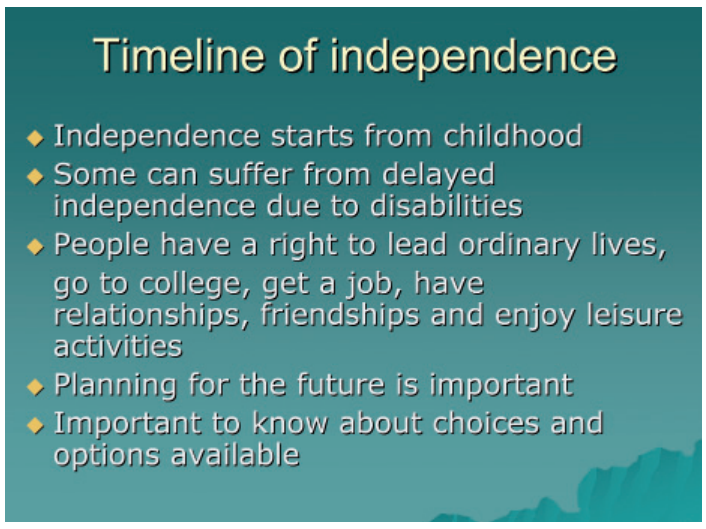
Over the next few editions of the MPS Magazine, we shall be including a selection of presentations given at the MPS Conference 2009.

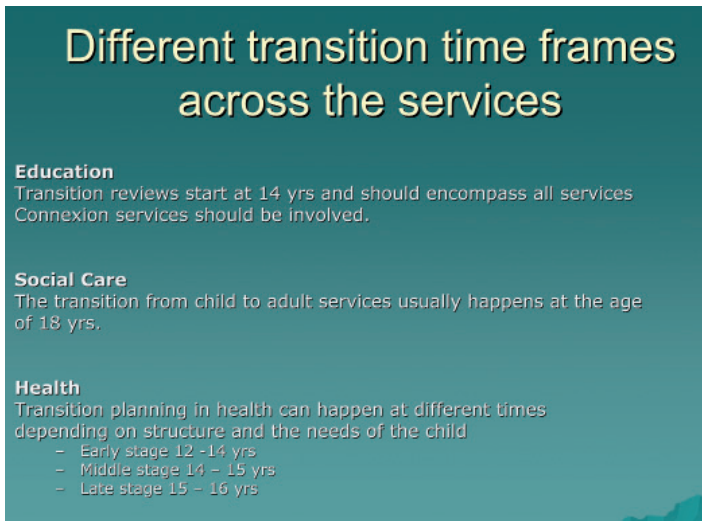
Independent Living: Avoiding the Pitfalls

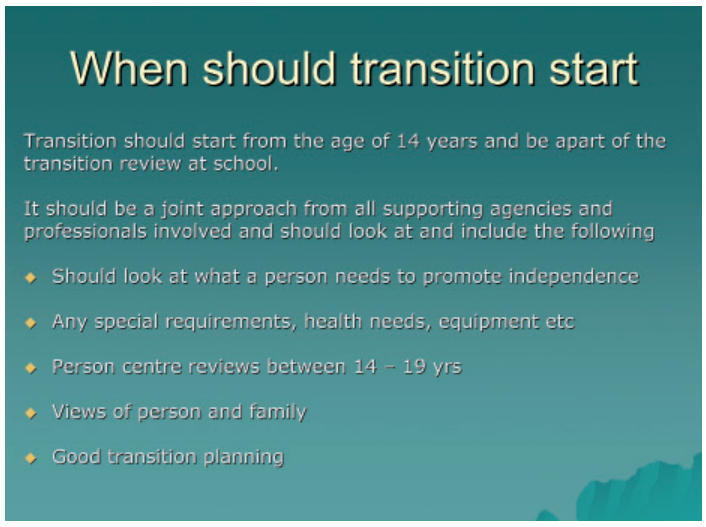
Sophie Thomas, Senior Advocacy Officer, MPS Society











Different assessments & support options

- Person centred planning
- Trans-active communication
- Section 140 assessment
- Health checks and Health action plans
- Pathway plans
- Social care assessments
- Community care assessments
- Common Assessment framework
- Carers assessments

Family participation

- Family members have a key role in supporting children through transition and independence.
- They often act as key workers and advocates for their children
- They very often put their children's needs ahead of their own
- Their views and needs should be taken into account.

Benefits

At 16 yrs, benefits may change and can move from parent to individual control. This is optional and for those who have capacity they may decide to take responsibility for these themselves. This affects things such as Disability Living Allowance.

Other benefits such as education grants, incapacity, income support and housing benefit can be applied for where applicable.

If social care is needed, direct payments and individualised budgets should be considered as well as applying to the independent living fund.

Independent living fund gives funding to enable a person to live in the community. It is used to purchase services from agencies or can be used to employ your own personal assistant.

Housing

- Right to have an informed choice of where and with whom you live - Council / Housing association housing, supported living, shared housing, home ownership schemes.
- Need to look at both long and short term needs
- Service providers need to look creatively at what is needed. I.e. new home, Shared or supported accommodation, adaptations
- Services and packages of care need to be coordinated and tailored to ensure that a persons needs are met.
- DFG for adaptations

Transport

- Vouchers for driving lessons
- Motorbility cars
- Disability Living Allowance
- Bus passes
- Train vouchers
- Vouchers of tokens to use taxi's or transport systems such as dial a ride which is a bus service which will transport people to various destinations whether it is access a day service or doing your weekly shop.
- Help with hospital travel costs



Summary

- Provision is very different between child and adult services
- It is Important to allow children young adults to experience different options
- Do start early and find out when things change in your local area
- Important to have a network of supporting agencies
- Be prepared for different services or reduction in support when move from child to adult services
- Take one step at a time
- Know that you can change your mind about things at any time.
- Always ask for records and copies of things
- Appoint an advocate if needed
- Parents need to allow their children to make decisions and choices but be supportive. A parents view is important but it is more important to put the young person first

New Clinical Study Group for Inherited Metabolic Disorders

Dr Chris Hendriksz, Consultant in Clinical Inherited Metabolic Disorders at Birmingham Children's Hospital reports on the new Clinical Study Group (CSG) for Inherited Metabolic Disorders (IMD) recently set up in collaboration with the UK LSD Patient Collaboration Group.

To improve research for patient benefit, the UK has been divided into Clinical Research networks and the individual networks have been assigned specific remits.

Due to the diversity and the fact that many rare disorders do not fit into one of the allocated groups, it was felt that patients from these groups might lose out.

A specific group was dedicated to deal with developing research around the use of medicines in children and was called the Medicines for Children's Research Network (MCRN). As this was the only group specifically looking at the needs of children, it was felt appropriate to approach them about inclusion into their remit. They were approached, but as adults with rare metabolic disorders only form a very small group, their inclusion was also requested.

This was agreed on the grounds that a CSG, or clinical specialist group, could be formed if adequate funding could be found to support it. The UK LSD Patient Collaboration Group kindly agreed to fund this group in principle for the next two years and will have a permanent member in the CSG. The Gaucher's Assocation is part of this consortium and it is hoped that this will lead to new opportunities for research involving patients affected by all rare inherited metabolic disorders.

A new development giving even more opportunities for research was the formation of a second group that will be developing studies but not dealing with medicine-only studies. This area would look at other research studies like pharmacovigilance, patient registries, developing new biomarkers or other basic science projects that will ultimately lead to patient benefit.

This may open the door to some funding streams, but more importantly, if studies are adopted into the network it will qualify for research support costs, meaning support from the research network partners like pharmacology, methodology and other specialist groups. It will also create training opportunities and members will have access to training programs such as communication in research, GCP training and other research related modules.

What's next?
The post of Chairman for this new CSG for Metabolic Storage Disorders was advertised nationally and I was delighted to be recruited to the post. The next step is to recruit additional members to the group who will be elected and appointed to form the clinical studies group for the next three years. Once the members have been appointed they will meet to develop a research strategy and start to prioritise projects. Only studies that have been open to national competition can be adopted but is unlikely to be a hurdle, as the different units tend to work closely together and especially as far as Lysosomal Storage Disorders are concerned, national collaboration has been achieved via the National Commissioning Group system.

You can find more information on the MCRN and the functioning of the clinical study groups at www.medicinesforchildren.org.uk. They are also slowly starting to build an information database for parents and patients so please feel free to comment or contribute to any of their calls for help if you are able to.

Only if we work together as patients, families, patient support groups and all healthcare professionals will we be able to decrease the disease burden for rare disorders.

Article taken from Gauchers NEWS
www.gaucher.org.uk

Introducing the Adult IMD Service at Salford Royal Foundation Trust

Hello, my name is Carly Bleakley and I am a Nurse Practitioner at Salford Royal Foundation Trust in the Department of Adults with Inherited Metabolic Disorders. My main role is the link nurse/disease specific nurse for patients with MPS diseases. We see lots of other Lysosomal Storage Disorders here including Pompe disease and Fabry disease to name a few.

We have 39 patients here with MPS diseases, we have 18 patients with MPS I, 13 of which are on treatment, we have 4 patients with MPS II, all are on treatment, 5 patients with MPS III, 6 patients with MPS IV and 6 patients with MPS VI, 4 of which have treatment.

I have been with the service for the last four years and my role in being the disease specific nurse for patients with MPS diseases began 18 months ago. Since then I have been familiarising myself with diseases and gradually introducing myself to the patients. It has been a big change for patients as they have moved here from the children's hospital and we have all tried to make it as smooth a transition as possible. I hope to be able to attend the children's hospital when patients are ready to transfer to introduce myself and so that they have a contact with someone before they attend here.

My main role is to see patients with MPS disease when they attend here for clinic appointments and to have telephone contact with them should they have any questions or queries.

All of our patients are on home treatment with the exception of two who have there ERT at their local hospitals. Although they don't all attend here for their ERT they are seen here every 6/12 months and have assessments every year.

If there are any parents with children/ adolescents who are getting ready to transfer from Royal Manchester Children's Hospital with MPS who wish to contact me, please see my details below.

Carly Bleakley
Nurse Practitioner
Adults with Inherited Metabolic Disorders
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Clinical Trials Update

MPS I

New study of intrathecal enzyme replacement therapy for cognitive decline in MPS I patients
The Los Angeles Biomedical Research Institute at Harbor-UCLA Medical Center in Torrance, California, in collaboration with the University of Minnesota, are starting a 24-month study to examine whether intrathecal enzyme replacement therapy with Aldurazyme will help to stabilise or reverse memory loss and cognitive ability in individuals with MPS I.

MPS I Intrathecal ERT for Spinal Cord Compression One-Year Extension Study Approved at the University of California and Los Angeles (UCLA) involving 10 patients
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 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 α-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.

The Primary Outcomes are: Safety of intrathecal enzyme replacement treatment by blood and spinal fluid tests each month; improvement in neurologic signs related to spinal cord compression, by neurologic examination and Japanese Orthopaedic Association 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.

The Secondary Outcomes are: 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.

MPS I Intrathecal ERT for Children Being Considered for Transplantation
The University of Minnesota recently has obtained U.S. 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 replacement 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 date of the transplant.

MPS II

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

MPS III

Shire Pharmaceuticals Group, as part of its research to evaluate new approaches to the problem of treatment of the central nervous system, is hoping to move its MPS IIIA 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 its research initiatives to include MPS IIIA. The Shire website is www.shire.com

MPS IV

The Enzyme Replacement Therapy phase I/II study is designed as an open-label, within-patient dose escalation trial in 18 patients followed by a treatment continuation phase. All patients have been enrolled in this study. During the dose escalation phase of the study, subjects will receive weekly intravenous infusions of BMN-110 in three consecutive 12-week dosing intervals. The objectives of the phase I/II study is to evaluate safety, pharmacokinetics, pharmacodynamics and to identify the optimal dose of GALNS for future studies.

BioMarin is also conducting a Morquio Clinical Assessment Program or MorCAP that involves about 15 centres in many countries and will evaluate the disease situation for patients globally. Finally they expect to have a phase III double-blind placebo controlled study that is looking to include 50 –100 patients from many centres. Additional information can be found at www.morquioBMRN.com

Article taken from the USA MPS Society's magazine.

Hunter Syndrome Intrathecal Enzyme Replacement Clinical Trial

Researchers at the University of North Carolina at Chapel Hill are conducting a clinical trial to learn if direct administration of recombinant enzyme into the fluid around the brain and the spinal cord is safe and a possible treatment for children with Hunter syndrome with developmental delays. The principal investigator for the clinical trial 'A phase I/II safety and ascending dose ranging study of idursulfase administration via an intrathecal drug delivery device in paediatric patients with Hunter syndrome who demonstrate evidence of central nervous system involvement and who are receiving treatment with Elaprase' is Joseph Muenzer, MD, PhD and the clinical trial is sponsored by Shire Human Genetics Therapies, Inc (Cambridge, MA).

Hunter syndrome is a genetic disorder in which the enzyme, iduronate-2-sulfatse is missing or not working properly. Enzyme Replacement Therapy using intravenous (IV) Elaprase (recombinant human iduronate-2-sulfatase) has been available for the treatment of patients with Hunter Syndrome since 2006. IV Elaprase administration is not expected to prevent or reverse developmental delay in patients with Hunter syndrome, because Elaprase has been shown to cross the blood-brain barrier and enter the brain or spinal cord after an IV infusion.

Currently, there is no approved therapy for treating the brain and spinal cord in patients with the severe form of Hunter syndrome. The goal of this study is to give a new preparation of iduronate-2-sulfatase (idursulfase-IT) directly into the fluid surrounding the brain and spinal cord (intrathecal administration). The new form of iduronate-2-sulfatase has not been used before in patients with Hunter syndrome and is considered investigational. It has not been approved by the FDA or any other regulatory agency.

This Phase I/II clinical trial is planning to enrol 16 patients with Hunter syndrome between the ages of 3 to 8 years with evidence of early neurocognitive decline using an open-label, three-dose trial design. This clinical trial will initially have both a treatment group (12 study patients) and a control group (4 study patients) with the control group eligible to receive intrathecal enzyme after a six month observational period. The monthly intrathecal administration of idursulfase-IT will be given using a Port-A-Cath ® II Low Profile ™ intrathecal implantable access system manufactured by Smiths Medical MC, Inc (St Paul, MN) that requires surgical implantation.

To be eligible for the investigational intrathecal enzyme replacement clinical trial, study patients need to have some developmental delay, but cannot be severely impaired, have received and tolerated a minimum of 6 months of weekly intravenous Elaprase and have adequate hearing (with or without hearing aids) to complete developmental assessments. Patients with Hunter syndrome are not eligible if they have a shunt for the treatment of hydrocephalus, have had a cord blood or bone marrow transplant or have other medical conditions that may place the individual at increased risk during the investigational clinical trial.

Patient Enrolment Complete for Phase I/II Clinical trial for GALNS for MPS IVA, Morquio Disease at three UK Specialist Centres

Biomarin announced on 13 July 2009 that patient enrolment has been completed for the PhaseI/II clinical trial for BMN-110 or N-acetylgalactosamine 6-sulfatase (GALNS), intended for the treatment of MPS IVA.

"The efficient enrolment of twenty patients is a critical milestone for the MPS IVA program and demonstrates both our commitment to this program and the support and enthusiasm of the MPS IVA patient community. Data generated from this study will be valuable in demonstrating safety and could be instrumental in designing a successful Phase III trial. Assessments from the Phase I/II study such as plasma and urine keratan sulfate levels, pulmonary function and walk tests will be helpful in determining optimal Phase III end points," said Henry Fuchs, M.D., Chief Medical Officer of BioMarin. "We appreciate the collaboration of the Morquio community in this important effort, and we hope to develop this new treatment as expeditiously as possible."

Christian Hendriksz, MBChB, MSc, FRCPCH, MRCP, Consultant in Metabolic Disorders, Birmingham Children's Hospital, UK, added, "Morquio is a serious and debilitating disease in which accumulation of keratan sulfate results in impaired breathing and walking, recurrent infections, impaired bone and joint function, dysmorphology and an overall impaired quality of life. The rapid accrual of patients for the Phase I/II study speaks to the magnitude of the unmet need for this disease."

The Phase I/II study is designed as an open-label, within-patient dose escalation trial in 20 patients followed by a treatment continuation period. During the dose escalation phase of the study, subjects will receive weekly intravenous infusions of BMN-110 in three consecutive 12-week dosing intervals. The objectives of the Phase I/II study will be to evaluate safety, pharmacokinetics, pharmaco dynamics and to identify the optimal dose of GALNS for future studies. BioMarin plans to provide an extension study in which all patients in the Phase I/II study will be eligible to participate.

MORCAP

Clincal trial for the treatment of Morquio Disease, MPS IVA

To the excitement of all the Morquio community, the clinical trial for the treatment of MPS IVA started in UK at the beginning of this year.

Birmingham Children's Hospital (BCH) was the first from three designated centres to carry out testing for the first phase of the study called MorCap or MOR-001. It was shortly followed by Great Ormond Street Children's Hospital (GOSH) and the Manchester Willink Unit. In total, thirty nine Morquio patients took part in MOR-001; nineteen at BCH, nine at GOSH and eleven at the Willink. After many tests and the hard work of all the medical staff engaged in the study, twenty committed patients were appointed to participate in the second phase of the clinical trial MOR-002, to receive enzyme infusions weekly for 36 weeks.

The first child to be infused, not only on this trial but first in the world, was Sultan Ali, pictured here with the ERT infusion team at Birmingham Children's Hospital. The infusion took place on 21st April in Birmingham. Olivia Vickery, pictured below with infusion nurses from GOSH, was the first to be infused in the London centre and it was carried out on her tenth birthday. The birthday cake proved to be a good distraction from needles! The last to follow was the Willink, not stopped by moving to the new hospital in Manchester's city centre, the first patient to receive enzyme infusion there was Katie Milby.

The youngest child on the trial is five years old and the oldest is eleven. We wish the best of luck to all the brave children on the trial and their families.

Results from both studies, MOR-001 and MOR-002, will help in designing potential enzyme treatment for Morquio disease.

Jolanta Turz ***Advocacy Officer***
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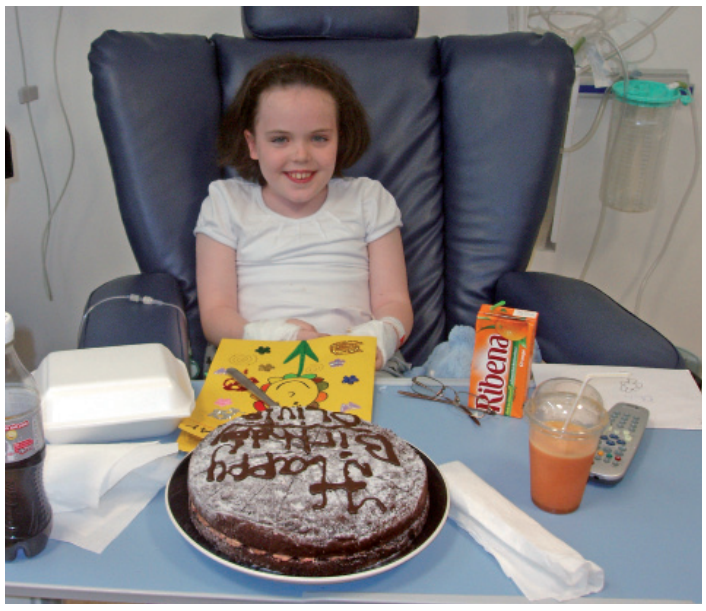


Another first for Birmingham Children's Hospital

A patient at Birmingham Children's Hospital is the first in the world to receive an innovative new enzyme treatment to help with their rare lysosomal storage disorder, Morquio A Syndrome (mucopolysaccharidosis IVA). This new drug candidate, developed by BioMarin Pharmaceutical of Novato, California, is the first and only treatment of its kind.

Dr Chris Hendriksz, a Consultant in Clinical Inherited Metabolic Disorders, commented:
"This is a very exciting announcement for our patients who have been waiting so long for a potential therapy. Hope of a potential therapy is now within reach for these patients, and Birmingham Children's Hospital is very honoured to be part of this exciting development."

Sultan Ali aged 9 from Moseley, was the first child in the world to receive the new intravenous infusion enzyme treatment on 21st April. For the next 72 weeks he will visit the hospital on a weekly basis to receive this treatment. It is hoped that this treatment will significantly improve his quality of life and prolong his lifespan. Sultan is joined by other children with the same disorders over the next few months that will be included in the same research study at Birmingham.

"Sultan called his new treatment his Spider juice as he is hoping for special powers like Spiderman" He is also looking forward to having more friends join him for the study that will initially last just over 18 months although treatment will be for life if it is successful.

This syndrome is very rare but its symptoms and long term effects can include: poor growth, abnormally formed bones and joints, heart valve problems as well as problems with breathing. Many patients become wheelchair dependent in their second decade of life and undergo numerous surgeries to alleviate life-threatening conditions caused by the underlying enzyme deficiency.

The Lysosomal Storage Disorders centre at Birmingham Children's Hospital supports one of the largest populations in the world affected by this rare disorder and over time has developed expertise in many fields caring for these patients. It is this expertise that attracted the biopharmaceutical company to the United Kingdom and specifically the local team to lead on this exciting project. The study will be expanded to Manchester and London over the next few months.

The new build Wellcome Trust Clinical Research Facility will be hosting the patients and the Medicines for Children Research Network will support this important project. To ensure that patients and their families have all the support they need the MPS Society is acting as advocates for this study.

Amicus Therapeutics

Amicus Fabry program begins Phase 3 following positive Phase 2 results shared in Spring 2009

In June, Amicus announced the beginning of its Phase 3 study of AT1001 (migalastat hydrochloride), the company's investigational drug for the treatment of Fabry disease. The company and the U.S. Food and Drug Administration (FDA) agreed on several important aspects of the trial, including the primary endpoint of changes in the amount of kidney interstitial capillary GL-3, the substrate that accumulates in the cells of Fabry patients. The FDA also agreed that Amicus is eligible to seek accelerated approval for AT1001. The accelerated approval regulations apply to products intended to treat a serious disease where there is an unmet medical need (for example, no approved therapy). A company can receive initial marketing approval for a qualifying product on the basis of a surrogate endpoint, and provide clinical outcome data at a later date.

"The start of our Phase 3 trial with AT1001 is a major milestone for Amicus and highlights our transition into a late-stage development company," said John Crowley, president and CEO of Amicus. "We are very pleased with the outcome of our interactions with FDA around the design of this pivotal study and are confident we have set the stage for a successful Phase 3 study. We continue to believe that AT1001 may be an important treatment option for patients who suffer with Fabry disease and a significant step forward for them and their families."

The Phase 3 study will evaluate the efficacy and safety of AT1001 in adult men and women with Fabry disease. This study will enroll individuals whose α-galactosidase A (GLA) mutation is responsive to AT1001 and who have never received enzyme replacement therapy (ERT) or who have not received ERT for 6 months or longer. The study objectives are to compare the effects of AT1001 versus placebo on kidney GL-3, kidney function, and urine GL-3.

Approximately 60 participants will be enrolled in this global study. Dosing is expected to begin in fall 2009.

Raphael Schiffmann, MD, Director of the Institute of Metabolic Disease, Baylor Health Care System Foundation in Dallas, commented, "Having been involved in Fabry research for more than 15 years and considering the Phase 2 data with migalastat, I am pleased that the Phase 3 trial is starting and look forward to continued involvement with this novel approach for the treatment of Fabry disease."

For more information on the Fabry Phase 3 clinical trial, please visit http://clinicaltrials.gov/ct2/show/NCT00925301

To read about the Fabry Phase 2 clinical studies and the Phase 2 extension study of AT1001, please visit http://tiny.cc/lktpi or patientadvocacy@amicustherapeutics.com

NEWS FROM THE PHARMA INDUSTRY

PRESS RELEASE: 25 June 2009

Supply shortages of Cerezyme and Fabrazyme - priority access for patients most in need of treatment recommended.

The European Medicines Agency's (EMEA) Committee for Medicinal Products for Human Use (CHMP) has recommended that patients who are in greatest need of treatment are given priority access to Cerezyme (imiglucerase) and Fabrazyme (agalsidase beta) during the expected supply shortage of these two medicines over the next few months.

Cerezyme and Fabrazyme are both used to treat rare, inherited enzyme-deficiency disorders. Cerezyme is used in patients with Gaucher disease, a disease in which patients do not have enough of an enzyme called alglucerase. Fabrazyme is used in patients with Fabry disease, a disease in which patients do not have enough of an enzyme called alpha-galactosidase A.

The supply shortage is caused by the shutting down of Genzyme's production site in Allston Landing, in the United States of America, where both medicines are produced. The company found a viral contamination (calicivirus of the type Vesivirus 2117) and has shut down the production facility for a sanitization of the bioreactors. The virus is not known to cause disease in humans, but it may affect the quantity, but not the quality, of the enzymes produced in the bioreactors. An in-depth investigation of the cause of the contamination is ongoing.

While the facility is shut down, no new stocks of Cerezyme and Fabrazyme can be produced. All batches manufactured prior to the detection of the contamination were tested and the Agency confirmed that they are suitable for release. To ensure that existing stocks last as long as possible until new batches can be produced, the European Medicines Agency has agreed to some temporary changes to the way these medicines are prescribed proposed by the company. These changes should be implemented immediately.

For Cerezyme, priority is given to infants, children and adolescents, and adults with active disease progression. These patients can continue to receive Cerezyme at the standard dosage schedule of one infusion every two weeks. However, adult patients without clinical evidence of active disease progression should receive Cerezyme at a reduced dose (half a dose once every two weeks) or at a reduced infusion frequency (once a month at their current dose).

For Fabrazyme, priority is given to children and adolescents, and adult male patients, who should continue to receive Fabrazyme as one infusion every two weeks. However, adult female patients, in whom the disease is less severe, may receive Fabrazyme at a reduced dose.

These are temporary recommendations and do not change the currently approved Product Information for either Cerezyme or Fabrazyme. It is expected that these changes will need to continue until the end of the year.

The Council Recommendation on Rare Diseases was adopted on June 9, 2009: Renewed commitment for national plans in all Member States by the end of 2013

Citizens of the European Union have the privilege to be defined not only by their geographical location or by their history, but also as people with civil rights enabling them to make collective choices. The European Parliament is elected by all European citizens, however less than 50% of them are actually voting - absenteeism is the first political party in Europe today. In the meantime, over 70% of new national laws and policies are the national transposition of laws and policies voted at the EU level. This European voting process involves ministers of all Member States and members of the European Parliament from all over the European Union. European citizens are not sufficiently aware of this process. The complexity but also the beauty of Europe today lies in this sophisticated articulation between the EU and national levels.

The field of rare diseases is an area of unique European added value for Community action. As such, it is a perfect example of how citizens from all over Europe have expressed their needs as patients and have called for action in order to shape a better environment for people living with rare diseases in Europe.

The momentum for an EU policy framework in the field of rare diseases is ripe. With the Commission Communication on Rare Diseases, adopted in November 2008, and its proposed Council Recommendation, adopted last month, we are where we wanted to be in order to take our cause to the next step and in particular toward the implementation of national strategies and plans in all EU Member States by 2013. For this next phase, we will rely on our committed staff and volunteers to act from three platforms: the EURORDIS Council of National Alliances; the EUROPLAN project - involving 24 Members States and EURORDIS; and the future EU Committee of Experts on Rare Diseases.

During the past four years, EURORDIS has relentlessly fuelled the political momentum, voiced the patient needs and expectations and contributed with concrete proposals. Our success is largely due to the high mobilisation of the rare disease community. It is also the result of the high quality dialogue and partnering between all stakeholders which enabled us to draw up common proposals. We now need to spread this momentum and key success factors in order to turn this political vision into concrete positive changes for people living with rare diseases in each Member State and beyond.

Yann Le Cam, Chief Executive Officer
Article courtesy of Eurordis (Newsletter July 2009)

New Study in Women with the Fabry Disease demonstrates significant benefits of treatment with REPLAGAL ® (agalsidase alfa) 0.2mg/kg on kidney function, heart function and burden of disease

Results from the largest and longest examination of enzyme replacement therapy (ERT) in female Fabry patients. Published in Genetics in Medicine.

The results from an observational study published in the June 2009 print issue of Genetics in Medicine demonstrate that enzyme replacement therapy (ERT) with REPLAGAL 0.2mg/kg is effective in treating some of the signs and symptoms of Fabry disease in women. Clinical benefits were measured, including stabilisation of kidney function and improvement in patients with stage 2 CKD, improvement in cardiac structure in some groups of patients, and reduction in pain and burden of disease. Authors concluded that this observational study, which included 36 women and last for 4 years, indicates that women with signs and symptoms of Fabry disease should be considered for ERT with agalsidase alfa, noting that the patient population studied may not be representative of the general Fabry disease female population.

'Our research has generated meaningful, measurable data which shows that long-term treatment with agalsidase alfa stabilises kidney function and improves heart function. This may result in important benefits for Fabry women, such as the reduced risk of heart failure,' said Professor Christoph Kampmann, MD, co-author of the study, Centre of Lysosomal Storage Diseases, Children's Hospital of the Universitatsmedizin Johannes Gutenberg University, Mainz, Germany.

Because women are commonly described as 'carriers' of Fabry disease, the true burden of their disease is often under estimated, which leads to delayed diagnosis and initiation of treatment, even though it is documented that without treatment, their lifespan is typically reduced by 15 years compared with the general population.1

'Like Dr Whybra et al physicians in the UK recognise that females can indeed carry a significant burden of Fabry disease requiring swift and correct diagnosis and management by an expert centre. This long term study allows us to better understand how women suffering from the symptoms of Fabry disease may benefit from enzyme replacement therapy with agalsidase alfa and is consistent with our findings in the female patients we treat here,' said Dr Derralynn Hughes, Senior Lecturer in Haematology, Lysosomal Storage Disorders Unit, Royal Free Hospital, London.

'Until the year 2000 Fabry disease was rarely recognised as causing symptoms in women. The findings of this clinical trial recognises that women can be symptomatic for Fabry disease and the burden of their disease can be very debilitating. This is what our Fabry members have been telling us for years,' said Christine Lavery, Chief Executive, Society for Mucopolysaccharide Diseases.

Key findings from the study were: Agalsidase alfa stabilises kidney function, improves heart function and reduces pain and severity of disease in Fabry women.

Kidney function
Stabilisation or improvement in kidney function was found in more that 90 percent of the women in the study. Average eGFR (estimated glomerular filtration rate) was 91 +/- 31.2mL/min/1.73m2/year at baseline and remained stable throughout the study. A subgroup of patients with moderately reduced renal function at base line (Stage 2 CKD, eGFR>60 and < 90ml/min/1.73m2) demonstrated a significant increase in eGFR after 1 year of treatment, which was sustained through 4 years. Reduction in proteinuria was also noted in patients with baseline excretion of >300mg/day.

Cardiac structure and function
LVM was significantly reduced in patients with baseline LVH after 1 year of REPLAGAL and remained reduced through the four years, with 52% of them showing decreases of LVM in excess of 20%. In addition, treatment with REPLAGAL resulted in clinical improvement of symptoms of heart failure for nearly one-third of the patients who were classified as having NYHA Class III heart failure at baseline. After four years of treatment with agalsidase alfa, only one patient remained in NYHA classification III, and no patients progressed onto a more severe stage of heart failure.

Pain and burden of disease
Significant benefits with regard to severity of disease and quality of life were demonstrated. Mainz severity score index (MSSI) significantly improved after 12 months of treatment ($p < 0.01$) and showed continued improvement over 4 years. Importantly, 12 out of 36 patients (33%) exhibited decreases in MSSI scores that moved them to a lower range of severity. Reduced Brief Pain Inventory (BPI) 'pain at worst' was observed; score at baseline was 4.6+/- 2.9 and declined to 3.3 +/- 2.9 after 12 months (P=0.001).

REPLAGAL was well-tolerated during the study. One patient experienced mild infusion reactions. No anti-agalsidase alfa antibodies were detected at any time during the treatment period. Five women experienced a stroke during the study (three of which had a history of stroke prior to initiating ERT).

Study limitations
The study was an open label, observational clinical trial conducted in a single centre. This site is a referral centre, and therefore, the patient population may not be representational of the general female Fabry disease population. The study did not include a concurrently followed untreated control group, which limits the strength of the conclusions regarding the effect of agalsidase alfa on organ-system involvement in women with Fabry disease.

Study background
Published data represents analysis of a prospective, single-centre, open-label clinical trial that was performed to evaluate the long-term response of female Fabry patients to enzyme replacement therapy. All enrolled patients were treated with agalsidase alfa for 48 months.

Agalsidase alfa was administered at a dose of 0.2mg/kg infused over 40 minutes every other week. Patients were assessed at baseline and at 12 month intervals. The following measurements were performed at baseline and at 12-month intervals: eGFR and proteinuria, plasma Gb3 and urine Gb3, left ventricular mass and New York Heart Association functional score, Brief Pain Inventory (BPI) and Mainz Severity Score Index (MSSI).

eGFR was calculated using the abbreviated MDRD equation. A responder analyses was performed comparing final and baseline assessments and was expressed as mL/min/1.73m2/year. Proteinuria was based on a 24 hour urine collection. LVM was expressed by standard echocardiographic techniques, calculated according to Devereux and was expressed as LVMi in g/m2.7.

Shire Press Release www.shire.com

Regenocyte: Stem Cell therapy successfully reverses effects of Fabry disease

BONITA SPRINGS, Fla., Feb. 11, 2009 – An international team of physicians and scientists have discovered a way to treat cardiomyopathy (heart disease) with adult stem cells, including a rare metabolic condition otherwise requiring heart transplant.

Florida-based Regenocyte Therapeutic is using stem cells extracted from patients' blood to repair damaged heart muscle, regenerate tissue and create new vessels to improve circulation. According to the organisation's director of Cardiology and Vascular Disease, Zannos G. Grekos, M.D., by applying specific growth factors to the patient's stem cells (in a lab) the team creates a new cell population which is educated to target the area of damage or deficiency when placed into the patient's heart and blood vessels.

'We've now treated close to 100 patients with their own stem cells and seen an average 22 point increase in ejection fraction (EF) with a significant improvement in heart failure classification – typically from a Class IV to a Class II status in less than 180 days,' Grekos states.

The cardiomyopathy treatment study, the first six months of which was published in December 2008 in Anti-Aging Medical News, follows patients through one year post-treatment with autologous adult stem cells, also called Angiogenic Cardio-Regenerative Progenitor cells (ACPs). Regenocyte's chief medical advisor Athina Kyritsis, M.D., announced that 'Across the board, no adverse effects from treatment were reported by patients and function plus quality of life measurably improved.'

Grekos and his team measured patients' heart function by cardiac nuclear scans, PET scans and echocardiographs.

Case Study: Cardiomyopathy from Fabry Disease
Robert Pleva of Fort Myers, Florida, aged 60, suffers from Fabry Disease, a rare enzyme deficiency that leads to multiple organ failure. His heart has been damaged by an attack in 1999. In addition to cardiomyopathy, Pleva had chronic high blood pressure, pulmonary hypertension (a severe lung condition) and mitral valve issues. He was living on kidney dialysis, awaiting a heart transplant with the hope that a new heart would make him strong enough to qualify for a kidney transplant.

Robert Pleva was treated with adult stem cell therapy in June 2008. Regenocyte announced that Pleva's ejection fraction has increased from 28 percent before stem cell treatment to 44 percent as of January 2009, six months after stem cell treatment. 'This was a case where the patient's only option for survival was heart transplant and that is no longer the case,' said Dr Grekos. 'We couldn't be more pleased with this outcome. He's off the heart transplant list and continuing to improve. Bob's dialysis time has been reduced by 10 percent, so we are looking at treating his kidney function as a next step.'

Pleva says the improvement in his health since adult stem cell therapy has been dramatic. 'I feel so good and have so much energy,' he explains. 'The best part about it is being able to do things I haven't done in a long time.'
An electrician with the Lee Country, Florida School Board for the past 26 years, Pleva describes concern prior to treatment that his debilitating symptoms would soon end his career. He is now back to working full time along with riding motorcycles, remodelling his house and working in his yard. 'The treatment has put me back on track,' he says.

Pleva's wife Roxanne, who he cites as his 'rock', says in addition to his increased energy, one of the biggest changes is in the reduction in the amount of her husband's medication. 'His blood pressure has come way down, so he's been taken off many of his prescriptions,' she explains. 'We're just taking it one step at a time… and I'm getting my husband back.'

Regenocyte Therapeutic is the first stem cell clinic in the United States to move beyond research and successfully treat end-stage diseases with adult stem cells. In addition to cardiac and vascular conditions, they have used adult stem cell therapy to help patients with severe pulmonary disease, early senile dementia and macular degeneration.

Paul Schwartz, Chief Operations Officer, says the company's progressive technology and stringent protocols contribute to the high level of efficacy.

'The patients' safety comes first,' he explains. 'We adhere strictly to ISSCR (International Society for Stem Cell Research) and World Health Organisation guidelines, and only use FDA-approved biologic factors. We believe we've found the right combination of biotechnology and medical expertise to advance adult stem cell therapy as the treatment that changes the future of medicine, particularly in dealing with diseases considered to be untreatable.'

Source: Regenocyte Therapeutic

New Study in Women with Fabry Disease, Demonstrates Significant Benefits of Treatment with Replagal on Kidney Function and Burden of Disease

In a Press Release issued by Shire Pharmaceuticals in June, Shire plc announced that results from an observational study published in Genetics in Medicine demonstrate that enzyme replacement therapy (ERT) with Replagal 0.2mg/kg is effective in treating some of the signs and symptoms of Fabry disease in women. Clinical benefits were measured, including stabilisation of kidney function and improvement in patients with stage 2 CKD, improvement in cardiac structure in some groups of patients, and reduction in pain and burden of disease. Authors concluded that this observational study, which included 36 women and lasted 4 years, indicates that women with signs and symptoms of Fabry disease should be considered for ERT with agalsidase alfa, noting that the patient population studied may not be representative of the general Fabry disease femail population.

Professor Christoph Kampmann MD, co-author of the study at the Centre for Lysosomal Storage Diseases. Children's Hospital, Mainz, Germany stated ' Our research has generated meaningful, measurable data which shows that long-term treatment with agalsidase alfa stabilises kidney function and improves heart function. This may result in important benefits for Fabry Women, such as the reduced risk of heart failure'.

The study is published in full in Genetics in Medicine Volume 11, Number 6 2009

Physiological principles underlying Intrathecal enzyme replacement therapy in Lysosomal storage diseases

Currently there are no reliable methods of therapy for most of the Lysosomal storage diseases associated with severe neurological manifestations. Intrathecal therapy, with enzyme being injected directly into the cerebrospinal fluid (CSF) of the cisterna magna, is being investigated in several forms of mucopolysaccharidosis, including MPS IIIA in mice and dogs, using models maintained by the Lysosomal Diseases Research Unit (LDRU) at the Adelaide Women's and Children's Hospital (mice) and Massey University (dogs). Stage I human trials are starting for MPS I patients.

The MPS Society in collaboration with the patient association for Lysosomal Storage Diseases in New Zealand has agreed to fund a research project seeking to investigate the routs of dispersion of enzyme from the site of the injection and its absorption into the central nervous system (CNS) in the dog model of MPSIIIA. The methodology will include Computerised Tomography (CT) and in particular, immuno-staining of recombinant human heparin sulphamidase (rhHSm) in a time/course experiment following injection, using both light and confocal microscopy. The rationale is that any new therapy should be scientifically proven and the underlying physiology understood so the clinical trial protocols can be optimised and adverse reactions minimised. This project will be led by Professor Robert Jolly, Emeritus professor and Professor H Blair, Acting Head at Massey University, Palmerston North, N. Zealand

Stem Cell Gene Therapy for Sanfilippo Disease, MPS III

1st Year Report, Alexander Langford Smith

Outline

Bone marrow or cord blood transplant does not treat the severe brain degeneration observed in Sanfilippo (MPS III A). We propose that this is because enzyme cannot pass from the blood into the brain and transplanted cells that move to the brain produce insufficient enzyme. We are developing a stem cell and gene therapy approach to overexpress the decificient enzyme, sulphamidase (SGSH) in haematopoietic cells. When transplanted cells move to the brain they will now produce more enzyme. We are studying this in the MPS IIIA mouse model.

Progress

We have optimised the isolation of bone marrow and enrichment of haematopoietic stem cells. We have successfully produced lentiviral vector and optimised the transduction of the haematopoietic stem cells to overexpress SGSH.

Initial bone marrow transplants have been performed and a good survival rate achieved. A three-fold increase in normal SGSH has been achieved in the spleen and a two-fold increase in the liver. This has reduced storage of heparan sulphase (the stored molecule in MPS IIIA) in these organs to undetectable levels by tandem mass spectrometry. We have also shown that a normal bone marrow transplant does not increase SGSH levels significantly in the brain, whilst we can achieve 10% of normal SGSH levels 5 months after transplant of bone marrow overexpressing SGSH from the lentiviral vector. We are starting our evaluation of heparan sulphate storage and behavioural effects to see if this is sufficient for disease correction.

Presentations

I presented my work at this year's annual British Society for Gene Therapy conference in Royal Holloway, where it was well received and it was also presented at the MPS Society's biannual conference in Northampton.

Baseline characteristics of patients in the Fabry Registry

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

Fabry disease, also known as Anderson–Fabry disease, is a rare, progressive and life-threatening inherited disorder, occurring in approximately 1 in 117,000 live births.1 It results from a defect in the gene for an enzyme that breaks down a substance called GL-3, so that GL-3 accumulates in and damages the cells.2 The progressive nature of the disease means that some effects on the body will not produce any symptoms for a long time; however, ultimately, there can be substantial damage to the heart, kidney and brain.3 There is growing evidence that Fabry disease is under diagnosed, possibly because symptoms and complications are non-specific and so are often mistaken for similar abnormalities resulting from more common disorders.4,5 Furthermore, there is still much to learn to improve the diagnosis and optimise the treatment of this disease. To this end, the Fabry Registry – an ongoing global observational database for monitoring patients with this disease – has been set up (www.fabryregistry.com).

As so few people have Fabry disease, it is important to collect information from both treated and untreated individuals to enable the medical community to learn about the natural course of the condition and the effects of treatment. The Fabry Registry collects data on patients with Fabry disease when they are first diagnosed and continues collecting clinical findings to see how their disease progresses over time, to gather data on treatment responses, and to help optimise patient care for this rare disease. This article describes the initial (baseline) results of this Registry.4 It is important to note that:

- All participating patients have a confirmed diagnosis of Fabry disease and volunteer to participate via their treating physician, irrespective of whether they are receiving treatment. All information is anonymised and remains confidential. The database is maintained by Genzyme Corporation.
- All physicians participating in the registry can access their patients' data or request analyses of larger data-sets to help answer medical questions. Comprehensive monitoring of Fabry patients, regardless of age, sex, or treatment should be conducted at regular intervals according to a minimum recommended level of assessments. This includes historical medical records, test results and procedures to evaluate stroke, heart disease, kidney disease, and symptoms like pain or those affecting the skin, eyes and digestive system.4
- The content, analysis and output from the Registry are overseen by independent expert physicians in Fabry disease.4

The Fabry Registry programme began in April 2001. Here we report the results from the first 1765 patients who had enrolled by December 2005, encompassing 34 countries (though most patients were from Europe or the USA), and 162 treating physicians.4 Ninety-two percent of patients participating in the registry were known to have relatives with Fabry disease. As such, when someone is diagnosed, it is advisable to also screen family members to see if they also have Fabry disease.

A substantial gap was observed between age at symptom onset and that at diagnosis for Fabry disease in both males (14 years) and females (19 years) (Figure 1).4 (Note that this figure shows median values that are sometimes used instead of an average; median means the middle value.) Median ages at symptom onset and diagnosis were 9 and 23 years (males) and 13 and 32 years (females), respectively.4 This long delay between the onset of early symptoms and a diagnosis indicate the difficulty of attributing early, non-specific symptoms to Fabry disease by physicians.

There is also growing awareness that Fabry disease can have a similar effect in females as males, and that the disease follows a similar course in both sexes, but that effects are delayed in women.4, 6 Figure 1 also shows such a delay in female patients: median onset of symptoms occurred 4 years later and median age at diagnosis was 9 years older in females than males.4 This contrasts with views held some years ago: that females acted just as carriers of Fabry disease and were clinically unaffected. In fact, females with Fabry disease are at substantial risk of Fabry-associated symptoms.5

Figure 1. Age at onset of Fabry symptoms and at diagnosis of Fabry disease.

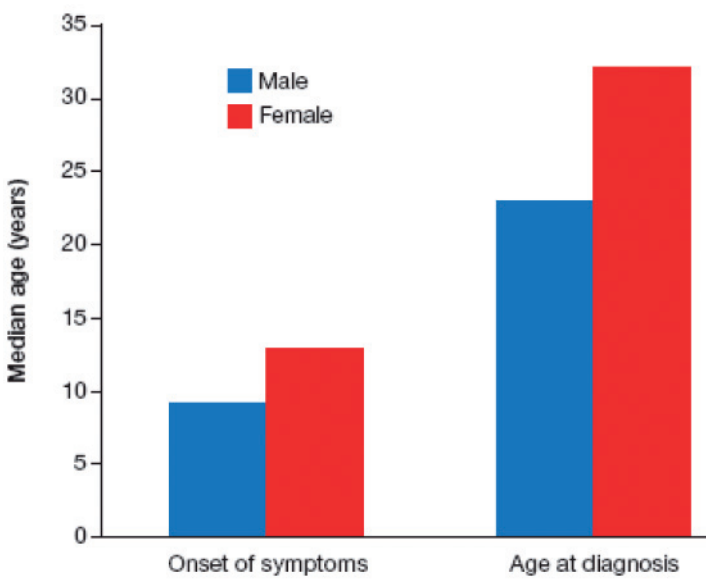
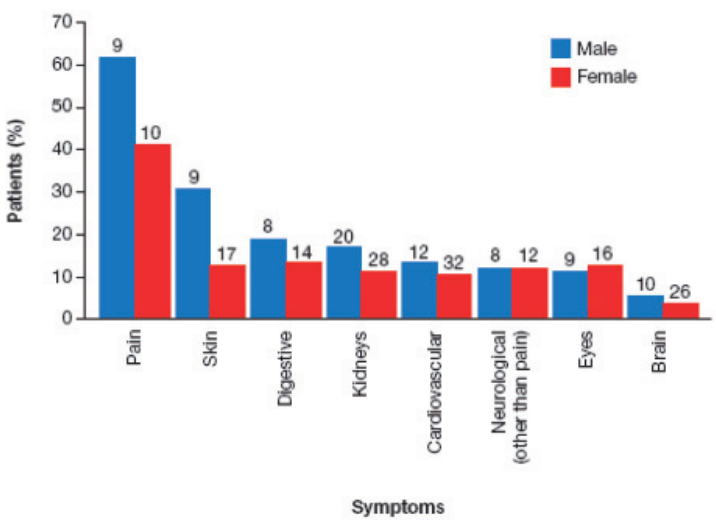


Figure 2 shows the most common first symptoms at Fabry disease onset, as reported by males and females enrolled in the Registry. In general, male and female patients reported a similar profile of initial Fabry symptoms with only minor differences. Pain was the most common onset symptom in males and females (62% and 41%, respectively). It is also notable that the incidence of potentially very serious indicators, such as cardiovascular symptoms and symptoms affecting the brain (e.g. stroke) were equally common in males and females, whilst renal symptoms were somewhat more common in males (17%) than females (11%). Note that for all symptoms, the median age of female patients was delayed compared with their male counterparts (Figure 2).

Figure 2. The proportion of patients with first Fabry disease symptoms (patients may have presented with more than one). (Median age at onset of symptoms [years] shown above each bar.)



Data of Fabry patients have been collected in the Fabry Registry regarding the age at which late, serious complications or events occurred in the kidney (i.e. dialysis or transplant), heart (e.g., heart attacks, heart failure, angina, heart surgery) or brain (i.e. stroke).

- Similar proportions of male and female patients had cardiovascular (heart) events (19% and 14%, respectively) or cerebrovascular (brain) events (7% and 5%, respectively), but more males (13%) than females (2%) had a renal (kidney) event.
- For those reporting declining kidney function, median age at occurrence was comparable (39 years for men, 37 years for women), but the onset of cardiovascular and cerebrovascular events was somewhat later in women (47 and 43 years) than in men (41 and 38 years, respectively).

To date, the Fabry Registry represents the largest effort to investigate Fabry disease, as described by patients and their physicians, and covers all age groups and parts of the world and enrols patients irrespective of their treatment status. As increasing numbers of patients agree to participate in the Fabry registry (more than 3000 Fabry patients are currently enrolled) it is expected that more important data analyses will be performed regarding the course of Fabry disease, both in treated and untreated patients. This will allow the effectiveness of interventions such as enzyme replacement therapy to be assessed and compared in different populations. Patient participation in the Registry, therefore, is fundamental to providing results that can increase awareness of common symptoms in all age groups, and lead to improved recognition of the disease, earlier treatment and the potential for improved outcomes for everyone.

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Declaration of interest: The author is employed by Genzyme Therapeutics, Oxford, UK.

Surgery in patients with MPS I: Results from the MPS I Registry

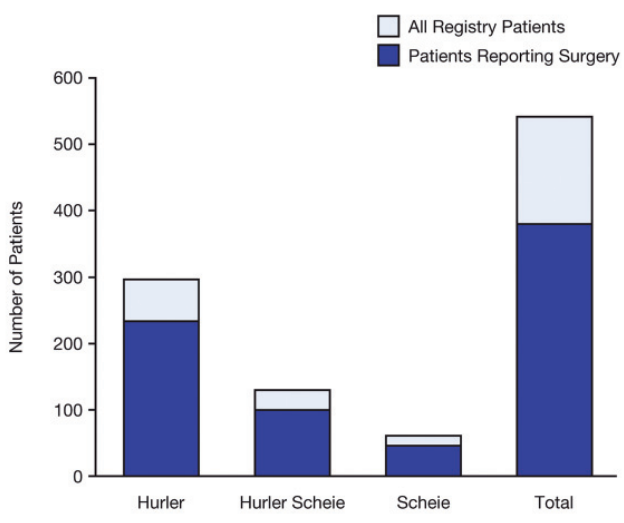
Emma James DPhil (Oxon)
UK Registry Co-ordinator, Genzyme

Arn P, Wraith JE, Underhill L. Characterization of surgical procedures in patients with mucopolysaccharidosis type I: findings from the MPS I Registry. J Pediatr 2009;154(6):859-64 e3.

Mucopolysaccharidosis type I (MPS I) is a rare and life-threatening inherited disorder, which is now treatable by enzyme replacement therapy or stem cell transplantation.1,2 MPS I is divided into three syndromes of increasing severity: Scheie, Hurler-Scheie and Hurler, with Hurler syndrome representing the most severe form. Because of the rarity of this disorder, the MPS I Registry has been set up to track the clinical onset, symptoms, treatment and outcomes of people with MPS I of any severity.3 This ongoing, international and voluntary disease registry programme collects routine clinical and laboratory data from individuals with MPS I. Currently, over 800 individuals with MPS I are enrolled. Here, the recently published results regarding surgery in MPS I Registry patients are summarised.4 Dr Pamela Arn and colleagues analysed surgical data collected from all of the 544 people who were participating in the MPS I Registry on or before October 2006. Surgical procedures were divided in 21 categories for analysis. Clinical procedures related to MPS I treatment (for example, bone marrow transplant) or monitoring were excluded from the analysis.

Surgery was very common in people with MPS I: 391 of 544 individuals (72%) underwent at least one surgical procedure, and a total of 1694 surgical procedures were reported. The median number of surgical procedures was four per patient of those who had surgery. (A median value is a kind of average, and just means the middle value.)
The overall burden of surgery was similar across the three MPS I syndromes and the proportion of those who had surgery was similar in those with Hurler (80%), Hurler-Scheie (79%) and Scheie (74%) syndromes (Figure 1).

Figure 1. The number Registry patients with each MPS I syndrome, and the proportion of these undergoing surgery. Note that 391 of 544 (72%) patients in the Registry reported 1694 surgeries.

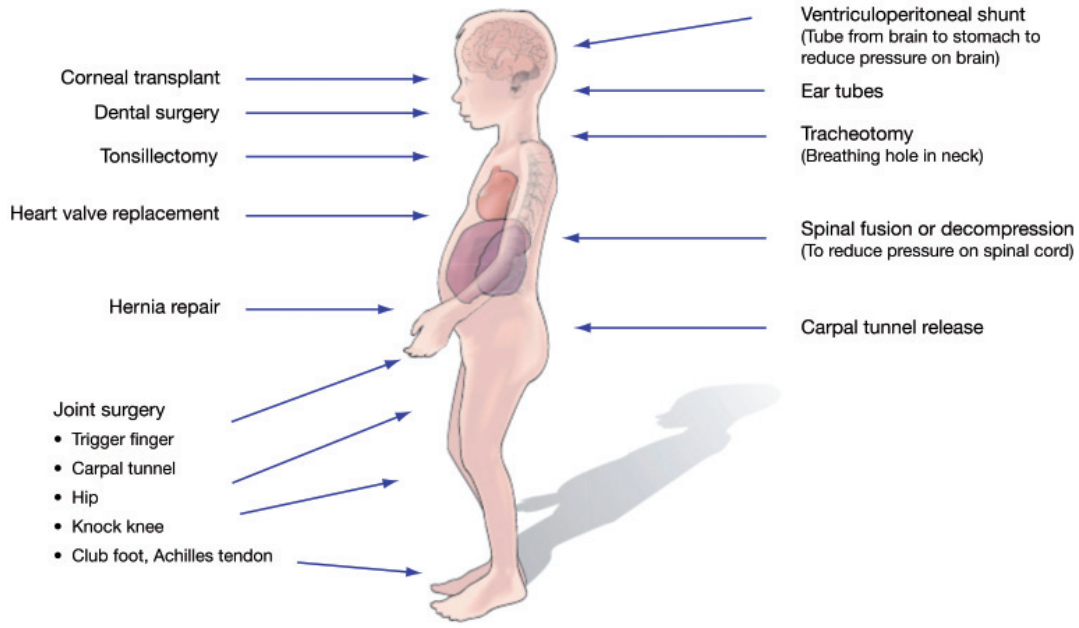


One important feature is the young age at which surgery was performed: by the age of four years, nearly half of all patients had undergone two or more surgical procedures. Another interesting finding from this study was the extent to which surgery was carried out before a diagnosis of MPS I was made. One or more surgical procedures preceded MPS I diagnosis in 36%, 46% and 63% of those with Hurler, Hurler-Scheie and Scheie syndromes, respectively. It is important that surgeons know which patients have MPS I, as these individuals may have compromised airways,5,6 and this potential risk during anaesthesia needs appropriate management.

The types of surgery performed most frequently were ear, nose and throat surgery (43% of all surgeries), abdominal/genitourinary surgery (21%) and orthopaedic surgery (19%) (Figure 2). Specifically, the most common surgeries in those with MPS I were myringotomies (a tiny incision in the eardrum to relieve pressure caused by the excessive build-up of fluid), hernia repair, and adenoidectomy/tonsillectomy. These are also the most common surgeries in non-MPS I affected children in the general population.

However, each of these surgeries is reported in at least one-third of MPS I Registry patients – which is a greatly increased number compared with that found in the general population. Of particular interest is the age at which these three surgeries are performed in patients with MPS I compared with children in the general population. Adenoidectomy/tonsillectomy is performed at a much younger age (average age of 2.8 years) in those with MPS I compared with an average of 7 years in the general population, whilst hernia surgery is more often performed at an older age (median of 2.5 years) in patients with MPS I than in the general population (where most hernia procedures in children are performed before 1 year of age). In addition, repeat hernia operations were more common in patients with MPS I than in the general population. Carpal tunnel syndrome (a pain or weakness in your forearm and hand caused by pressure on a nerve in your wrist) is unusually common in children and adolescents with MPS I: it was among the five most common surgeries reported in all three syndromes. In contrast, this condition is very unusual in non-adults in the general population. In fact, when carpal tunnel syndrome does occur in children, an MPS disorder is one of the most common causes.7

Figure 2. Common surgeries in people with MPS I.



The results from this analysis of surgery in the MPS I Registry provide 'alarm' signals to help doctors recognise possible MPS I and diagnose this condition earlier. These may include:

- adenoidectomy/tonsillectomy performed at, or before, 5 years of age
- hernia after the age of 1 year, or any child undergoing a repeat hernia surgery
- carpal tunnel syndrome in a child of any age
- or two or more unrelated surgeries before 4 years.

In the presence of any of these signals, physicians should consider the possibility of an MPS disorder especially if they also see any other common indicators of MPS, such as coarse facial features, enlarged tongue, corneal clouding (cloudiness of the usually clear front of the eye, reducing sight) and joint stiffness, as well as any abnormalities in the valves of the heart.

If MPS I can be diagnosed at an early age this will allow prompt treatment. It also alerts surgeons and anaesthesiologists to special surgical management needs (which would otherwise be overlooked if MPS I went undiagnosed), allowing safer and more successful surgery. The ongoing commitment of people providing their data is crucial to sustaining the success of the Registry programme, and this kind of information will help improve the lives of all individuals with MPS I disorders.

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Declaration of interest: The author is employed by Genzyme Therapeutics, Oxford, UK.

Turkish MPS Society
'A Dream Come True'

On 4 July 2009, 34 people from various parts of Turkey met up in Istanbul in a conference room of a hotel. They elected the management committee of The Turkish MPS Society. After many years of waiting, we now have an MPS Society in Turkey. This is a personal dream come true for me.

It has been very difficult to establish the Society, because the rules regarding charitable organisations state that there should be an office open to members and public as well as inspectors.

Apparently, there are companies in Turkey who specialise doing the necessary paperwork and registering charitable organisations to their address. It was with the help of Biomarin Turkey representative Bertrand Souleres that one of these companies was contacted and the Society was set up. His office also organised the MPS families' get together in Istanbul.

Without his help, the Society would not have been set up at this moment in time. The Society and I are very much indebted to him. I also would like to thank Christine for supporting me whenever she could to make this happen. We will very much need her help in the future as well.

I am constantly in touch with the Chairman, Nalan Cetin. She is the mother of a Hunter boy and is also from the city where I come from, Izmir.

Their website is being prepared. Their logo has been designed ready for the committee's approval. Nalan has had several newspaper interviews and appeared on local TV. She has been to see the consultant at the big regional hospital and is in touch with several families and several patients are on ERT.

I wish them all the best of luck in their new endeavour and will support them as much as I can. **Fer Pidden**

WORLD Symposium 2010

The WORLD Symposium 2010, co-presented by the Lysosomal Disease Network and the National Institute of Neurological Diseases and Strokes, is approaching.

WORLD Symposium 2010 – 2/10/2010-2/12/2010 – University of Minnesota – Hilton Miami Downtown; Miami, FL – The Lysosomal Disease Network is a research 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.

Topics: - Newborn Screening – New Advances and Therapies in Gaucher Disease – Fabry Disease – Mucopolysaccharidosis – Batten Disease – Pompe Disease – Mucolipidosis – Sphingolipidoses – Oligosaccharidosis

For more information: Office of Continuing Medical Education University of Minnesota Phone: 612-626-7600 or 800-766-8636 / Fax: 612-626-7766 University Park Plaza, Ste 601 2829 University Ave SE Minneapolis, MN 55414, USA E-mail: cme@umn.edu
Web: www.LysosomalDiseaseNetwork.org or www.cmecourses.umn.edu.

The Kakkis EveryLife Foundation

Dr Emil Kakkis was most supportive of the MPS Society in his capacity as Medical Director of BioMarin. Last November Dr Kakkis left BioMarin to set up his own Foundation with the primary aim of:

- Establishing a new Office of Drug Evaluation for Genetic and Biochemical Diseases at the FDA, consolidating expertise to ensure safe, effective and timely patient access to needed treatment.
- Create a new standard for surrogate measures of treatments for rare disorders, and allow these treatments full access to the accelerated approval pathway for life threatening diseases.
- Devise new clinical study designs for rare diseases that account for disease complexity to properly capture treatment effects on all aspects of the disease.

The focus of the Foundation is not about changing safety standards but to level the playing field between large market drugs in which surrogates are commonly allowed and to improve the way efficacy is assessed for clinical disease using novel study designs appropriate for rare diseases. The strapline for the Foundation is 'Cure the Process'. For more information visit **www.kakkis.org**

Elaprase Approval in New Zealand

LDNZ was very pleased to announce on 2 April 2009 that they have successfully achieved the first new approval for an Enzyme Replacement Therapy for a Lysosomal disease since Gaucher disease treatment was approved over a decade ago in New Zealand. Pharmac has confirmed to us that the exceptional circumstances application for a young boy with Hunter disease has been approved for a two year period. Treatment with Elaprase is expected to commence within a few months.

This is a significant breakthrough for LDNZ and the patients we represent. We have worked very hard over many years to build an environment where a positive decision would be achieved despite the very difficult situation regarding medicine funding in New Zealand. However this is just one treatment for one patient, so there is a lot more work to be done to ensure other NZ patients also gain appropriate access to these therapies.

PTAC (Pharmac's technical advisory committee) have not recommended a general listing of these new treatments on the pharmaceutical schedule, but indicate that a case by case assessment through the exceptional circumstances application system should be preferred. That is obviously a tactic they have chosen in order to minimise the financial impact on their budget, but it seems to us they may not have had all of the most up-to-date information on which to base their recommendations. Anecdotally we know of studies that are being done on various patient groups with these diseases, and given the very strict evidence-based approach Pharmac always takes, it will be essential to have all of the most recent data available if their views are to change towards a more liberal approval.

On 19 August 2009 Jenny Noble from LDNZ wrote to share with us a very auspicious day in New Zealand. After 10 years of advocacy she was very pleased to announce that Jack Peacock, Hunter Syndrome, had received his very first infusion of Elaprase today.

We were delighted to hear that Jack is doing really well and so far has no side effects. The family are on cloud 9 and can't wait to see the changes taking place.

This is such an amazing turning point in New Zealand and LDNZ recognise there is still much work ahead to get patients with Pompe and then Fabry treated.

We are very excited to get to this point in the funding issues and know that we still have much work ahead of us.
Jenny Noble Lysosomal Diseases New Zealand

E-mail: jenny.noble@xtra.co.nz
Website: www.ldnz.org.nz

Did you miss the Vancouver Symposium? Or were you unable to catch all the sessions while you were there?

All scientific sessions were videotaped and are available for viewing, along with their accompanying slideshows, at www.goldinfo.org.

To view the sessions on the GOLD website, you will need to register if you have not done so before. Registering only needs to be done once. Click on 'Video Presentations' under 'Education and Information' on the GOLD homepage's left sidebar. Then, click on 'register here' in the first paragraph, which will open a registration form. You will be asked to give your email address as a username, choose a password and select the organisation of which you are a member: Please select Canadian Society for Mucopolysaccharide and Related Diseases Inc. from the drop-down menu. GOLD will not give your information to third parties. All presentations from the family sessions are available for download at www.mpssociety.ca

Taiwan MPS Society celebrates MPS Awareness Day 2009

The Taiwan MPS Society celebrated International MPS Awareness Day on May 15th this year by presenting "Appreciation Awards" to ten dedicated volunteers. Last year, we presented this award to three MPS doctors. This was the second year of making presentations with the primary goal of raising awareness of MPS. We will continue giving out awards to different fields that help MPS children in any way. Next year, we are planning to give the awards to MPS families that help bring a good attitude in our MPS Society.

Noah’s Ark Children’s Hospice

Noah’s Ark is a children’s hospice service supporting life-limited children and their families to live positive lives together by providing practical help and emotional support.

Currently the Noah’s Ark Team is committed to providing help for families in their own home. The service is small but growing steadily and over 50 families are already benefiting. Noah’s Ark will eventually have a hospice building and help families living in the London Boroughs of Barnet, Enfield, Camden, Islington and Haringey.

In partnership with Barnet and Enfield Primary Care and Hospital Trusts, Noah’s Ark is funding a play specialist and a respite nursery nurse to provide sessions in the family home. They offer play for the children and support them in coping with treatments or other anxieties. Parents may join in the sessions or take a break for themselves. Brothers and sisters can always be included!

The Volunteer Co-ordinator is recruiting and training a team of family support volunteers to give a helping hand to families. They can offer practical help such as shopping, light housework, gardening or driving. They can be an extra pair of hands for a family, playing games with the children or helping the whole family to enjoy a day out together.

A part-time social worker is available to give extra help to a number of families. She can also be contacted by families and professionals for advice and signposting to other services.

Families registered with Noah’s Ark will receive invitations to outings and other special events held several times a year. These events are free and transport can be provided.

During 2009 a bank of trained carers is being set up to work alongside parents or to look after the children while parents have a short break.

For more information please contact:
Noah’s Ark Children’s Hospice, Ganwick House, Wagon Road, Barnet, Hertfordshire EN4 0PH
Tel: 0208 449 8877
Email: info@noahsarkhospice.org.uk
Website: **www.noahsarkhospice.org.uk**

Hunter Disease eClinic

This amazing new educational software, developed by Sick Kids’ Lysosomal Research Group and launched at the Canadian MPS Society’s International MPS Symposium, is now available through the Hospital for Sick Children’s website. The direct links to this educational software are:

eBook: http://www.sickkids.ca/research/lysosomalresearchgroup/contents/Hunter/Hunter_eBook.asp

eClinic: http://www.sickkids.ca/research/lysosomalresearchgroup/contents/Hunter/Hunter.asp

Users need “Flash Player” installed on their computers in order to be able to view the program, and should be reminded not to use the browser ‘Back’ button.

The Hunter eClinic can also be accessed from the Lysosomal Research Group’s website under “NEW: Educational Resources” on the group home page: http://www.sickkids.ca/lysosomalresearchgroup/

Family Fund extra

The Family Fund helps families with severely disabled children and young people to have choices and the opportunity to enjoy ordinary life.

We give grants for things that make life easier and more enjoyable for the disabled child, young person and their family, such as washing machines, driving lessons, computers and holidays.

We will consider any grant request that will make a difference the lives of a disabled child, young person and their family.

Families can apply for things from attraction tickets to zoo trips, but we also encourage requests from young people that have particular meaning to their age group such as driving lessons, laptops and equipment for college.

The Fund is a registered charity, helping around 50,000 families in the UK with £30.5 million in grants a year.

The Family Fund (www.familyfund.org.uk) has expanded its eligibility criteria to help more disabled children and young people. The age limit has been raised from 16 to up to 18th birthday, the income limit remains at £23,000.

The Family Fund is always looking for new ways for disabled children's families to keep their costs down.

Family Fund extra is a new online buying and lifestyle club that will help the families of the 770,000 disabled children across the UK make their money go further.

Separate to the Fund's grant making, **Family Fund extra** will help all families with disabled children, as well as disabled young people themselves, enjoy online discounts of up to 25 per cent from leading retailers like Comet, Argos, Haven Holidays, BSM and Stone Computers.

Extra click brings extra help.

You can help the Family Fund to raise money so we can help more families by clicking on extra every time you shop online. Family Fund extra has over a hundred leading retailers registered such as Amazon, Tesco, M&S, Thomas Cook, HMV and John Lewis who give us money back when you shop with them at no extra cost to you!

To find out more, go to **www.familyfundextra.org.uk**

FOR SALE!

Convaid Safari Tilt wheelchair. Transport model. Size 16" to fit child aged 12 upwards
Compact folding wheelchair that tilts in space. Degree of tilt adjusts from 5° to 45° for stabilising and feeding. Crash tested and can be used on school transport. Comes with 5 point harness, swing away footplates. Bought new Dec 07 £1600, Excellent condition.
£500 ono. Telephone **01225 764851**

The Challenging Behaviour Foundation

The Summer issue of 'Challenge' (newsletter of the Challenging Behaviour Foundation) is now available, this issue focusing on education for children and young adults with severe learning disabilities.

Policy and practice regarding children with learning disabilities have changed radically in the past 40 years. In the main feature, 'Home or away', Peter McGill (University of Kent Tizard Centre) examines recent research and issues for future provision, coming to the surprising conclusion that, despite the move away from long-stay hospitals and policies of inclusion, we are now excluding from their local communities nearly as many children with learning disabilities as we did in the 1970s.

In accompanying articles, a parent and an independent special needs education provider debate whether out of area residential school placements are a "scandal" or "solution" for families. Peter McGill sets out three recommendations for change if we are to reduce the need for residential school and other residential placements: "it would clearly be much better if far fewer children and young people had to leave their families and travel halfway round the country in order to access specialist knowledge and experience."

Other articles include: work by Westgate College to provide individually tailored Further Education opportunities for a group of students with severe learning disabilities and challenging behaviour; information about the new Deprivation of Liberty safeguards and what this means for both individuals and care providers; news of a new national initiative led by the Challenging Behaviour Foundation: the Challenging Behaviour National Strategy Group; Regular articles include 'your questions', 'what parents say', new resources from the Challenging Behaviour Foundation, 'Comment' by Tony Osgood, Lecturer in Intellectual & Developmental Disability, Tizard Centre, University of Kent, and editorial by Vivien Cooper, Chair of Trustees and Founder of the Challenging Behaviour Foundation.

'Challenge', the newsletter of the Challenging Behaviour Foundation, is produced three times a year and is available free of charge by emailing: info@thecbf.org.uk or downloading from www.challengingbehaviour.org.uk

Helen Marron
The Challenging Behaviour Foundation

Email: info@thecbf.org.uk
www.challengingbehaviour.org.uk
General Enquiries: Tel. 01634 838739
Family Support Worker: Tel. 0845 602 7885 (individual telephone support for families at the cost of a local call)

The Challenging Behaviour Foundation is a registered charity (no. 1060714) supporting families caring for individuals with severe learning disabilities.

Visit **www.challengingbehaviour.org.uk** to make a donation today. Thank you for your support.

Is Disability Living Allowance (DLA) under threat?

Many disabled people are concerned that the future of DLA is under threat. The government has published a green paper looking at the future of the social care system called 'shaping the future of care together'. It proposes to look at how to pay and provide care and support to the growing population of older people and the number of young people with disabilities.

One of the proposals is that benefits such as Attendance Allowance (AA) are put into a central pot which would be allocated to local authorities to fund support services. However, many believe that DLA could also be included into this and this has not been ruled out.

For many people DLA along with AA is a life line to help pay for the extra provisions and equipment that is needed and not funded through social care.

The Child Poverty Action Group (CPAG) have claimed that they have had assurance from the DWP that DLA will not be involved but few people are satisfied with this as it has not come from government ministers.

Individuals and charities have been asked to make their representation and views known to government and have until the 13 November to do this.

If you would like to make representation regarding this matter, you can do so by either going to the Disability Benefits Consortium website www.disabilityalliance.org or going to the benefits and work website www.benefitsandwork.co.uk

Sharing the learning

Sibs

For brothers and sisters
of disabled children & adults

Sibling Group Leader Conference
Saturday 10 October 2009
10.00 – 16.30
City of London School, London

If you support siblings of children with disability or chronic illness this day is for you. In the morning, three different sibling groups will be demonstrating their group work, so that you can observe and participate in an active sibling workshop and real siblings.

For more information call
Chris at Sibs on **0114 2302548**

Changing the world for children with genetic disorders

Friday 2nd October 2009

Jeans for Genes Day 2009 takes place on **Friday 2nd October**. It's your chance to make a difference to thousands of children and their families across the UK who are living with genetic disorders. Register for a free fundraising pack at www.jeansforgenes.com and organise a Day at your office, nursery, school or club. For a £2 donation (£1 for children) everyone can wear their jeans, have fun and raise money.

Your pack contains inspiring ideas along a musical theme to help you raise even more. Everything from lunchtime karaoke, busking in the canteen to a pop quiz in the pub.

This year, the money you raise will benefit the MPS Society and nine other charities. You'll be helping to fund projects such as: retreat weekend for children with a rare premature ageing condition, Cockayne syndrome; a five day summer camp for children caring for a parent with Huntington's disease; two study days focussing on rare chromosome deletions, bringing together families and professionals to learn from each other.

The new collection of merchandise features a wonderful women's black cotton t-shirt with a gold-foil 'double helix' chain design. With matching cotton shopping bags and handbag-sized umbrellas, it's the design to be seen in this summer. It also comes in beautiful bright colours for girls, boys and men.

And for the youngsters there are four brand new Fifi and the Flowertot badges to collect. Featuring the TV character and her friends, the glow-in-the-dark badges will brighten up coats and bags. Buy them online at **www.jeansforgenes.com/webshop**

Among the celebrities supporting the Day this year are radio presenter Zoe Ball and her dad Johnny, supermodel Jodie Kidd, actress Jaime Winstone and singer/songwriter Lemar.

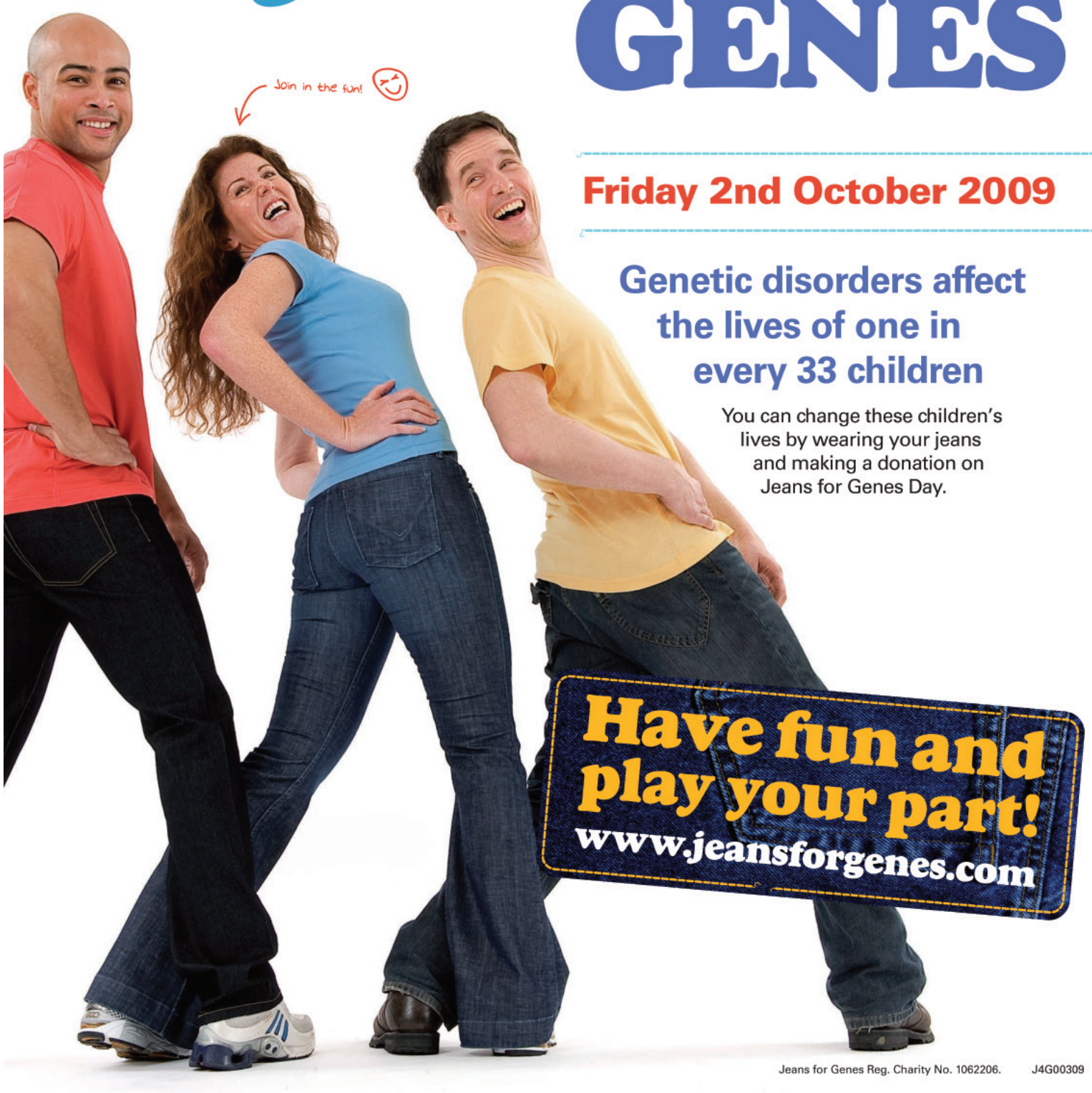
Sign up now and be part of the fun and fundraising on Jeans for Genes Day 2009 at **www.jeansforgenes.com**

Changing the world for children with genetic disorders

Jeans for Genes Day

We want YOU! for Jeans for Genes

GET BEHIND JEANS FOR GENES



Friday 2nd October 2009

Genetic disorders affect the lives of one in every 33 children

You can change these children's lives by wearing your jeans and making a donation on Jeans for Genes Day.

Jeans for Genes Reg. Charity No. 1062206. J4G00309