MPS

Society for Mucopolysaccharide Diseases

What's Inside...

Recent events including the MPS Disney Conference in Florida

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

Winter 2009

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

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

Front cover photos:
Josefine (MPS IV) and the Pollock Family at Disneyland USA

Society for Mucopolysaccharide Diseases

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Magazine Deadlines

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

Winter 2009

CHIEF EXECUTIVE'S REPORT

This time last year, the MPS Society's Board of Trustees presided over one of the most challenging budget meetings in recent years. Despite the enormity of the credit crunch and ensuing recession and thanks to so many of our members, corporate and charitable trusts and extreme prudence by the staff team, we have just about broken even. Having said that, we are going to need all your help this year with fundraising and donations to ensure we can continue to weather the recession where many charities are reporting an 18 – 20% drop in income.

As many of you know, the MPS Society has been a Jeans for Genes partner charity from the Jeans for Genes conception and this has enabled the MPS Society to fund pioneering and ground-breaking research that has or is likely to lead to therapeutic approaches for those affected by MPS, Fabry and related diseases. Four years ago at the peak of Jeans for Genes the Society received £450,000. Regrettably we have seen a considerable decline in income available to the beneficiary charities and may only expect to receive up to £140,000 from the 2009 Jeans for Genes Appeal. This huge drop in income cannot be allowed to translate into an equivalent drop in MPS research. Whilst we are committed to working with Jeans for Genes to reduce their cost/income ratio and generate levels of income previously received, we also need your help to identify other funders to fill this void too.

The MPS Society, whilst retaining its autonomy, strongly supports the efforts of the pharma industry in bringing to the market place new therapeutic approaches. In 2009, we have seen 20 children with MPS IVA, Morquio disease recruited to the MorCap Enzyme Replacement Therapy Clinical Trial in London, Birmingham and Manchester and we are hoping Phase III/IV will be underway later in 2010. In the last MPS Magazine I alluded to a forthcoming MPS IIIA, Sanfilippo A Natural History Study. Whilst progress is being made to roll out this study we are not yet able to offer further information. Rest assured though that once we are, those families whose child or children who have a diagnosis of MPS III, Sanfilippo, will be written to individually advising of the MPS IIIA Sanfilippo study. We appreciate that those families with children with Sanfilippo type B, C and D may also receive this information but we do not necessarily know the sub type of the Sanfilippo disease the children and young adults have.

There are other clinical trials taking place in the United States. For MPS I and MPS II intrathecal Enzyme Replacement Therapy is being explored for MPS I and further on in this magazine you can see details of the MPS II phase I/II intrathecal enzyme replacement therapy.

As we celebrated the festive season we recognise that again this year many of our members have in different ways suffered loss. Whether it be an MPS diagnosis, disease progression, the death of an MPS loved one or as the MPS family grows, the loss of non MPS family members, our thoughts are with you all as we move into 2010.

The Society, supported by its Board of Trustees and led by a wonderfully motivated staff team, have an exciting events programme for the coming year and we hope to see as many of you as possible at the MPS Family Weekend at Camelot, at the All Ireland MPS and Fabry conference in Templepatrick Belfast, the MPS III Expert Meeting and all the other activities in the Events programme featured in this magazine.

Finally, I know you would want to join us in sending best wishes for the future to Miriam Blowers and Sarah Irvine who left their employment with the Society in December. Miriam and her family say goodbye to the UK in January to start a new life in Australia. Sarah left with regret after her partner accepted a new job in Eastern England. We shall miss them both and in January we will be recruiting for Miriam and Sarah's successors.

Christine Lavery
Chief Executive

Winter 2009

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 Meeting held 11-12 September 2009.

Personnel

Election of Officers

In the absence of any staff member, the Trustees duly elected unanimously the Trustees to the following positions:
Chairman – Barry Wilson
Vice Chair – Bob Devine
Treasurer – Judith Evans
Wilma Robins was unanimously co-opted to the MPS Board of Trustees.

Risk Management

Chairman, Barry Wilson, reported that the Volunteer Handbook has now been completed and extended his thanks to the Senior Advocacy Officer. It was agreed that Trustees would review the MPS Risk Management Register at the next Management Committee Meeting. The Chief Executive and Finance Officer are working on the Asset Register, Business Continuity and Staff Handbook.

Policies

The Chief Executive advised the Board of Trustees that the policy review timetable had been updated. Trustees reviewed the policy on Board Diversity and acknowledged by virtue of the Society's constitution the Board does not reflect the ethnic diversity of the members of the MPS Society and will continue to review Board membership on an ongoing basis. It was agreed that a money laundering policy would be brought to the next Management Committee.

Jeans for Genes (J4G)

The MPS Board has had considerable concern over the governance of Jeans for Genes particularly over the past twelve months. After considerable discussion the Board agreed that professional advice should again be sought and the situation reviewed again at the next Board meeting. Due to the above the Society's independent Trustee on the Jeans for Genes Board has resigned.

MPS Research Grants

The Chief Executive Officer informed Trustees of recent discussions she has had with a potential high level donor interested in funding MPS III research at the Manchester Stem Cell Group. The Trustees also considered a proposal for a Brains for Brain fundraising initiative in the UK.

International Collaboration

It was agreed that the Chief Executive will participate in the WORLD LSD meeting in Miami in February 2010.

Thank you to all our supporters!

The MPS Society is very grateful to our fundraisers and supporters for all their hard work in raising money through organised fundraising events, sponsored events and other activities, big or small.

We are so appreciative of your support and for thinking of the MPS Society. We get a number of requests each year to attend cheque presentations or give talks on our work. We always like to do these when possible but to minimise the costs to our charity, try whenever possible to coincide these with other visits in the local area or en route to other meetings or events. Thank you to all our fundraisers for their continued and very vital support. We need you!

For more information or to request a fundraising pack, please phone us on 0845 389 9901 or email us at fundraising@mpssociety.co.uk.

Office of the Scottish Charity Regular (OSCR)
We are pleased to announce that the Society for Mucopolysaccharide Diseases is now a charity registered in Scotland no. SC041012.

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.

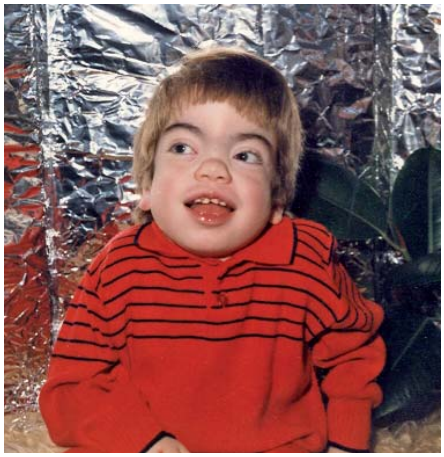
Visit **www.mpssociety.co.uk** to download the application or phone us now on **0845 389 9901**.

Members' Announcements

In Memory

It was with enormous sadness that we acknowledge the sudden death of Peter Robins, aged 62 years on Monday 25 May 2009.

Peter, husband of Wilma and father of Gethin (pictured below) who died aged five years in 1984 has been a huge supporter of the MPS Society since Gethin's diagnosis with Hurler Disease. Peter and Wilma have rarely missed an MPS Conference and Peter has unstintingly supported his wife, Wilma's, role as MPS Trustee since her appointment over 20 years ago.



Wilma continues as an MPS Trustee and we send our heartfelt sympathy at this sad and difficult time. To date, the Society is very grateful to have received donations totalling £1487 in memory of Peter. Peter is pictured below with Wilma at the Childhood Wood.

New Members

Mr Tonge has recently been in contact with the Society. Dylan has been diagnosed with alpha-Mannosidosis. Dylan is almost 4 years old. The family live in London.

Mr and Mrs Chowdhury have recently been in contact with the Society. Shahadat and Shafaat have a diagnosis of Hunter disease. Shahadat is 2 years old and Shafaat is 6 months old. The family live in the South West.

Deaths

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

Amie Oliver who suffered from ML III and who died on 3 May 2009 aged 18 years.

Santana McDonagh who suffered from ML II and who died on 3 June 2009 aged 2 years.

Faiza Shaikh who suffered from Sanfilippo Disease and who died on 24 October 2009 aged 21 years.

Mark Fitzgerald who suffered from Hunter Disease and who died on 1 November 2009 aged 32 years.

Nicole Pickard who suffered from Sanfilippo Disease and who died on 7 December 2009 aged 19 years.

Josephine Kembrey who suffered from Sanfilippo Disease and who died on 7 December 2009 aged 18 years.

Daniel Ellis who suffered from Sanfilippo Disease and who died on 20 December 2009 aged 18 years.

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

WellChild Awards 2009

On Monday 28 September 2009 the MPS Society attended the fifth annual WellChild Awards in London. This was an evening to celebrate some inspirational children and to reward the dedication of some of the many professionals who support and care for young people across the UK.

The winners for this year represent all that the Awards should stand for - courage, care and dedication. There were various awards presented on the night including Bravest Child in various age categories but also Awards to those in the medical profession such as Best Nurse and Best Medical Team.

We were delighted however that Dr Ed Wraith was winner of the Best Doctor award. Dr Wraith is a Consultant in Paediatric Inherited Metabolic Disease at Royal Manchester Children's Hospital. He is at the forefront of care and resarch, runs clinical trials and is widely respected by his peers and the families that he works with. One parent described him as 'a very special human being'.

We would like to congratulate Dr Wraith, pictured here with TV presenter Chris Hollins, on his award from all at the MPS Society. Prince Harry is Patron of WellChild and has attended the Award Ceremony for the past two years.

For more information on WellChild visit **www.wellchild.org.uk**

CONGRATULATIONS! to Professor Ed Wraith

After over two decades devoted to the clinical management and treatment of children with Mucopolysaccharide and other lysosomal storage diseases Dr Ed Wraith, Consultant Paediatrician specialising in Lysosomal Storage Diseases at the Royal Manchester Children's Hospital has been made a Professor.

On behalf of the MPS Society, Trustees, staff and its members, we send our heartfelt congratulations. No one is more deserving.

Enzyme Unit Open Afternoon at Great Ormond Street

On Tuesday 1st December 2009 the Enzyme Unit at Great Ormond Street held an open afternoon.
The meeting celebrated the Unit's move into the main hopsital pathology block at Great Ormond Street Hospital. The programme included a welcome from Derek Burke, and talks on the past, the present and future direction of the Unit from speakers Liz Young, Prof. Bryan Winchester, Prof. Simon Heales, Kevin Mills and Katie Bainbridge. This is very exciting and we look forward to the future as the Unit's research and development evolves alongside the diagnositic activities.

WHAT'S ON 2010!

MPS CLINICS

Friday 15 January	MPS I BMT under 6 years
Friday 22 January	MPS I BMT over 6 years
Tuesday 2 February	Bristol clinic
Friday 12 February	Birmingham clinic
Friday 23 April	MPS I BMT under 6 years
Friday 23 April	MPS I BMT over 6 years
Friday 14 May	Northern Ireland Clinic
May TBC	Newcastle Clinic
June TBC	MPS I BMT Teenage / Transition clinic
Friday 9 July	Birmingham clinic
Friday 16 July	MPS I BMT under 6 years
Friday 23 July	MPS I BMT over 6 years
Friday 15 October	MPS I BMT under 6 years
Friday 22 October	MPS I BMT over 6 years
Friday 26 November	Birmingham clinic
November TBC	Northern Ireland clinic
December TBC	Cardiff clinic

CONFERENCE EVENTS

12 - 16 April	Sibling Weekend
23 - 25 April	Young Adult Weekend, Blackpool
14 - 16 May	All Ireland MPS Conference
29 - 31 May	MPS Family Weekend, Camelot Lancashire
20-27 June	International MPS Conference, Adelaide
27 - 28 August	MPS III Expert Meeting, Northampton

Date for your diaries!

Expert Meeting on Sanfilippo Disease, MPS III

27 - 28 August 2010

Northampton Hilton

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.

Joanne graduates from University

Here is a photograph of Joanne, who has Morquio, MPS IVA, taken at Glasgow University in June this year just after her Graduation ceremony. Joanne graduated with Joint Honours in Social and Economic History and French after five long years of studying - four years at University and one year spent abroad in France where she worked as a teaching assistant at Lycee Paul Cezanne in Aix en Provence.

I must emphasise that I am writing this from my experience of having to let my precious daughter 'fly the nest' when all my instincts were to keep her safely at home, but she wouldn't have achieved much there! When I last wrote an article for the MPS magazine, we had just returned home from helping Joanne to move out to Aix en Provence - how typical of Joanne that when she left home she also moved abroad! I must admit I found the first few weeks very difficult but she quickly sorted out internet access and emailed me regularly so that, as she said, I would know she was still alive! As the months went by, and she was obviously coping admirably, I was able to cope too and the year passed very quickly; I'm sure she had many adventures and she certainly took every opportunity to travel within France when she wasn't working but that is her story to tell - and there are some things that Mothers just don't need to know! However, Graham, my husband, and I did fly out to visit her in Aix a few times which was always a treat, particularly the trip we made in December when we experienced for the first time a magical continental Christmas market.

When she returned from France, she was adamant that she wasn't going to move back home and quickly found a flat in Glasgow - at least she was living in the same country as us now!

The next two years passed very quickly and the final exams were certainly a stressful time - more so for us, than for Joanne, who coped in her usual pragmatic way.

At last, on 25th June we were all set for her Graduation, however, like all graduates Joanne was feverishly applying for jobs and the previous day had to travel to Bristol for an interview; her Graduation was almost scuppered by a series of unfortunate events which left her stranded at a rail station miles away from Glasgow late at night, having missed her connection through no fault of her own and with no mobile phone signal. So, come the morning of the Graduation we had no idea where she was - situation normal really! An early morning phone call confirmed that she was back at her flat but with a damaged wheelchair, so instead of spending the morning preening ourselves, we were heading to the flat with a toolbag!

However, we made it to the University in time for Joanne to register, collect her robes and join her friends in the Bute Hall for the ceremony which was spectacular; we were fortunate to be seated adjacent to the dais so had a superb view as Joanne received her degree - it was all over so quickly. We then made our way out to the Cloisters for celebratory drinks and to meet up with all the other Graduates and their proud parents where many photographs were being taken and fond farewells bid to both tutors, lecturers and friends. It was a wonderful day.

Joanne continued her search for gainful employment and, on the morning that she completed her 100th job application, she heard that she had secured a job with Cancer Research UK in Oxford. She had two weeks to move out of her Glasgow flat, find another one in Oxford and complete the move which she did with very little help from us. She has now been in the job for three months, has settled happily in Oxford and is relishing the challenges of this next stage of her life.

We remain her very proud parents, quite in awe of all that she has achieved through her own determination and willpower. **Judith Evans**

A Grandfather's Story

The pain was excruciating as my wife Kathie and my eldest daughter Sarah helped me into the car. It was 22 April 1999 and I was just out of hospital following a hernia operation. We were on our way to see Rachel who had just given birth to Ryan, our first Grandchild. Little was I to know that only five weeks later Ryan would also be having an operation for a hernia. This was to be the first of his many hospital visits over the next ten years.

Ryan was a beautiful baby but used to cry a lot from what, we now know were the result of classic Hunter disease symptoms. Over the next two and a half years we were to get used to not knowing why he had so many medical problems. Then, one day, someone in hospital recognised the typical facial look that so many Hunter children have and the diagnosis soon followed. Ryan had MPS II, a particularly rare form which had all of the physical symptoms but also the brain strain as well.

I remember Rachel telling us the news as she sat crying her eyes out whilst cuddling Ryan. What on earth was Hunter's and what could we, as Grandparents do to help. So began the quest for information, this in the days before the wonderful MPS Society was known to us and which today would be the first port of call. Our doctor could not help as he had never heard of it. For me it meant trawling the internet and learning as much as I could myself. What I found out was not encouraging; Ryan could have some remedial surgery to relieve the pain he was obviously in but there was no cure.

What we all learned pretty quickly was that you have to fight to get the best you can for a disabled child. I am immensely proud of the way Rachel (Ryan's Mum and my daughter) has managed to get the best for Ryan by finding her way through loads of bureaucratic minefields. We also had help from the most unexpected quarters, David and Victoria Beckham provided a wonderful state of the art buggy when they heard Ryan did not like the traditional wheel chair. Dreams Come True gave him a tricycle specially adapted so that he thinks he is pedalling and steering – neither of which he can now do.

In 2006 we started a campaign to try and get ERT treatment brought forward from April 1st 2007. We finished up with contact with Tony Blair and various other MP's as well as publicity from ITV, BBC and national and local press. Whilst we were not successful, we learned a lot about how to lobby for the help needed to care for Ryan. During this time we came across some wonderful, helpful people and one or two well meaning but totally incompetent ones; luckily there are far more of the former but you have to work hard to find them. A lesson Rachel says she has learned is not to be afraid to speak your mind, as people are not always right.

Early in 2007 I started Ryan's web site so that friends and family, some of whom were reluctant to keep asking, could find out how he was getting on. It has also attracted emails from some families abroad who have stumbled across it. In March of that year Ryan went to hospital to have his Portacath fitted so that he would be ready for his ERT. There were complications with a bit of the tube breaking off and getting lost. So, the operation finished up taking seven and a half hours and Ryan finished up in Intensive Care. Another worrying period for us all. On 4th April we arrived in Manchester for his first treatment which went reasonably well. In those days he had to sit held tight by Rachel and myself (in relays) for over three hours. This on top of the journey to and from the hospital, which could vary from one and a half hours to double that, each way. This meant one day a week being written off but the rewards soon became apparent with a more talkative and active Ryan. There was also less crying and more smiling which we assumed was because of a reduction in the pain and discomfort he had been in.

On Ryan's birthday my Godson ran in the London marathon with the proceeds going to the MPS Society. We raised some £11,000 which we felt was some small reward for all the help we had received from them. On the day, Clare Stevens, who also has two boys with Hunters came down to support us. She has been a constant support and friend for Rachel and Bob and illustrates the importance of getting to know the parents of other Hunters children.

In June the ERT treatment took place at home in Harrogate and life became much easier, especially when a back pack was used which meant Ryan could move around. This continues to this day with some wonderful nurses from Health Care At Home looking after him. I am normally there to help them and give Rachel and Bob a much needed break.

In 2008 Ryan had to move from his local school where he was very happy. The headmistress bent over backwards to keep him longer than she should have. Thanks to her and his two fantastic carers he could not have stayed as long as he did. He moved to Springwater School for children with special needs where he is also very happy.

In October 2008 Ryan was admitted to Manchester Children's Hospital for a course of antibiotics and physiotherapy to clear a lung infection which had been making him rather ill. There followed several more visits culminating in a Gastrostomy in November; this because he had been aspirating at a far greater rate than at first thought. After managing to pull the first feeding tube out Ryan eventually returned home in time for Christmas. However, he was now on three feeds a day with no more food by mouth; a devastating thing to happen, for food was the highlight of Ryan's day. Now the family had to get used to a new regime with Ryan missing out on family meals. He could not understand why he could no longer eat and to this day I am unable to eat in front of him.

On Ryan's 10th birthday we decided to have a party for all the wonderful carers who had helped us over the years. Ryan was delighted to see so many friendly faces and the day was a great success. He has the ability to make everyone who gets to know him, fall in love with him. Although he doesn't now speak, other than the odd word, one of his smiles makes the dullest day feel full of sunshine.

Helping to look after a Hunter's boy is something Grandparents can do to a greater or lesser degree depending on how close they live. We are able to baby sit so Ryan's parents can have a break from the 24/7 care he needs at home. We can stay at Martin House Children's Hospice when he goes there for respite care; if we are there with him it means his sister, Rebecca, can also attend - something she loves. Giving Rebecca extra attention is also a help as she sees Ryan being the centre of everything in the home. Being available to listen when sometimes life seems to be just too much is appreciated.

People ask how we cope with a child with Hunter's. The answer is that we try to make sure today is a happy one. Tomorrow we start all over again. However, we must also look to the future and right now that means planning to cope with Ryan's increasing lack of mobility. Rachel, Bob and I all have bad backs and Ryan is a heavy boy, so we will have to get funding somehow for a stair lift and hoist for the bath.

Photo this page: Ryan and his family on Boxing Day 2009 with left to right: Dad, Aunt Sarah, Mum, Ryan, Michael and Ryan's sister Rebecca
Photo left: Mum and Dad on Ryan's 9th birthday

Finally, apart from those already mentioned I must thank the following for their amazingly helpful staff: The Willink Clinic Manchester, Harrogate Hospital Woodlands Ward, Harrogate Social Services and the Harrogate Community Nursing Team

This, in a nutshell, is my story. You can read much more on www.ryanseeber.co.uk or just Google Ryan Seeber and his site will be the first entry you come to. How cool is that? Michael Briggs, Ryan's Grandfather

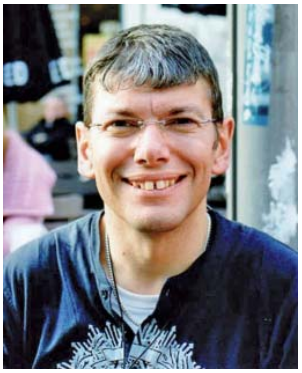
Are you a grandparent whose family is or has been affected by MPS or Fabry?

How did you feel when your grandchild was diagnosed and how has this impacted on your family? Do you have any stories to tell, would you like to share your experiences?

We would like to reach out to more grandparents as you often provide a vital role in looking after the grandchildren, supporting their Mums and Dads as they juggle the demands of life with the extra problems of caring for a child with special needs. We would like to help you share your stories and support you too.

Please get in touch with us at newsletter@mpssociety.co.uk or phone us on 0845 389 9901.

Living with Fabry



Hello everybody, where have the months gone, or do they just go faster when you get older? Unfortunately I don't know the answer to that one.

So how have the last few months been for you? For me it has been very busy, partly because I had a recurrent infection in my left leg three times in three months and the final time I spent seven days in hospital. I was told that I was quite poorly, but because I am so used to having fevers since childhood, I was not aware how serious my condition was. I was able to still smile when the nurses came around.

It was also quite amusing to be the 'special' patient with that strange condition. I had more doctors come to see me than I had good meals (hospital food does not get any better). However, putting that aside, it has been about 4 or 5 years since I stayed in hospital and I noticed there had been a big change in the attitude of the staff. Although they didn't know much about Fabry Disease, they knew more than they did during my previous stays and I was treated with more respect. This really shows how all the work that has been done on improving general knowledge about lysosomal storage diseases is getting better, but there is still more work to do in raising awareness.

I dare say you were all worried to hear and experience a shortage in Enzyme Replacement Therapy. This I found quite worrying as did the rest of my family. I have been moved over to Replagal and had my first infusion recently. It was great to have it done and over so much faster, but I still needed to sleep after it. I would be interested to know how others are getting along with the switch.

Recently I was invited to meet with some doctors for a conference. It was interesting to meet consultants that many of us know and also some that we don't know who work within the area of LSD's behind the scenes and carry out vital research work for and on our behalf. The key message that I got from the meeting was that we will be working together with these doctors and scientists for many years. Communication between 'us' and 'them' needs to improve as we NEED EACH OTHER. The way that we can do this is when we visit our consultants to be open and honest, the British way is to say 'I'm ok' or 'I'm alright.' Well, if you're not, tell the doctor so it can be logged. This information can then be used for your benefit but it builds up the total picture. Think of it as if we are all part of a jigsaw and the doctors are putting it all together. If we don't allow our information to be put into the picture then we will never get a bigger picture. We all have a part to play in this.

I was asked to talk at the conference and it got me thinking about my family after we received our diagnosis, just how it affected us and where we are eight years on. I remember feeling pleased that at long last I had a diagnosis. My immediate feeling was elation and I wanted to know all I could and what I would have to do to get 'cured' but as time went on and I got to know the staff at my specialist centre, I realised that it was not so straight forward.

We, as in my sister, father and mum, found ourselves in two camps. My dad and sister had so much on their plate looking after mum after her stroke that taking on Fabry Disease was too much. As for me, being the kind of person I am, I wanted to know everything and then I would share it with everyone else. I realised however that I was not ready for what I learned, but how was I to know unless I looked and neither was my family. The family ship Hedgecock had really hit the iceberg. It took a long time for me to get myself into a position where I could start to deal with all that had happened.

It also took Diane, my sister, some time especially after the deaths of Uncle Robert and our mum and dad. In fact, it took a few years for both of us and we have both been at each end of the spectrum of both health and emotion. The thought that we could end up like mum and the death she had played on our minds constantly. Everybody said that it might not necessarily be like that. Despite that, I was convinced my life was over.

But I am still here and I will stay here as long as I can and do as much as I can because I have so much to do. As I did not pass any exams in school I have taken on a part time post graduate teaching course. It is a two year course but I am finding it hard to organise myself so any advice would be most welcome!

As one nurse said to me while I was in hospital, you can always manage one smile a day, always do it for yourself and always smile with your heart. So, no matter what happens, smile at yourself every morning. **Ian Hedgecock**

This is Ian Hedgecock's second article in a series of articles focussing on his experiences of living with Fabry. If you have any feedback or would like to share your experiences, please do contact us at newsletter@mpssociety.co.uk

Me and my new hip

Hi! My name is Amy Bray. I am 18 years of age, I was diagnosed with Muclipidosis type lll at the age of 3½. I have had several operations; however my condition has affected my hips the most. Dr Ed Wraith at Royal Manchester Children's Hospital explained to me that my hip joints aren't formed correctly and to make this worse the blood supply to them is poor therefore in effect the bone is dying off.

Back in 2003 it was decided that I needed an operation to build a shelf on my right hip, as my hip was unstable and there was risk of dislocation. This was a success but did not relieve me of my pain. Over the next 6 years I have had less and less mobility and more pain.

Upon my 16th birthday I had to change to the adult clinic at the Northern General Hospital Sheffield and my new consultant was to be Mr Kerry. It was suggested that in the near future I would need a full hip replacement but this was to be the first decision I had to make on my own, as previously my parents had had to make these choices for me. After seeing Mr Kerry for the first time I had to decide how much longer I could cope with the pain, the deterioration of the hip joint and how my life was being affected by less and less mobility.

Like many other sufferers I had seen enough of hospital, therefore as I had just started college I decided I would postpone my operation. During the next 12 months I found I had been taking more pain relief, the pain was affecting my sleep pattern more and I was having to depend on my wheelchair more and crutches around the house which I had never had to do before. At the beginning of this year I decided I needed to take the decision to have the operation. After scans Mr Kerry explained that the hip joint had deteriorated and was much worse than thought. Mr Kerry had the difficult decision as to what type of joint to use and this would have to be specially made because of my small frame due to my condition.

The operation was scheduled for 10th September 2009 but this had to be postponed for two weeks as more time was needed for planning. One of the three surgeons that would be present at the operation took my notes to a conference in America, the procedures for the operation were discussed and delegates agreed that Mr Kerry and his team were going in with the best possible options.

As the day neared I was extremely nervous as this was also my first experience in an adult hospital, therefore my Mum and Dad wouldn't be able to stay with me during the nights which I was used to previously. Mr Kerry had explained that if he couldn't replace the hip, then the worst possible option would be to remove all the damaged bone and leave me without a hip joint.

The day of the operation arrived and I was very worried. I have a needle phobia and I was not looking forward to being a pin cushion for the next 1-2 weeks. When I arrived at the hospital the first thing I had to have done was a blood test. I was upset at this point but in the end the nurse had to bribe me with a bar of Galaxy. I was then prepared for theatre and the next thing I knew I was coming round from the operation.

When Mr Kerry came to see me after he explained to my family that he had managed to carry out a full hip replacement, even though during the operation he had found my hip joint was much worse than anticipated and success was looking doubtful. However, in the end the smallest possible hip joint of 40mm was used, much to his delight.

After a week in hospital and already in much less pain than before, I had managed to walk, climb stairs and get in and out of a car. Therefore I was discharged to recover further at home. Even though progress is slow, just four weeks after my surgery I am already walking further than I could before. **Amy Bray**

MPS CLINICS

Sharing experience of Ametop cream

I would like to share our experience of using Ametop cream. We had been using Ametop cream on our son Tom who has MPS VI for infusions that initially he had through his arm with no problems. Anyway the veins for Tom don't like to play very well. So in January he had his first port fitted under the left side of his chest. However, it wasn't until June that the nurse and myself noticed bruising appearing to the side of his port.

Bruising can occur so at this stage we were not particularly worried. However, slowly the bruising spread then it started to get scabby and crusty.
I took Tom to the doctors. They weren't sure what was wrong but Tom was treated for an infection and given antibiotics. For a short while it appeared to be getting better but it wasn't to be. The out line of his port then became visible and was coming to the top of his chest. A swab was taken and came back clear. In the meantime the port was coming further out and after a few discussions with Manchester Children's Hospital Tom went down there to get it checked over.

It is believed to have been caused through the use of Ametop cream as it was slowly burning the skin and damaging the port. I obviously hadn't known this. The port would have to be removed as it was rejecting itself and a new one replaced. I feel responsible for this, but not realising the damage and strength the cream could actually cause and having used it before, I just carried on using it for his port. If I had known before, I would never have used Ametop.

It's upsetting to think Tom had to go through another operation because of all this. I would like to advise this cream not be used in the community, especially with long term treatments. I would hate anyone else to go through a similar experience to Thomas and ourselves.

My intention in writing this article was not to scare anyone abut having a port. Thomas is doing ok now with his new one but we now only use Emla. I know that some people do use Ametop without problems but just wanted to draw attention to this. Tom was very brave with all that he had to go through and showed such courage. **Kim Coney**

Editor's Note: Please contact your specialist consultant if you have any concerns or questions regarding this article.

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

Northern Ireland Clinic

11 December 2009

On a cold Friday morning I set off for Northern Ireland to support the MPS clinic, held in Antrim Area Hospital.

As always, Dr Fiona Stewart, Dr Joanne Hughes and their reliable team were there to offer clinical support to families. In addition Dr Simon Jones from Manchester flew back to his home roots to provide expert advice from across the water. This was Dr Simon Jones' first clinic in Northern Ireland but he was made to feel very welcome and soon settled in.

The clinic ran very smoothly and most people attended their appointments. Pictured below is Nathan Worsford who has Hunter Disease.

A big thank you goes to all the Doctors and nurses who helped make this clinic a success.
Sophie Thomas

Manchester BMT Clinics

16 October 2009

It was a very busy clinic but everybody managed to turn up. The biggest attraction of the day was the lift! I met with girls: Melissa, Holly, Cody and Demi-Leigh. All of them were chatty and looked very stylish. The boys: Jamie, Mikko and Harvey were testing all the toys and didn't pay much attention to anything else. All children looked great and… I'm sure a few inches taller!

23 October 2009

After a week I arrived in Manchester again. It was another busy clinic and the turn up was excellent. This clinic was dominated by the boys. Bradley, Isaac and Matthew were engaged with recent football events and secondary school experiences. Callum was looking forward to going to the Disney Conference in Florida. Charlie, Steven and Leighton were expressing their views on Spiderman. Rachel and Kiera, neither interested in football nor discussion about super heroes, waited patiently for their appointments.

Both clinics ran to time and went really smoothly. It was great to meet up with children and their parents again. I would like to thank Jean, Ed and the Manchester team for all their support and making us welcome.

Linda Warner

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

Photo top row: Charlie Escanlonilla, Steven O'Reilly, Leighton Barker and Rachel Rothwell.
Second row, left to right: Holly Campbell; Demi-Leigh Rodden; Melissa McKie and Cody Taylor.
Third row, left to right: Harvey Houghton; Mikko Astle; Bradley Evans

Great Ormond Street Hospital MPS III Clinic

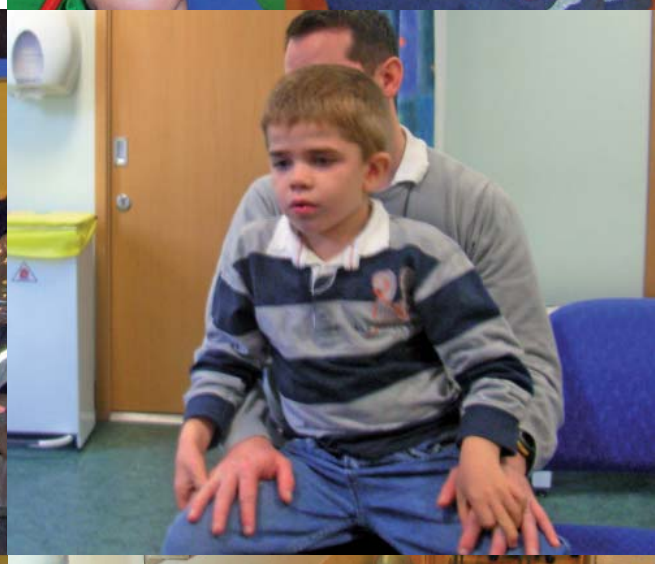
19 November 2009

It was a very windy day when I arrived at Great Ormond Street Hospital, but this weather didn't dampen the high spirits of the children attending this morning's MPS III clinic.

The clinic was full of laughter as Dr Vellodi, Victoria, Sonia and Michelle were kept very busy throughout the morning. Outside in the waiting area, Ana was busy drawing pictures, Daniel was listening to his mum reading a story, Ben and Ollie were pleased to see their friends, Tommy, Ollie and Ashley!

We did have one worrying moment, when Ana's buggy was taken to the 4th floor by accident when the receptionist thought it belonged to Ben and Ollie Illingworth. But it was all resolved and Ana and her buggy were reunited again very quickly!

As the children left the clinic and the toys, books and crayons were put away there was no evidence that anyone had even been there! **Linda Warner**



Photos above: Daniel Zaldua, Ben and Oliver Illingworth, Ana Jabeen and Tommy Thompson

Cardiff Clinic

4 December 2009

It was a cold and wet day when I arrived in Cardiff, but this was soon forgotten as families arrived for their appointments at a busy MPS clinic.

It was lovely to everyone again as it has been a year since I was at the Cardiff Clinic. We all watched as the reception staff were busy getting into the spirit of Christmas by hanging up decorations and dressing the tree, by the end of the afternoon the Children's Outpatient's Dept had been transformed into a Grotto.

For many of our families this was a great opportunity to chat and catch up on what has been going on since they were last together. Despite being a full clinic, we managed to finish on time!

I would like to once again thank Dr Shortland and his team, Dr Wraith and everyone else who made me feel so welcome on the day. **Linda Warner**

Photos clockwise from top right: Marshall Dale (MPS III), Georgia Lewis (MPS III), The Medical Team; Christopher and Steven Jones (both MPS III), Abigail Harvey (ML II)

Scottish Respite Weekend

The MPS Society offered individuals and families living in Scotland a short break at a holiday park on the coast of Ayr. All families who had expressed an interest and were able to attended. The holiday included accommodation, free passes to many of the onsite activities and meals.

The idea of the holiday was to offer families some respite from their daily routine, allowing them time to enjoy a weekend away catching up with old friends and making new friends.

There was plenty to keep the children occupied with the huge arcade, swimming pool, outdoor play area and funfair.

Although the weather was relatively kind to us it was a little on the cold side. For those who were brave enough the park had direct access to the beach which had magnificent views across to the Isle of Arran.

The only down point was that some of the parks amenities and accommodation was run down but despite this, many people said that they had a great time and were thankful for the break away. **Sophie Thomas**

On Friday 11 October 2009 a large group of us travelled to Scotland by Ryanair. The weather was cold and windy but that didn't matter because you were having so much fun you didn't notice! There were many activities such as swimming, bowling, mini golf and much much more. There were loads of lovely food places such as Burger King, a restaurant (which we booked for a buffet which was absolutely delicious), a Chinese stall (which looked very nice as there was always a long queue) and many more! The supermarket sold many things such as food, drinks, toys and clothes (all you'll ever need within a few days)! The rooms were clean and tidy with two bedrooms, a living room, a kitchen and a bathroom/shower room. Overall we think Haven was great and would definately stay there again. **Lauren Page**

Thank you to the MPS Society for organising the weekend in Ayr. It was good to get away for a couple of days. **Andy Robertson**

Childhood Wood Planting Day

On Friday 23rd October 2009, the Society held its annual Tree Planting Day at the Childhood Wood. Seven families attended the day to plant trees and release balloons with their own special messages.

We were extremely lucky with the weather, the sun shone throughout the day and there was warmth in the air and not a rain cloud in sight!

Some families had joined us earlier for a buffet lunch at the Clumber Park Hotel and we were warmly welcomed by Commander Judith Helen Swann, Christine Lavery, Barry Wilson and Wilma Robins.

Other families decided to meet us at the Wood, where we were all welcomed by Paddy Tipping MP.

As we gathered at the Childhood Wood, Councillor Tom Pettengell read the names of the children and adults being remembered today, this was followed by Wilma Robins reading a beautiful poem 'Remember'.

Families and friends were then able to take their time to plant the sapling trees and enjoy the tranquillity of the Wood. Commander Judith Helen Swann, Paddy Tipping MP, Councillor Tom Pettengell and Jacqueline Pettengell all planted trees on behalf of the families who were unable to attend the Childhood Wood Tree Planting Day.

Finally, as the day drew to an end, everyone gathered for the Balloon Release in memory of their loved ones.

Christine Lavery thanked families and dignitaries for making this day such a special and memorable day.

For my part it was a privilege to be allowed to share in this very special day. **Linda Warner**

The costs of the planting day were covered by the Geoff and Fiona Squire Foundation, who have made a generous grant towards the costs of our Childhood Wood programme in 2009/2010.

Photos clockwise from top right: Councillor Tom Pettengell and his wife, Jacqueline Pettengell; The Richardson Family; Paddy Tipping MP and Councillor Tom Pettengell; The Childhood Wood.

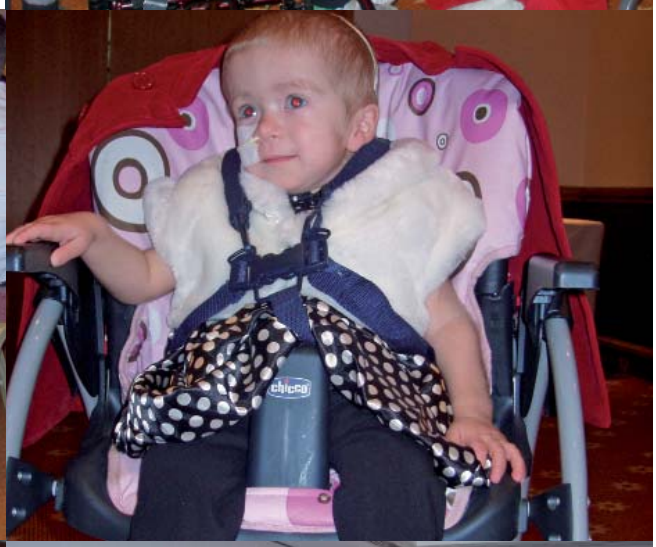
Scottish Christmas Party

On Sunday 29 November 2009 the MPS Society held a Scottish Christmas Party at the Edinburgh Hilton Hotel.

Gina and I flew up from Gatwick at the crack of dawn, looking forward to a nice lunch and fun and games with the children. Six Scottish MPS families were able to join us on the day and we all met at 12 noon with drinks, and then sat down for a lovely Christmas lunch with Turkey and all the trimmings.

After lunch, we were entertained with some balloon modelling and a magic show. The balloon models were brilliant - we had a palm tree with a monkey, an alien and a spider, to name but a few. The magic show kept us all entertained, kids and adults alike and some mums and dads were called upon to be the magician's assistants!

Our last guest of the day was Father Christmas who arrived with the reindeers at Edinburgh airport to give the children an early Christmas gift. We hope you enjoyed the day as much as we did and got an opportunity to meet old friends and chat with new ones. Thank you for coming! **Antonia Anderson**



Disney MPS Conference

We started off early on Sunday 13 December and made our way to Gatwick airport. When we arrived most of the families were already there. Booking in took some time due to us being such a large group. We boarded the plane and had a perfect experience of flying. There were no delays through security and the flight left on time. Our group were fantastic on the plane, the children were occupied by their own TV screens in their seats with a choice of films and cartoons.

On arrival at Orlando International Airport we proceeded through security and found our bus eventually to take us to Disney Coronado Springs Resort accommodation. The families were all booked in, our rooms were clean and we had maid service every day. The American people were all so friendly, in fact by the end of the week it was all too much! You just can't be happy and smile like that all of the time.

All of our families were given theme park tickets to the major theme parks to visit over the coming days. They included The Magic Kingdom, Animal Kingdom, Epcot Centre, Blizzard Beach and Hollywood Studios. The hotel had an outside pool which we had a dip in whilst Christine soaked up the sun in her swimming costume reading a book before the days of work ahead.

The Thursday evening soon came round. We met all the families for the conference registration, then we all sat down and had a buffet meal. We all then attended break-out sessions on various diseases. Christine Lavery ran one of the sessions on Morquio Disease. We met some amazing individuals sharing their stories and giving advice on their personal experiences.

Friday morning soon came round. Breakfast was from 7am followed by an all day conference with childcare. Dr Joseph Munzer gave an overview on Management of all aspects of MPS Diseases with time for questions at the end. Dr Mark Sands gave a presentation on Treatment and Research updates which was then followed by the American MPS Society's AGM.

The afternoon sessions were split into two one hour sessions ranging from Adolescence and MPS, Living with Loss, Reproductive issues and Palliative Care. This was all followed by an Awards banquet including a three course meal and various presentations. Christine Lavery was very surprised to receive a Lifetime of Achievement award.

The following evening the UK MPS Society hosted our own private meal. We travelled to Contemporay Hotel and on the 15th floor we had a meal and watched the amazing fireworks display over the Magic Kingdom.

Sunday came and it was time to travel home. We only had a short delay and then we were on our way. Most of the children slept on the way back ready for the cold and snowy journey back home for Christmas. **Gina Page**

We would like to thank you all for such wonderful trip to the US MPS Conference and Disneyland. We had such a wonderful time, full of memories that will be with us all for a lifetime. Thank you for all your wonderful organising before and during the trip it must have been hard work for you all but it was really appreciated and made the trip so easy for us.

It is difficult to isolate the highlights as there were so many. For Amy, it was all the rides especially Expedition Everest (3 times), Test Track and Soarin' at Ecpot. She also loved Blizzard Beach, where even her first wasp sting could not overshadow a wonderful day.

Sophie loved meeting the characters and all the parades. At one parade she jumped so much in her chair several people came up to afterwards and told us they couldn't stop watching her. She loved "It's a Small World", which we are still singing and the show 'Dream along with Mickey' in front of Cinderella's Castle.

Padraic and I had so many highlights, watching the joy in Amy and Sophie faces, meeting all the families, the US MPS conference and seeing Christine receiving her well deserved Lifetime Achievement award, the wonderful meal at the Disney's Contemporary Resort and watching the incredible Wishes Fireworks display from the balcony.

But if we had to choose one moment for us which summed up the trip it was one morning at 4am when Sophie woke most mornings! Sophie started to pat my face, I kept my eyes closed tight in the hope she move on to her Dad. When she started saying "Mickey says, Mickey says" I couldn't resist and I asked her "What does Mickey say?". She answered "Mickey says I have a dream and it will come true." Well if Mickey says so, who knows.

Thanks to all those who made this trip possible.
Tina, Padraic, Amy and Sophie O'Connor (pictured below)

Christine receives Lifetime Achievement Award

During the Award ceremony at the Gala Dinner, the National MPS Society Life Achievement Award was made to the UK MPS Society's Chief Executive, Christine Lavery, with enormous pride and pleasure. Christine, pictured below with Barbara Wedehase, thanked the Board of Directors, the Executive Director and all the members of the National MPS Society for making this award in recognition of over 28 years working with MPS Families. **Christine Lavery**

We would all like to send our dearest regards and a huge thank you for taking us to Florida. We all had a fantastic and magical time when we visited Disneyworld. This is Callum pictured below, enjoying himself.

The conference was very interesting and we met a lot of new friends and interesting people. **The Pollock Family**

Disney MPS Conference

Thank you ever so much for organising the Disney Conference trip, we had the holiday of a lifetime and a brilliant experience. We all loved it and were thrilled by it all. Hannah and Emily had a superb time on the many rides, shows etc. Hannah particularly enjoyed the Speedway cars, Dumbo ride and carousel at Magic Kingdom, the water rapids, safari and Dinosaur rides at Animal Kingdom - please see attatched photos. We got to meet some great people and catch up with old friends, the girls made lots of new friends too!.

The conference was really helpful - we found the Thursday evening breakout session with fellow MPS1 BMT parents extremely useful, something we would like to do again maybe at a U.K. conference? The opportunity to chat with professionals and other parents at other times during the conference was also really good. We meet up with some American and Canadian families that we had got to know previously through family web sites so that was a unexpected bonus. **Nic Cooper and family**

Khan family with Winnie the Pooh

Clinical Trials Update

MPS I, II and VI

A clinical trial of human growth hormone (HGH)
This clinical trial is being conducted at the University of Minnesota Children's Hospital. Children with MPS I, II and VI with short stature are invited to participate. HGH is a US Food and Drug Administration-approved treatment for short staure, however there is no data at this time on using this treatment specifically in children with MPS. The goal of this clinical trial is to determine what, if any, effect HGH has on growth velocity, bones and cognitive functioning of children with MPS I, II and VI. For additional information contact Linda Polgreen MD, Polgr001@umn.edu

MPS I

Extension Study of Intrathecal Enzyme Replacement Therapy for MPS I
This study is currently recruiting participants.
This is a one-year extension study of the use of laronidase into the spinal fluid to treat spinal cord compression in mucopolysaccharidosis I. Spinal cord compression occurs in this condition due to accumulation of material called glycosaminoglycans (GAG). Laronidase is the manufactured form of the enzyme alpha-l-iduronidase that is deficient in mucopolysaccharidosis I patients. The aim of this study is to determine whether laronidase is safe and effective when given into the spinal fluid as a potential non-surgical treatment. This study is a Non-Randomized, Open Label, Uncontrolled, Single Group Assignment, Safety/Efficacy Study for spinal cord compression due to mucopolysaccharidosis I. If successful, intrathecal delivery could represent a practical, straightforward method of treating central nervous system disease due to lysosomal storage.

Clinical Trial Sites: Los Angeles Biomedical Research Institute at Harbor-UCLA, USA and Helsinki University Central Hospital, Finland

For additional information contact Patricia I Dickson, MD pdickson@ucla.edu

MPS II

A Multi-Center Observational Study Evaluating Anti-Idursulfase Serum Antibody Response in Hunter Syndrome Patients Enrolled in the Hunter Outcome Survey (HOS) Receiving Idursulfase Enzyme Replacement Therapy
This study is currently recruiting participants.
The objective of this study is to evaluate the effect of anti-idursulfase IgG, IgM & IgE antibodies on idursulfase safety (measured by infusion related adverse events) between patients who develop anti-idursulfase antibodies and patients who do not after long-term idursulfase enzyme replacement therapy. This is a multi centre, observational study in a prospective cohort of affected patients.

Primary Outcome Measures:
- To evaluate the relative risk of experiencing an infusion-related adverse event given anti-idursulfase antibody positive status relative to anti-idursulfase antibody negative status. [Time Frame: 2 years] [Designated as safety issue: Yes]

Secondary Outcome Measures:
- To measure the mean (and percent) difference in urinary GAG level between the groups of IgG anti-idursulfase antibody positive and anti-idursulfase IgG antibody negative patients. [Time Frame: 2 years] [Designated as safety issue: No]

Detailed Description:
This study is being conducted to satisfy post-marketing commitments to monitor anti-idursulfase antibody development in Hunter syndrome patients after long-term idursulfase enzyme replacement therapy. The study will be conducted as a sub-study within the Hunter Outcome Survey (HOS). Hunter syndrome patients in the HOS who have previously received idursulfase as well as treatment-naive patients who will begin idursulfase treatment within 30 days of study enrollment will be included.

Research Sites: Children's Hospital & Research Center Oakland, California, USA; Hospital de Clinicas de Porto Alegre, Servico de Genetica Medica, Porta Alegre, Brazil; Birmingham Children's Hospital, UK

MPS II

A Safety and Dose Ranging Study of Idursulfase (Intrathecal) Administration Via an Intrathecal Drug Delivery Device in Paediatric Patients With Hunter Syndrome Who Have Central Nervous System Involvement and Are Receiving Treatment With Elaprase®
This study is currently recruiting participants.
This Phase I/II study is designed to obtain necessary safety and exposure data, as well as secondary and exploratory outcome measures, to be interpreted and used in the design of subsequent clinical trials. This is a Phase I/II Randomized Safety and Ascending Dose Ranging Study of Idursulfase (Intrathecal) Administration Via an Intrathecal Drug Delivery Device (IDDD) in Paediatric Patients With Hunter Syndrome Who Demonstrate Evidence of Central Nervous System Involvement and Who Are Receiving Treatment With Elaprase.

Primary Outcome Measures:

- Safety of intrathecal idursulfase administration as assessed by AEs, changes in clinical laboratory testing, 12-lead electrocardiograms, CSF chemistries, and anti-idursulfase antibodies [Time Frame: 6 months] [Designated as safety issue: Yes]
- Safety, tolerability, and long term patency of the IDDD in the paediatric Hunter syndrome population [Time Frame: 6 months] [Designated as safety issue: Yes]

Secondary Outcome Measures:

- Single and repeated dose pharmacokinetic parameters of idursulfase-IT, given in conjunction with Elaprase, in CSF and blood [Time Frame: Weeks 3 and 23] [Designated as safety issue: No]
- Change from baseline in CSF biomarkers by dose group and in comparison with untreated patients [Time Frame: 6 months] [Designated as safety issue: No]
- Change from baseline in urinary GAG and degradation byproducts by dose group and in comparison with untreated patients [Time Frame: 6 months] [Designated as safety issue: No]

Inclusion Criteria:

A deficiency in iduronate-2-sulfatase enzyme activity of ≤10 % of the lower limit of the normal range as measured in plasma, fibroblasts, or leukocytes (based on normal range of measuring laboratory); AND a normal enzyme activity level of one other sulfatase as measured in plasma, fibroblasts, or leukocytes (based on normal range of the laboratory performing the measurements).
The patient is male and 3 to 8 years-old, inclusive.
The patient has evidence at Screening of early stage (duration and severity metrics per protocol) Hunter syndrome-related CNS involvement, defined as:

- The patient is assessed to be between 2 and 3 standard deviations below the mean overall IQ of the healthy population, OR
- There is evidence of a change of ≥1 but ≤2 standard deviations decline from a previous protocol-defined neurodevelopmental assessment; at enrollment the subject should satisfy protocol-defined CNS parameters. The duration of protocol-defined neurologic involvement is at least 3 months but less than 36 months as documented in the patient's medical history.

The patient has received and tolerated a minimum of 6 months of treatment with weekly intravenous idursulfase, and has received 80% of the total planned infusions within that time frame, including having received 100% of the planned infusions within 4 weeks immediately preceding the surgical insertion of the IDDD.

The patient must have sufficient auditory capacity, with or without aids, to complete the required protocol testing, and be compliant with wearing the aid on scheduled testing days.

The patient, patient's parent(s), or legally authorized guardian(s) must have voluntarily signed an Institutional Review Board / Independent Ethics Committee-approved informed consent form after all relevant aspects of the study have been explained and discussed with the patient. The guardians' consent must be obtained. There are also exclusion criteria to be considered.

Clinical Trial Centre: University of North Carolina at Chapel Hill, USA. One or more non US sites are currently being considered.

MPS IVA

Enzyme Replacement Therapy phase I/II Study

This open label within-patient dose escalation trial in 18 patients at three UK sites is nearing completion and will be followed by a treatment continuation phase. An announcement is expected soon regarding the Phase III/IV clinical trial.

Fabry Disease

Stroke in Young Fabry Patients (sifap1): Frequency of Fabry Disease in Young Stroke Patients

This study is currently recruiting participants.

More than one million people in Europe suffer from a stroke every day. Normally older people have a stroke, but also a significant number of younger people between 18 and 55 years. Usually, these cannot be explained by the classical risk factors such as diabetes, overweight and high blood pressure. New studies indicate that in about 1 - 2 % of the younger stroke patients the cause could have been an undiagnosed genetic disease, the so called Fabry disease. The purpose of this study is to determine in a large number of young stroke patients, how many strokes were caused by Fabry Disease.

The aim of the study is to determine the frequency of Fabry disease in an unselected group of young patients (18 - 55 years of age) with acute cerebrovascular event (CVE).

Rolfs and co-workers have shown a high frequency of Fabry disease in a cohort of patients with cryptogenic stroke (4 % [28/721]) aged between 18 and 55 years. This corresponds to about 1.2 % in the general population of young stroke patients. Therefore the authors stated that Fabry disease must be considered in all cases of unexplained stroke in young patients, especially in cases with the combination of infarction in the vertebrobasilar artery system and proteinuria.

Cryptogenic strokes are cerebrovascular lesions of unknown origin. Clinical and laboratory data show that Fabry disease is itself a risk factor for accelerated atherosclerosis and cardiac and renal disease, which can lead to emboli and hypertension. The pilot-phase started April 2007; the official study started January 2008.

For additional information contact Arndt Rolfs, Prof., MD
arndt.rolfs@med.uni-rostock.de

Fabry disease in females
– learnings from the Fabry Registry

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

Wilcox WR, Oliveira JP, Jopkin RJ et al.
Females with Fabry disease frequently have major organ involvement: lessons from the Fabry Registry. Mol Genet Metab 2008; 93: 112–128.

Fabry disease, is a rare but often severe inherited disorder [1]. As a result of its rarity, it is difficult to conduct large enough clinical studies to provide definitive information about Fabry disease [2,3]. Most doctors have little or no experience in managing people with the disorder, meaning that there is limited knowledge about Fabry disease within the general medical community. As there is still much to learn about Fabry disease, the Fabry Registry, an ongoing global observational registry for monitoring patients with this disease, has been set up (www.fabryregistry.com). One of the objectives of this Registry is to collect data on people with Fabry disease, to characterise the symptoms of the disease and how it progresses over time [4].

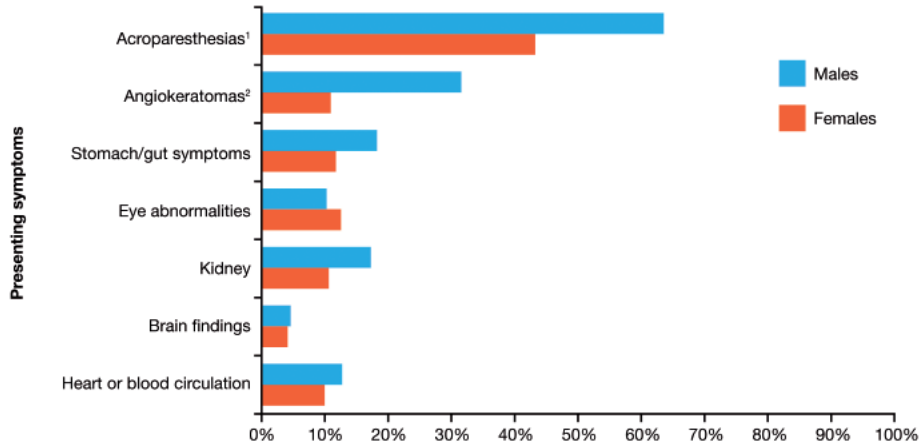
This article is the second of a series for the MPS Magazine about the Fabry Registry, and is based on a recent publication by Dr William Wilcox and colleagues, addressing what happens to women and girls with Fabry disease before treatment with enzyme replacement therapy [5]. Until recently, in contrast to males, females with Fabry disease were thought to be mostly asymptomatic (that is, they had no symptoms or only minor symptoms of this disease throughout a normal lifespan). However, some recent reports have shown that females may, in fact, develop substantial Fabry disease symptoms [6,7]. Here, we will focus on results from the Fabry Registry in large numbers of females prior to treatment, and compare these with data from male participants in the Registry to see how Fabry disease affects women and girls [5].

An effort was made to include all females with Fabry disease in the Registry, regardless of the presence of symptoms.

- Thus, this study includes 1077 women and girls in addition to 1159 males.
- The median age at which symptoms started was 13.0 years in females (9.0 years in males), and diagnosis was far later in females (median of 31.0 years) than in males (median of 24.0 years). (A median value is a kind of average, and just means the middle value.)
- Over two-thirds (69.4%) of females had Fabry disease symptoms.

The most important finding in females is that 20% reported major clinical problems involving their heart, kidneys or brain compared with about 31% of males. In addition, for girls, the most frequently reported symptoms at enrollment were (Figure 1):
- acroparesthesias (tingling, numbness and/or 'pins and needles' sensation) (43.3%)
- eye abnormalities (12.5%)
- gastrointestinal symptoms (11.9%)
- angiokeratomas (small dark red- or purple-coloured raised spots) (11.2%)
- kidney symptoms (10.6%).

Figure 1. Fabry disease symptoms at presentation to the doctor.



[1]Tingling, numbness, and/or 'pins and needles' sensation
[2]Dark red or purple raised spots

However, if follow-up data were combined with data at enrollment, the frequency of angiokeratomas was comparable for both sexes (about 18% in each) and stomach pain and diarrhoea occurred more often in females than in males.

A serious potential complication of Fabry disease among females was an effect on the heart, occurring in 14% of females and 19% of males. The most common heart problems in women were the enlargement of the left pumping chamber of the heart (termed left ventricular hypertrophy or LVH) and abnormal heartbeat rhythms (called arrhythmias). These occurred in 18% and 7% of females respectively. Heart problems tended to happen about 10 years later in life among women than in men.

Strokes occurred in 4.2% of females compared with 6.7% of males, and the first stroke happened, on average, about 5 years later in women than in men. Significant kidney problems were common in women (10.6%). Many women (39%, or 135 out of 346 for whom protein in the urine was measured) had high levels of proteinuria, which indicated that they are at risk of having kidney disease. About 2% of women versus 14% of men underwent kidney dialysis or transplant. However, among women who had any kidney problems, the mean age at which these happened was similar to the age that men reported these problems.

Both sexes had lower quality of life, as measured using a standard questionnaire, compared with scores for the general U.S. population.
• Mean scores for males aged 18–25 years were generally lower than normal, whereas the quality of life only worsened dramatically among females by the age of 25–35 years.
• Both male and female patients reported the lowest quality of life between the ages of 35 and 55 years.
• Notably, women tended to have an even poorer quality of life than men at 55–65 years and when older than 65 years.

The proportion of women in the Registry who were treated with enzyme replacement therapy was much lower (33.8%) than in males (81.9%) with some apparently significant differences in practice; for example, only 53.1% of females with LVH had enzyme replacement therapy compared with 84.9% of males with LVH. It remains to be seen if this may eventually change as new data in women with Fabry disease and severe symptoms emerge [5–7].

In summary, a high proportion of females with Fabry disease show signs and symptoms of this disease, and a substantial proportion have serious problems affecting the heart, brain and kidneys. It is evident that females remain under-diagnosed, incompletely monitored and undertreated. This report should finally dispel the misconception that Fabry disease does not affect women or that it does not affect them severely. As such, the individuals participating in the Fabry Registry are contributing to fundamental changes in the perception, understanding, and management of Fabry disease.

References

1. Meikle PJ, Hopwood JJ, Clague AE et al. Prevalence of lysosomal storage disorders. JAMA 1999;281(3):249–54.
2. Pinto R, Caseiro C, Lemos M et al. Prevalence of lysosomal storage diseases in Portugal. Eur J Hum Genet 2004;12(2):87–92.
Date of preparation: October 2009
Code: FABR-UK-10/09-1696
3. Poorthuis BJ, Wevers RA, Kleijer WJ et al. The frequency of lysosomal storage diseases in The Netherlands. Hum Genet 1999;105(1–2):151–6.
4. Eng CM, Fletcher J, Wilcox WR et al. Fabry disease: baseline medical characteristics of a cohort of 1765 males and females in the Fabry Registry. J Inherit Metab Dis 2007;30(2):184–92.
5. Wilcox WR, Oliveira JP, Hopkin RJ et al. Females with Fabry disease frequently have major organ involvement: lessons from the Fabry Registry. Mol Genet Metab 2008;93(2):112–28.
6. Deegan PB, Baehner AF, Barba Romero MA et al. Natural history of Fabry disease in females in the Fabry Outcome Survey. J Med Genet 2006;43(4):347-52.
7. Wang RY, Lelis A, Mirocha J et al. Heterozygous Fabry women are not just carriers, but have a significant burden of disease and impaired quality of life. Genet Med 2007;9(1):34-45.
Declaration of interest: The author is employed by Genzyme Therapeutics, Oxford, UK.

NIHR Medicines for Children Research Network

Clinical Inherited Metabolic Disorders Clinical Studies Group

Chris Hendriksz – Chairman of the Clinical Studies Group for Inherited Metabolic Disorders, Birmingham Children's Hospital
Tanya Collin-Histed - Coordinator UK LSD Patient Collaborative Group, Dursley, Gloucestershire
Laura Pilkington - Clinical Studies Group Administrator, MCRN Coordinating Centre, Liverpool
Andrew Rose - Industry Liaison Manager, MCRN Coordinating Centre, Liverpool

Challenges of research in rare Inherited Metabolic Disorders

Research studies in populations affected by rare Inherited Metabolic Disorders (IMD) is poorly funded and regarded scientifically insignificant due to small numbers involved in these studies. To get reasonable numbers it is important to collaborate on studies and involve as large a population as possible. One possible solution is to establish national research groups that could influence the national research agenda. Potential researchers and commercial companies could then approach a single group for advice and will also know that maximum participation is possible even over wide geographical areas.

Involving patients and their families

Patient/parent participation is crucial and with all this in mind a Clinical Studies Group (CSG) was formed with the help of the UK Lysosomal Storage Disorders (LSD) Patient Collaborative Group and the National Institute for Health Research (NIHR) Medicines for Children Research Network (MCRN). The UK LSD Patient Collaborative group had a similar vision that collective action is likely to be more beneficial for patients and for that reason they were approached to help establish this CSG. The group is made up representatives from the Gauchers Association, The Society for Mucopolysaccharide Diseases (the MPS Society), Battens Disease Family Association, Niemann-Pick Group (UK) and the Pompe Association. The group first met in January 2007 and agreed to operate as a forum to discuss issues common to all groups including working with the Pharmaceutical Industry, the development of homecare services for patients, new born screening, the development of metabolic networks in the UK, the need for research into the brain and representation on the Health Technology Assessment longitudinal study into enzyme replacement therapy for LSD's.

What is a Clinical Studies Group?

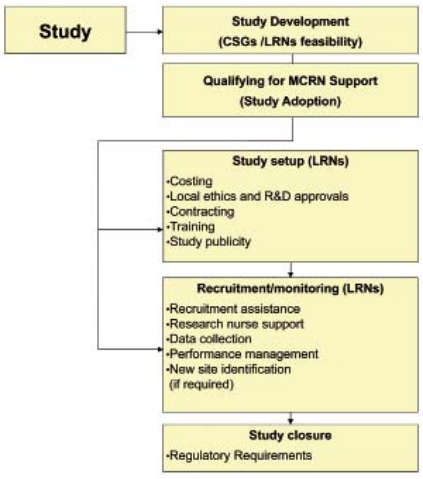
The NIHR MCRN has been created to improve the coordination, speed and quality of randomised controlled trials and other well designed studies of medicines for children and adolescents including those for prevention, diagnosis and treatment. MCRN CSGs are the drivers of the network and are the primary route by which clinical studies are considered in the development of the MCRN portfolio.

A CSG for IMD has now been formed and consists of the following members: 4 Paediatric Metabolic Consultants, 1 Adult Metabolic consultant, 1 Clinical Psychologist, 1 Physiotherapist, 1 Pharmacist, 1 Metabolic Consultant from the devolved nations (Scotland, Ireland and Wales) and a representative from the UK LSD Patient Collaborative Group. The member for the UK Collaborative Group is a Clinical Research Nurse. She will be liaising with the wider group and helping to set the research agenda from a patient perspective. The group is also supported by the NIHR and the MCRN with administrative support and an additional research fellow in Pharmacology. Current members are from London, Birmingham, Manchester, Liverpool and Glasgow, and will be representative of the community caring for those affected by IMD.

Remit of the CSG

Reactive Role	Proactive Role
To provide advice to investigators for the development of research proposals and funding applications	To identify gaps in current research within specialty areas and determine research priorities
To provide advice to commercial companies through the MCRN Industry Liaison Team	To develop clinical trials or other well designed studies to address research gaps
To advise the MCRN Study Adoption Committee regarding the quality of studies submitted to run through the network	To participate in strategic decisions of the MCRN through representation on the MCRN Board

What is the process and assistance offered for potential studies?



Future vision

It is our hope that similar networks will be formed across the world and that international collaboration will become the norm rather than the exception for those affected by rare Inherited Metabolic Disorders.

Contact details

CSG Chair: Chris Hendriksz chris.hendriksz@bch.nhs.uk **UK LSD Patient Collaborative Group: Tanya Collin-Histed** tanya@gaucher.org.uk
CSG Administrator: Laura Pilkington laura.pilkington@liverpool.ac.uk **Industry Liaison Manager: Andrew Rose** Andrew.rose@liverpool.ac.uk

The NIHR Medicines for Children Research Network is part of the National Institute for Health Research Clinical Research, and forms part of the UK Clinical Research Network. The Networks Support and deliver high quality clinical research studies.

2009 South East Asia Joint Symposia on Rare Diseases in Taiwan

This meeting, held on 9-11 October 2009 in Taipei, Taiwan, was organised and opened by Virginia Tsai, Founder and President of the Taiwan MPS Society and mother of David who lost his life to MPS II.

At the opening Gala Dinner, Guest of Honour Dr. Roscoe O. Brady from the US National Institute of Health gave his view of the insights and perspectives on progress made in the LSDs over the last 20 years. Dr Brady was born in Philadelphia in 1923, attended Pennsylvania State University, and received his medical degree from Harvard Medical School in 1947. Dr. Brady described his accomplishments that include the identification of the metabolic defects in hereditary lipid storage disorders including Gaucher disease, Niemann-Pick disease, Fabry disease, and Tay-Sachs disease. Dr Brady has applied this knowledge in developing diagnostic tests, carrier identification procedures, and the prenatal detection of lysosomal storage diseases. His recent research interests include the ongoing development of enzyme replacement therapy for metabolic storage disorders other than Gaucher disease, as well as gene therapy for these conditions.

The aim of the meeting brought together MPS families from Taiwan, Hong Kong, Malaysia, South Korea and the Philippines. International speakers from outside the region included: Christine Lavery, Overview of the MPS Society in the United Kingdom; Kirsten Harkins & Barbara Wedehase, Experience sharing by patient groups in Europe and North America and; Wendy Boon, Experience sharing by patient groups from The South Asia Pacific.

These along with specialist talks on ophthalmology, diagnostics, Ronald McDonald House and Quality of Life issues as well as a patient testimonial by William Luo, MPS II, made for a very interesting programme.

One of the highlights of the visit to Taiwan was having the privilege of seeing the children's choir of the Taiwan Foundation for Rare Disorders perform. It was mesmerising watching these amazing children with different rare disorders sing their hearts out and in tune! We loved it and the children themselves clearly loved performing.

I am sure Barbara, Wendy and Kirsten join me in thanking Virginia, her daughter Julie, son Edward and all her family and MPS supporters for making our time in Taiwan so memorable.

Photo top right: Virigina Tsai, Wendy Boon, Christine Lavery and Kirsten Harkins

Prof. John Hopwood is awarded 2009 CSL Florey Medal

9 November 2009
Professor John Hopwood, South Australian Scientist of the Year 2008, will this evening follow in the footsteps of some of Australia's greatest medical researchers, including immediate past winner Professor Ian Frazer and Nobel Laureates Professors Barry Marshall and Robin Warren, by being awarded the 2009 CSL Florey Medal.

Professor Hopwood today joins a cohort of extraordinary Australian medical researchers who are at the forefront of breakthroughs globally, and who have dedicated their lives to basic research all the way through to making a difference to our lives through prevention and treatment.

This prestigious honour is part of the Australian Institute of Policy and Science's Tall Poppy Campaign and takes its name from Australia's first Nobel Laureate in Medicine, Sir Howard Florey, who made an indelible impact on world health by developing penicillin. The winner receives a $25,000 prize and a hand sculpted silver medal, joining the most recent winner, Professor Ian Frazer in 2007.

Hopwood first formed the Lysosomal Diseases Research Unit in 1976 in South Australia. This Unit is the only group researching lysosomal storage disorders nationally and is the largest multidisciplinary group working on them world-wide. The Unit is world-renowned for its research capabilities and the translation of research findings into state-of-the art diagnostic services.

Lysosomal diseases are genetically inherited and affect about 1 in 5,000 babies, with recent findings suggesting maybe up to about 1 in 1,000. Symptoms are progressive and impact on many body organs including the skeleton, heart, lungs and brain with devastating effects.

Hopwood's Unit has generated several world firsts, particularly the isolation of genes involved in some of these disorders and the development of first ever FDA-approved treatments for two disorders which were marketed world-wide in 2005 and 2006. These outcomes meant improved quality of life for patients and multi-million dollar royalty returns to South Australia, representing the largest public sector commercialisation outcome for that state, and possibly Australia.

Ongoing research commercialisation is well advanced to enable screening and the development of effective therapies for lysosomal disorders affecting the brain.

Hopwood says: "we are just tipping the iceberg." He is most of all excited about the discoveries that link lysosomal disease to unexplained cancers, stroke and neurological disorders that concern us all, as well as his team's achievements in helping the many children affected.

Professor Peter Rathjen, AIPS Director, Deputy Vice-Chancellor (Research) at the University of Melbourne and Chair of the 2009 CSL Florey Medal Selection Committee says the bi-annual accolade was created in 1998, the centenary of Sir Howard Florey's birth, to celebrate world standing achievements in biomedical science and human health advancement by Australian researchers.

"Many top ranking scientists were in the running for this year's medal, a testament to the excellence in Australian medical research. Professor Hopwood was the winner, reflecting the quality of his long term dedication from basic research through to clinical outcomes," he says.

"John Hopwood is an Australian medical researcher whom we should celebrate, and who should stand high on the Australian and world stage for his achievements, alongside other great Australian medical researchers."

John Hopwood comments: "It is important that biomedical scientists receive recognition for the perseverance that is required for real outcomes through research. For this I am thankful for this award. It is important that we also recognise all the clinicians and scientists involved. But neither they nor I would be where they are without the patients and families, nor would our work have any meaning without their need."

CSL, Australia's leading biopharmaceutical company, became the principal sponsor of the Florey Medal this year as part of its commitment to recognising and promoting scientific excellence in Australia.

"Australia has much to celebrate when it comes to achievements in medical research, and we need role models who can inspire a new generation of innovative scientists. Professor John Hopwood is one such person and an extremely deserving winner of the 2009 CSL Florey Medal" Dr Andrew Cuthbertson, Chief Scientific Officer at CSL said today.

Citation for Professor John Hopwood

Professor Hopwood is honoured with the 2009 CSL Florey Medal for his life-long work into the diagnosis and treatment of genetically inherited disorders that affect children with devastating clinical effects leading to progressive destruction of the brain and other organs.

Born in Melbourne, and educated at the Swinburne Institute and then Monash University, Hopwood first formed the Lysosomal Diseases Research Unit in 1976.

From basic science to clinical trials, through to clinical outcomes and commercialisation, Professor Hopwood has dedicated three decades of research, discovery and personal perseverance in the service of human health advancement for the sufferers of lysosomal storage disorders (LSD) which affect at least one in 5,000 babies if not one in 1,000, as recent research suggests. Recently, lysosomes have become implicated in otherwise unexplained stroke and heart disease, cancers and neurodegenerative disorders.

Professor Hopwood today heads a group of more than 30 researchers through SA Pathology at the Adelaide's Women's and Children's Hospital, focusing on the diagnosis and treatment of LSD, and was awarded the South Australian of the Year (Science) Award in 2008 amongst other accolades. Hopwood and his team have developed a novel program to enable newborn screening for these disorders. His unit has achieved world-first treatments for two lysosomal storage diseases that have dramatically improved clinical outcomes for patients worldwide, numerous patents, as well as FDA-approved drugs and commercial licenses and attracted over $30 million in competitive research funding, to date.

John Hopwood's research is world leading. He has displayed continuous and humble dedication and effectiveness in relating to LSD patients and their families alongside his research team, clinicians, global colleagues and pharmaceutical companies. John is a man of vision who, through basic research, persistence and commitment, has accumulated and harnessed scientific knowledge and techniques to achieve his stated goal of "early diagnosis and effective therapy of lysosomal storage disorders". Without his efforts, thousands worldwide would be without diagnosis, therapy or the hope that either would ever come.

2009 CSL FLOREY MEDAL SELECTION COMMITTEE
Professor Peter Rathjen - Chair
Deputy Vice-Chancellor (Research) – and AIPS Director
The University of Melbourne

Professor Warwick Anderson AM
Chief Executive Officer NHMRC

Professor Suzanne Cory
Director Walter Eliza Hall Institute

Professor Geoffrey Donnan
Director Florey Institute

Steven Dower
Head of Protein Biochemistry
Honorary Professor, Bio21 Institute

Professor Ian Frazer
Director Diamantina Institute for Cancer, Immunology and Metabolic Medicine, QIMR

Professor Peter Schofield
Executive Director and CEO Prince of Wales Medical Research Institute

Professor Fiona Wood
Winthrop Professor Burn Injury Research Unit
The University of Western Australia

7th International Fabry Meeting

My name is Leslie Hilliard and I went to the the 7th International Fabry Meeting in Vienna in November.

The results that were shared at the meeting, particularly those by Dr. Derralynn Hughes from the Royal Free Hospital in London on Chaperone Therapy AMIGAL 1001 which is still in the trial stages, looks very promising.

There was also a very interesting talk by Dr. S Keshav on 'Nutrition and Digestion' which was very informative.

It was also very good to see some old friends from all over Europe and make new ones too. I would like to thank the sponsors for their support and to say how important these meetings are, to all of the Fabry patients, and how we all look forward to the next meeting. **Leslie Hilliard**

Photo right: Les Hilliard and Latif Jekhakha

International MPS Network Meeting

The organisers and representatives of ten MPS Societies around the world met on 19 December 2009 in Orlando, Florida following the National MPS Society Disney Conference. The minutes of the International MPS Network Meeting held in Northampton, UK in August 2008 were approved.

A Memorandum of Understanding was approved as the modus operandi of the International MPS Network. A review of current membership was undertaken and it was agreed that those not currently in the MPS Network should be invited to apply in preparation for approval at the next International Network Meeting 22 – 23 June to be held in Adelaide, Australia prior to the International Symposium on Mucopolysaccharide and related diseases.

David Oliver, President of the Australian MPS Society presented and received feedback on the draft 'Family Programme' for the 11th International Symposium on Mucopolysaccharide and related diseases. The theme of the Symposium is 'Translating Research into Clinical Reality' and runs from 23 – 27 July 2010.

Thomas Baum from the German MPS Society presented a proposal of what constitutes a paediatric or adult medicine specialist diagnostic, clinical and treatment centre for Lysosomal Storage Diseases. This included designated specialisms in anaesthesia, haematology, genetics, ENT, cardiology, neurology, ophthalmology, respiratory, urology, dermatology, psychology and orthopaedics. This intiative which very much replicates the UK National Commissioning model, is to welcome representatives of smaller populated countries or those with small populations and a large land mass with the formula of a specialist centre having at least '50 patient years' experience.

Barbara Wedehase opened up for discussion the '2014 International MPS Symposia Bid Guidelines. A timetable for receiving the bids was agreed along with criteria for bid presentations. It was agreed that up to two representatives each from Genzyme, Biomarin and Shire will be invited to hear the bids and each company will have one vote. The bids will be heard on the morning of 23 June at the International MPS Network Meeting in Adelaide, Australia. The result of the bid will be announced at the 11th International Symposium on Mucopolysaccharide and related diseases.

Dr Mark Sands, from the Sands' Laboratory Washington University School of Medicine, gave an authoratative and thought-provoking presentation on the importance of Peer Review as a prerequisite to patient organisations funding research. Dr Sands started by describing Peer Review as the 'Evaluation of a person's proposed work and performance by a group of people in the same occupation, profession or industry'. He went onto state that critical evaluation of proposed work is good for the researcher, good for the patient organisation and good for the entire system. A model of peer review was discussed and many questions ensued.

Dr Myra Roche, University of North Carolina at Chapel Hill shared some of her experiences researching 'How Parents Search, Interpret, and Evaluate Genetic Information Obtained from the Internet'. Although all of her research was with parents of children who do not have an MPS disease her finding were translational and highlighted the prevalence of Internet use and how information is used.

Finally Dr Ann Barbier and Dr Charles Richards, Principal Medical Director, Shire Human Genetic Therapies updated the Network on two areas of interest. Firstly the MPS II Intrathecal ERT clinical Trial. This is currently recruiting at the University of North Carolina at Chapel Hill where the principal investigator is Dr Jo Muenzer. Details of the clinical trial itself can be found in the Clinical trial section of this magazine. Currently Shire Human Genetic Therapies are considering one or more additional clinical trial sites. Secondly, Dr Richards spoke of the MPS IIIA Natural History Study and potential MPS IIIA Intrathecal ERT Clinical Trial.

First MPS patients day in Russian history

Following my visit to Moscow in June 2009 to speak to LSD Patient Associations supporting Gaucher, MPS diseases, Niemann Pick and Pompe disease, a further meeting was held in December 2009 to look at patient access to Enzyme Replacement Therapies.

To facilitate discussions the UK MPS Society supported the Russian patients' associations 'First MPS patients day in Russian history' by providing materials on presentations it used to campaign for funded ERT back in 2004. There were more then 70 participants in this meeting which enabled all patients and their carers to discuss urgent and important questions relating to clinical management and access to ERT. At the end of this meeting all patients and their families made a resolution to approach the Russian President with a request to the government for a reimbursement programme for Enzyme Replacement Therapy.

We wish all Russian patients, their families and health professionals success in achieving their goals.

Living with Morquio

My name is Josefine and I live in Norway. I am twelve years old and have Morquio syndrome. I am in the last grade of Primary School and I have four siblings. My leisure time activities are being with friends, listening to music, sports, computer games, going to the cinema, different types of handiwork and cheerleading. If you have Morquio syndrome and are about the same age as me, I would like to hear from you. We could perhaps get to know each other and share experiences.

When I was little and went to kindergarten I stumbled and fell a lot. My caregiver talked to my parents and they took me to a doctor at the hospital. It took about two years before I was diagnosed with Morquio because the Norwegian doctors were not quite sure what was wrong with me. When I met Dr. Ed Wraith from Manchester at Frambu when I was five years old he confirmed that I have Morquio. I do not have the most severe form of the disease, but I have already had surgery on my hips. Sometimes I have pain in my wrists and in my legs, especially if I have been very physically active during the day.

The bad things about having Morquio are that I can not jump on our trampoline and do gymnastics as my siblings do. One of my sisters is on the national team for cheerleading and she is going to compete in Florida, US next year. I wish I could too. She is very lucky, but I know it is a lot of hard work for her too. What else bothers me is that some people treat me as if I am six or seven years old because I have short stature. I also often get a bit angry and sad because I move much more slowly than my friends.

I really like to go downhill skiing with friends and family. I have sit skis and I am quite good at it. Last winter my father and I almost had an accident when we flew three metres up in the air before landing with a big bang in the ski hill. We use helmets. Some other sport activities I like are ice pigging and swimming.

I enjoy school and have a lot of good friends. The subjects I like the best are mathematics, gymnastics, cooking and health, handiwork and English. The name of my gymnastics teacher is Harriet. She is a very good teacher. She has borrowed five wheelchairs so all the pupils in my class can learn how to use them. When we have races I always win. Another person who means a lot to me is Bodil. She is my physiotherapist. She has known me since I was little and she supports me when I need it.

When I get older I hope to go to a MPS Conference abroad. Norway is a small country and there are no girls of my age with Morquio syndrome here. I therefore hope to get to know somebody from Great Britain. My mother would also like to get in touch with some mothers who have children with Morquio at the same age as me.

Josefine
E-mail: jossi_rosa@hotmail.com

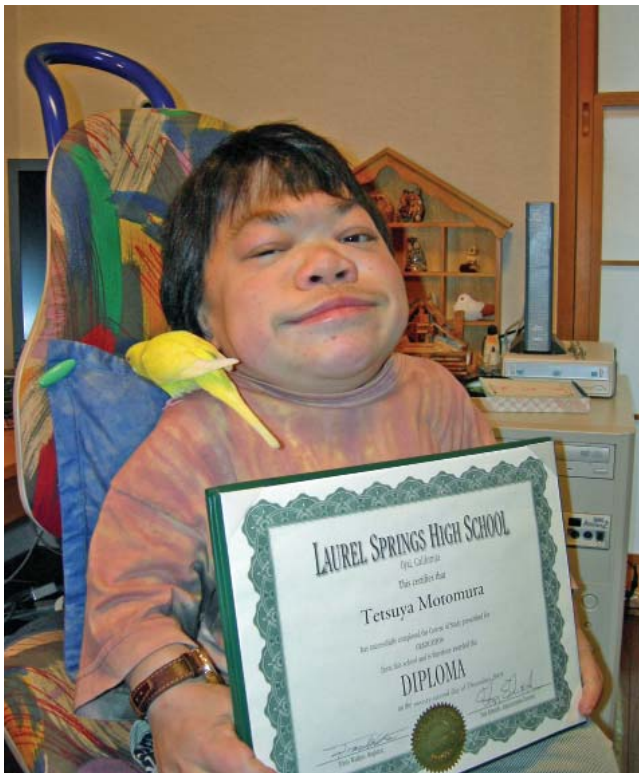
Signe (my mother)
E-mail: signeh@live.no

Destiny in the Wind

This was a dance performance which took place in September 2009 in New York, inspired by the life of Tetsuya Motomura (pictured below) who had Mucolipidosis.

Kazuma, the creator of 'Destiny in the Wind' says that 'Before my little brother Tetsuya passed away it was his wish that I tell people of his existence and struggle. We discussed creating a dance piece based on the imagery he had whilst listening to his favourite piece of music, 'Chikyu ni Kanpai' by Takefumi Haketa. His imagery was so vivid, telling and self-reflective and matching the music so beautifully. I tried to do justice to his creativity by developing this piece in accordance with his imagery, but unfortunately, I was never able to show it to him in his lifetime. I have since choreographed the piece and performed it in two cities in Japan. Now I am ready to bring it back to New York developed with my dance partner, Kana Ote, along with paintings by my mother, Sally Marshall-Motomura.'

Tetsuya Richard Motomura
There was a man less than 3 feet tall that lived to the age of 23. With great difficulty and pain, negativity never touched him as he lived with all his might. He attended Thomas Norman High School in New York and later graduated from an online course. Even with his rare condition of Mucolipidosis, he touched the hearts of many with his humour, power to imagine and passion for life.



Challenging Behaviour Foundation

'Breaking News', the Winter issue of 'Challenge' (newsletter of the Challenging Behaviour Foundation) focusing on short breaks for families caring for individuals with severe learning disabilities described as having challenging behaviour is now available.

In 2006, Mencap's 'Breaking Point' survey highlighted the fact that 6 out of 10 families were not receiving short breaks that met their needs. Three years on, little has changed. 'Breaking News' includes a special report from the Challenging Behaviour Foundation and the Tizard Centre (University of Kent) highlighting findings from a survey of over 300 families across the UK.

Over a quarter of families reported their family member had been excluded from a short break service. "Families are expected to cope without support and on a long-term basis with a situation which qualified professionals say they are unable to manage, is too difficult for them or beyond their expertise."

The report includes accounts from three families, which highlight the need for inclusive short breaks services, alongside a summary of what families feel would improve the quality of short breaks and suggested actions. A positive family experience ('Getting it right') and report from an Aiming High for Disabled Children 'Pathfinder' demonstrate positive family experiences of short break services.

Other articles include:
'Comment' by Tony Osgood, Lecturer in Intellectual & Developmental Disability, Tizard Centre: "A break isn't about having a holiday or 'a well earned rest' but often about merely surviving."
'Specialist equipment and safety adaptations': new products described include magnetic shower and towel rails, curtains with Velcro tab tops and "virtually indestructible" mattresses.
News of a new national initiative led by the Challenging Behaviour Foundation: the Challenging Behaviour National Strategy Group

'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

If you would like any further information then please do not hesitate to contact us.

The Challenging Behaviour Foundation
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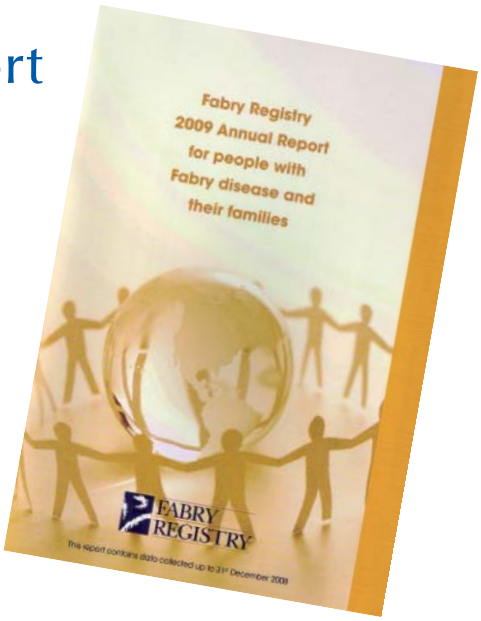
Fabry Registry 2009 Annual Report



The Fabry Registry 2009 Annual Report is available now at no charge from the MPS Society. If you would like a copy, please contact us at newsletter@mpssociety.co.uk.

The booklet tries to answer the questions that are frequently asked about the Fabry Registry by people with Fabry disease and their families. It covers information that has been covered about people living with the disease and the different medical problems they encounter.

The booklet provides 'collective' information about numerous participants worldwide, and gives an overview of their medical problems, which may sometimes be serious and life-threatening. However, Fabry disease is 'individual' and each affected person has their own clinical paths.

For Sale

Alvema 400 classic standard from Rainbow Rehab. It has a low back and the seat is 15" wide. **£100**
For more information please contact the MPS Office on 0845 389 9901.

MPS website - Kids Section!

We are developing the MPS website and have recently added a new section for Kids. We would like to use this section to expand on our support to siblings and children affected by MPS and related diseases.

Do you have a brother or sister affected by MPS? Would you like to tell us about your family and share your stories. Please email your stories to **newsletter@mpssociety.co.uk**.

Can you help with our media campaign?

Do you ever wish that more people knew what you meant when you started talking about MPS or Fabry?

One way to help change this is to spread awareness through the media - newspapers, magazines, TV and radio for example. We are looking to promote MPS Awareness Day through the media and need your help. Would you be willing to provide a case study, a story about your or your family's experience of living with MPS or Fabry? Perhaps you have an unusual story about receiving the diagnosis, have achieved something amazing with MPS or Fabry, are thinking of doing some fundraising, or simply want to tell your story.

To find out more please email us at **fundraising@mpssociety.co.uk**

RAISING AWARENESS

MPS Awareness Day

15 May 2010

One baby every eight days in the UK will be born with an MPS or related disease

Each year the Society celebrates International MPS Awareness Day on 15 May. This is a day devoted to raising awareness of MPS and Related Diseases

Help us celebrate International MPS Awareness Day on Saturday 15 May 2010

This year we're asking all our members, Friends and supporters to do something, big or small, to mark MPS Awareness Day

Download our Awareness Day flyer from **www.mpssociety.co.uk** or give us a call on **0845 389 9901** to find out more.

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**

Changing the world for children with genetic disorders

Friday 1st October 2010

Jeans for Genes is the national charity that holds Jeans for Genes Day. Jeans for Genes aim to provide funding for the care and support of children and their families affected by genetic disorders as well as funding research into the genes responsible and the development of effective treatments and cures.

Jeans for Genes Day takes place on the first Friday in October. Next year's event takes place on Friday 1 October 2010. Wearing 'jeans' is a great reminder of the 'genes' that you're raising money for.

For more information on how you can help Jeans for Genes, support the campaign all year round and education and awareness opportunities visit **www.jeansforgenes.com**.

The MPS office celebrating Jeans for Genes Day

Dear All

We hope you are all well in the MPS Office. You all work so hard to keep the Society running efficiently and smoothly, we would like to thank you for all the effort and hard work that you put in.

This year we did our usual thing for Jeans for Genes in our place of work, N.C. & B. Lunt Pharmacy. We all wore our jeans and also had some Fifi badges for sale on the counter. Our colleague Becky, pictured below, did her usual dressing up bit as a rock chick! We made a grand total of £134 from donations and the sale of badges. **Karen and Andrew Weedall**

If you celebrate Jeans for Genes Day, please let us know how it goes!

WANTED!

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

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FREE!

Minimum sponsorship required

Help us raise funds by making an exhilarating 10,000 feet freefall parachute jump.
No experience is required and if you raise the minimum amount of sponsorship
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or email **fundraising@mpssociety.co.uk**