CARE TODAY, HOPE TOMORROW

The **Society for Mucopolysaccharide Diseases** the MPS Society) is a voluntary support group, founded in 1982, which represents from throughout the UK over 1200 children and adults suffering from MPS and Related Diseases, their families, carers and professionals. It is a registered charity entirely supported by voluntary donations and fundraising and is managed by the members themselves. Our aims are to:

To act as a **support network**

To bring about more **public awareness**

To promote and support **research**

What we offer

Advocacy Support

Telephone Helpline

MPS Befriending Network

Support to Individuals with MPS

Regional MPS Clinics

Information Days and Workshops

National & International Conferences

Sibling Workshops

Information Resources

Quarterly Magazine

Bereavement Support

Research & Treatment

'Mucopolysaccharide and Related Diseases are individually rare; cumulatively affecting 1:25,000 live births. One baby born every eight days in the UK will be diagnosed with an MPS or Related Disease. These multi-organ storage diseases cause progressive physical disability and, in many cases, severe degenerative mental deterioration resulting in death in childhood.'

Cover photograph:
Myles Broughton and Sarah Long (MPS Members) with Jeremy Hunt MP at the Society's 25th anniversary reception at the Palace of Westminster

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Magazine Deadlines

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

Become a Friend of MPS

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

CONTENTS

CHIEF EXECUTIVE'S REPORT

Summer 2008

It is over four years since the last big step in the development of a new treatment for an MPS disease. Casting our minds back to that time there was real optimism that enzyme replacement therapy for MPS IVA, Morquio disease was on the horizon. There have been a number of deadlines for starting the natural history study, all been and gone. Nevertheless behind the scenes scientists have been working dedicatedly to make the progress needed to bring ERT for MPS IVA to clinical trial. This dedication at last looks as if a clinical trial is in reach.

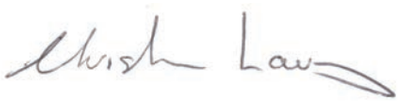
Dr Tomatsu from the University of Louis in the USA has dedicated much of his professional life to finding a treatment for Morquio disease. Having made significant progress, a little known Biotech company became involved. Subsequently, Vivendy took over but there has not been any clinical trial programme launched, as yet.

On 5 June 2008 as can be seen later in the magazine, BioMarin, who developed ERT for MPS I and MPS VI, announced a clinical programme for MPS IVA starting in the first quarter of 2009. This will no doubt come as welcome news to those affected by Morquio disease and their families.

The Morquio conference on 29-30 August 2008 is taking place at the Hilton Hotel, Northampton. This conference was planned in the Summer of 2007 when it was recognised that we could not give adequate time to the many clinical management issues that need to be addressed. As it happens, the time couldn't be happier and I am pleased to announce that both Dr Tomatsu and Dr Emil Kakkis for BioMarin will present at this first ever conference specifically to address the clinical management and treatment options for Morquio disease.

There are still too many members waiting for similar news. Albeit slow due to the sheer challenge of addressing the CNS involvement of Sanfilippo disease, we do hope to have news in the not too distant future. In the meantime, the MPS Society has agreed to invite families of children with MPS III, Sanfilippo Disease born in 1990 or more recently, to participate in a natural history study being co-ordinated by Dr Jean Michel Heard from Paris, France. We hope to get this underway very soon.

Finally, the International Symposium on Mucopolysaccharide Diseases took place in Vancouver, 26-29 June 2008. In the next issue we shall bring to you all the breaking news gleaned as well as some of the presentations.



HIGHLIGHTS from the MANAGEMENT COMMITTEE

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Management Committee Meeting held on 2 - 3 May 2008.

Governance

The Chief Executive confirmed to Trustees that the Charity Commission return on behalf of the Board of Trustees had been completed and a draft sent to the Chairman for his consideration. The Trustees agreed the content of the Charity Commission return and it was duly signed by the Chairman.

Personnel

Trustees were advised that all the staff appraisals have now been carried out and have been countersigned. Two staff are due to undertake appraisal training in readiness for the implementation of a new performance related appraisal system to be introduced in late 2008.

Applications for funding for a MPS helpline post and maternity cover for the Volunteer and Events Co-ordinator are also in the pipeline.

Policies

The Trustees reviewed and agreed unanimously without amendment the following policies: Conduct of CEO, Staff Conduct, Trustee Conduct, Mobile Phone, Managing Abusive Phonecalls, Overseas Travel and Subsistence, Financial Controls, Volunteer Carers Conduct, Redundancy, Private Shareholding and Financial Interests, Media Handling.

Risk Management

It was agreed that no changes to the risk register were necessary at this time. A report written by Sue Cotterell was tabled giving an update on health and safety procedures within the MPS Office.

Advocacy Support

The Senior Advocacy Officer tabled a report that was distributed giving Trustees an overview of the range and scale of work currently being undertaken by the advocacy team. Trustees were advised that the changeover from support by geographical area to disease type has been welcomed generally except for a small handful of families and this is being managed.

Research Grants

The Chief Executive advised Trustees that there had been regular contact with the Society's grant recipients over the past three years. In respect of the Genistein programme for MPS III at the University of Gdansk, Poland, the Society has received a letter requesting a fourth year extension to this project at a cost of £40,000. The Trustees considered this application and agreed in principle MPS is committed to funding this but at the present time is not in a position to grant the full £40,000. It was agreed the Chief Executive would write to all MPS Societies to seek financial support.

The Society approved a research grant to fund a unique, psychosocial research project for Morquio disease, 'Tipping the Lens', at the University of Bath. The aim of this research, the Trustees were informed, is to gain an insight into how individuals with Morquio disease see themselves.

*The **MPS Annual General Meeting 2008** took place on Saturday 3 May at Splash Landings Hotel during the Alton Towers Family Weekend. Turn to page 11 for the minutes of this meeting.*

ANNOUNCEMENTS

New Members

Mr and Mrs Powell have recently been in contact with the Society. Caitlin has a diagnosis of Sanfilippo Disease. Caitlin is two years old. The family live in the West Midlands.

Mr and Mrs Basharat have recently been in contact with the Society. Ana has a diagnosis of Sanfilippo Disease. Ana is three years old and the family live in the South of England.

Mr and Mrs Johnson have recently been in contact with the Society. Espen has a diagnosis of Morquio Disease. Espen is 15 years old and the family live in the South East.

Ms West has recently been in contact with the Society. Lyla has a diagnosis of Hurler Disease. The family live in South Yorkshire.

Mr Mohammed Pravaz has recently been in contact with the Society. Sabhaa has a diagnosis of Fabry Disease. Sabhaa is 12 years old and the family live in the Midlands area.

Deaths

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

Maggie Stokes who suffered from MPS I, Hurler Disease and who died on 24 March 2008 aged 7 years.

Caitlin Powell (MPS III) with her sister Chloe.

John Stokes

My niece's baby, John, was born on 15 January 2008 and was diagnosed with MPS VII, Sly Disease. He was born in Sligo hospital, Southern Ireland. John lived just over ten weeks. He needed a bone marrow transplant but had a lot of problems and was not well enough to have the transplant.

Sadly, John passed away on 28 March 2008. He is sadly missed by his parents, Eddie and Mary, and sister Leanne. Below is a photo of John with his parents and sister.

It hurts to know that we can't see you every day
To rock you to sleep at night or kiss your pain away
To pick you up whenever you fall
To be there when you need us whenever you call

It was hard to let you go
To watch the angels take you away
Knowing that God would watch you every night and day

We held you until the end, our hearts crushed and sore
We didn't want to let you go but we could do no more
All we ever wanted was to hold you tenderly
Praying that God would keep you here
And let you stay with me

We know the time was coming for you to go away
We knew we had to let you go
But we still begged you to stay

But you looked at us with those eyes
And you seemed to say
I have to go now I hope you will be ok

MPS Events 2008

CONFERENCES

Friday 29 – Saturday 30 August	Morquio Conference

CLINICS

Friday 11 July	Manchester BMT Clinic
Tuesday 5 August	Bristol MPS Clinic
Thursday 2 October	GOSH MPS III Clinic
Friday 10 October	Manchester BMT Clinic
Friday 17 October	Manchester BMT Clinic
Tuesday 4 November	Bristol MPS Clinic
Friday 21 November	Birmingham MPS Clinic
Friday 21 November	Cardiff MPS Clinic (tbc)

REGIONAL PROGRAMME

Sunday 13 July	Childhood Wood 15th Anniversary Remembrance Day
Friday 25 – Monday 28 July	Wiltshire Sibling Weekend
Friday 3 October	Jeans for Genes Day
Friday 24 October	Childhood Wood Planting Day
Thursday 30 October	Newcastle Get-Together
Sat 29 - Sun 30 November	MPS Adult Weekend

Cancellation of Scottish MPS Conference

It is with great regret that the MPS Society had to cancel the Scottish conference, 12 - 13 June 2008, due to a lack of interest. All funding needed was secured for this event. The families in Scotland have been written to separately regarding the cancellation.

We would look forward to hearing any suggestions as to what you feel would be helpful as an event to hold in Scotland so that we can support families in Scotland appropriately.

Please do get in touch if you have any further questions. The Advocacy Support Team are here to help with any needs you may have from housing adaptations, DLA forms, education queries and much more.

If you would like someone to be in touch with you regarding any of this issues above or even if it is something entirely different please don't hesitate to contact us on 0845 389 9901 or email mps@mpssociety.co.uk.
Miriam Blowers, Volunteer and Event Co-ordinator

Alton Towers Family Weekend

On Saturday 3 May MPS families travelled to Alton Towers to stay for a weekend of fun!

As families arrived at the registration desk I took a deep breath as it was all about to begin. All 35 volunteers had arrived, been briefed on the weekend and were being handed their care plans so they knew which families to find later on that day during lunch.

Some families had travelled for hours but this didn't seem to worry many of the children that were desperate to explore Splash Landing Water Park!

After a yummy buffet lunch and lots of catching up of old friends and some families meeting other members for the first time ever, we all headed down to the Water Park to brave the heat and rides! I couldn't believe how warm it was in the indoor swimming area and there was plenty to explore.

I was exhausted by now and thought a nap would be needed but it was straight into planning for the evening meal and making sure the children's dinner run smoothly with the right sized bowls and spoons. At about 6pm the children, who had met their volunteers by now, headed off (there were minimal tears) for dinner with the volunteers. Parents were left to mingle and enjoy a drink at the bar before heading downstairs for the Annual General Meeting and a very posh Gala Dinner.

Meanwhile, the children's meal seemed to be going smoothly and it was almost time for the children to head downstairs for some great face painting, craft making, DVDs and a magical show!

The parents enjoyed some creamy soup, chicken and amazing rich chocolate truffles for dinner and by 9.30pm it was time for the disco! Well, I have to say that I was rather impressed at some very brave parents who took to the dance floor and were pulling some impressive moves while holding children in both arms! Helen Patterson our trusty, faithful, volunteer co-ordinator hosted pass-the-parcel and plenty of disco dancing games that involved the whole family.

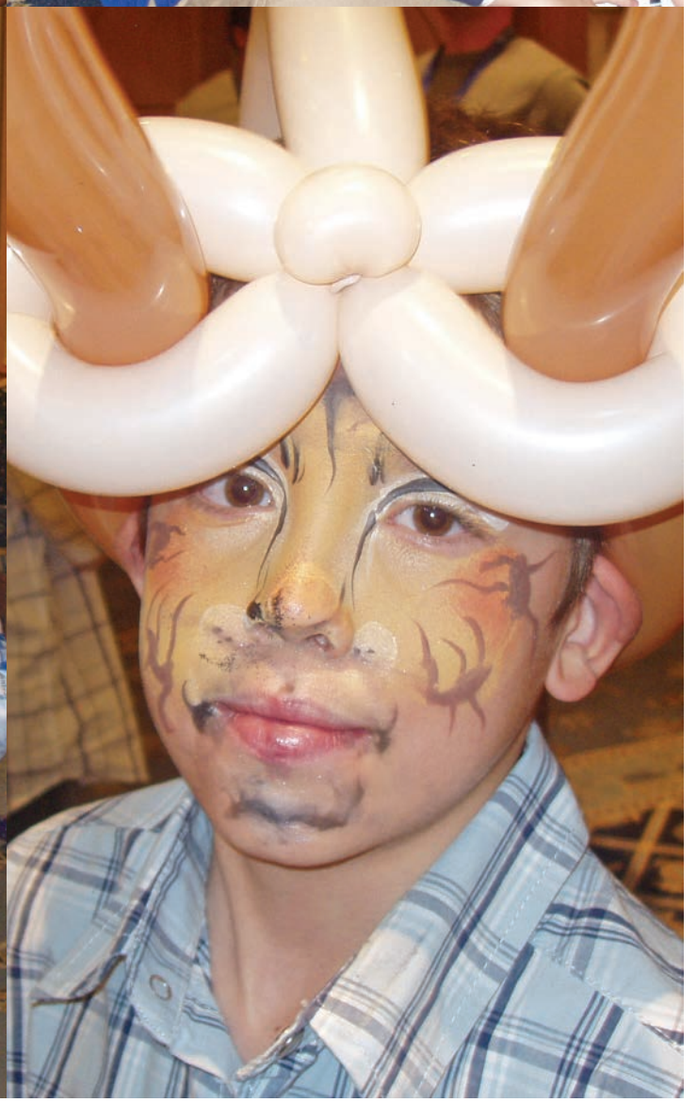
By 11.30pm I was pleased as the Event Co-ordinator that everything had gone so well. The Alton Towers team were wonderful and nothing was too much for them. The volunteers once again went that extra mile caring for the amazing MPS brothers and sisters and everyone seemed to have a ball.

Most MPS families the following morning were heading into the Alton Towers Theme Park, where I have heard it poured for most of the day! I, however, headed home. Rides are not my favourite thing in the world.

Thank you all for coming and making it such a great event and I look forward to seeing you at another MPS Conference or Family Weekend in the future.
Miriam Blowers, Volunteer and Co-ordinator
m.blowers@mpssociety.co.uk

EVENTS

Here are a selection of photos from the children's evening entertainment.



'You all deserve a massive pat on the back for everything you do as a Society.'

'A big thank you to all, fantastic as usual, brilliant weekend.'

'We really enjoyed the weekend, the volunteers put their heart and soul into helping the children have a happy time.'

'We thoroughly enjoyed the weekend, it's a great opportunity to see families and MPS staff.'

MPS Annual General Meeting 2008

The Annual General Meeting of the Society took place at the Splash Landings Hotel on Saturday 3 May 2008 at 7pm.

The minutes of the Annual General Meeting held on 1 July 2007 were distributed in advance to those members present and were accepted as true and accurate.

The Chairman, Barry Wilson, presented the Trustees report. This is published in the Annual Report and Accounts for the year ending 31st October 2007.

The Treasurer, Judith Evans presented the Statement of Accounts for the financial year ending 31st October 2007, the details of which are also to be found in the Society's latest Annual Report. It was proposed and seconded that the auditors, McLintocks and Partners, Chester be appointed the Society's auditors for the financial year ending 31st October 2008.

There being no other business the Chairman thanked members and guests for coming to Alton Towers and making the weekend such a success.

Thank you from the Murphy's

Hi! I am just writing say a huge thank you to all who helped organise the Alton towers weekend. As I know for a fact, me and all my family enjoyed it loads.

Saturday
Managed to get into the car, fully dressed, fed and off we go. After three long stops and four hours later we finally get there. Hooray! Although we were too late for lunch, in we slipped. Ooh, wish mum and dad would hurry up. Of course me and Tara can't eat anything because we were so excited. Back in Splash Landings again.

At 3pm we checked into the room. Before even unpacking, me and Tots are changed and downstairs, ready to swim, swim, swim. After an hour or two, I was starting to become wrinkly, maybe it was time to get out now. And of course we need to get ready for tonight. Getting ready didn't take long, so we all got a chance to sit and relax, watching a bit of TV.

Dinner now and it went quite quickly compared to others I'd been to. Tara and I then went to the entertainment piece of the night, Tara loved it, and so did I seeing her as the magician's assistant. I met some really nice people there including Emma and Amber, also our volunteer Anna is mint. Finally at the disco, dad and I were very happy as we both won prizes for being the best dancers. Funny I have to admit and maybe a little embarrassing but good. It was great to meet up with friends again, even though a few faces from Blackpool and Scotland were missing, but we still all had a fantastic time. End of day one.

Sunday
Once again up early. Breakfast is a quick and easy affair and now we were up and ready for the rides that the day has in store. The first ride we went on was probably the smallest in the park, the Old McDonald's tractor ride. As Tara and my dad headed off for the nutty squirrel ride, myself and my mum Maria, headed for the spinball whizzer. Didn't mum scream on that? We then joined up and went on the biggest ride Tara had ever gone on, the beastie, so tame compared to the spinball whizzer. I could tell you about all the rides we went on, but that might just bore you!

After a few rides we meet up with some more MPS families. The younger ones went on the driving test; I was much upset, being too tall. After a while the big group was spilt. The night before I had arranged to meet up with Emma, to go on a few rides. So we went on Oblivion a few times together which was scary. While I was off on the scary rides mum and dad went on battle gallions. A friendly word of advice for anyone about to go on it, don't. They looked like they were ducked under a shower. At 6.15 we go on the monorail back to the hotel, as we were staying the extra night so there would be time for more rides tomorrow.

As we said goodbye to the people we met that day and the one before, we could tell tonight wouldn't be the same without the company of our friends in Blackpool and Scotland and also the ones just leaving us… Although we thought it was going to be bad, the night was great, we drove to a country pub and had some food, and trust me I hadn't laughed that much all weekend…

Monday
I would go through all this day, but as you can probably tell, the day was full of rides, rides, and more rides.

Thanks to all who came to the weekend, to the MPS Society, the volunteers, to the families who went, it wouldn't have been the same without all of you.
The Murphy Family (Ivan, Maria, Kate and Tara (MPS I))

Thank you from the Cooper Family

Our weekend at Alton Towers kicked off with Saturday lunch. This was a good opportunity to quickly catch up with some familiar faces and meet some new families. When we had checked into The Splash Landings hotel we had looked at the Cariba Creek Water Park from reception and so were eager to venture down to the water park after lunch.

First of all Hannah played in the toddler pool area and loved the two little water slides there. When we went to investigate the main pool and surrounding area we had no choice but to get very, very wet and have lots of fun in the many sprays, gushes, torrents and canons of water. Later we discovered The Flash Flood slides and the outside pool. Hannah thought these were great and it was magnificent to hear her laugh as she disappeared off down the slides.

Late afternoon we went to our room for an hour to have a bit of a rest before the evening entertainment started. By this time Hannah had had a long and very busy day and was tired, we were anxious about how she would cope with the evening ahead especially as we were going to be in the Gala Dinner.

Hannah's volunteer helper, Jo, helped her at the evening meal and ensured that Hannah had a fantastic time during the children's entertainment. The adults and children met up at about 9.30pm after a lovely meal. By this time, and much to our relief, Hannah was all geared up and ready to enjoy the music and have a dance. She danced until about 11pm, by which time she was absolutely exhausted and ready for bed, but still smiling!

After breakfast on Sunday we caught the mono-rail into the theme park. It had started to rain and Hannah was very tired so we spent a couple of hours on a few of the attractions and then headed home. Hannah was fast asleep in the car by the time we had left Alton Towers and slept most of the way home. We had a superb weekend - a BIG thank you to everyone who helped organise the event. **Cooper Family**

Hannah (MPS I)

Calvin (Fabry) and Kyle Hall on their journey home from the Alton Towers weekend.

25th Anniversary celebrations at the Palace of Westminster

On Wednesday 14 May 2008 the MPS Society along with Jeremy Hunt MP hosted the 25th Anniversary reception at the Palace of Westminster. It was a beautiful day and the terrace pavilion was the perfect location to hold the celebrations.

Following a welcome speech from Jeremy Hunt MP, Barry Wilson, Chairman of MPS Trustees, reflected on the Society's achievements. Dr Ed Wraith presented a talk on how clinicians and the Society work together and Professor Bryan Winchester celebrated the research achievements over the past 25 years. Lady Shauna Gosling looked into the future and the importance of the MPS Society before the closing remarks and toast from Jeremy Hunt. Jeremy, Sarah Long and Myles Broughton cut the celebration cake.

Thank you to the speakers, families and professionals who so kindly gave of their time to support the MPS Society at this special event. Here are a selection of photos taken on the day. **Miriam Blowers, Volunteer and Event Co-ordinator, m.blowers@mpssociety.co.uk**

PALACE OF WESTMINSTER

On behalf of my sister, Juanita Davenport and our families, may I take the opportunity to wish the MPS Society even greater success in the years to come. It must give you a great sense of pride and achievement to see the Society go from strength to strength.

We thoroughly enjoyed our visit to Westminster as guests of Jeremy Hunt MP and the Society. We mingled and spoke to many different people.

Thank you once again for our invitation and for all the hard work by everyone at the MPS office in doing such great work. ***Sharon Chatting, Juanita Davenport and families***

Photo to the right, left to right: Leslie and Jayne Hilliard, Sharon Chatting and Mark Taylor, Phil and Juanita Davenport

MPS Awareness Day

The Society celebrated MPS Awareness Day on 15 May 2008. This is a day devoted to raising awareness for this group of 23 Mucopolysaccharide (MPS) & Related Diseases.

Spreading MPS Awareness at Great Ormond Street

Chris and Linda survived the rush hour journey on the tube into London including the sudden downpour and Chris managed to get all the MPS awareness teddy bears there safe and sound for the afternoon despite some strange looks on the tube!

As usual the MPS III clinic was very lively as families arrived mainly together, giving a great photo opportunity and time to show off their MPS ribbons and share lots of laughter and catching up with one another or meeting for the first time.

Ashley Brown kept everyone entertained with his big smiles, whilst Harris was busy tidying up the children's books and toy bricks. Sophie and Nathan made new friends with Yassin and Harris.

Then it was time for our afternoon MPS Awareness event of the day which quite literally went with a big bang! New to us, we soon discovered that the hospital has recently brought in a ban on latex balloons. Having blown up four already to promote our stand, the plan was to discreetly let them down with the kind assistance of Niamh, unfortunately with all good plans and teamwork this wasn't to be! The first balloon was very quiet but the second balloon made so much noise everyone quite literally ducked for cover and therefore we certainly made a big awareness on MPS Awareness Day!

Chris and Linda would like to say a big thank you to all the staff at Great Ormond Street especially, Dr Vellodi, Dr Cleary, Niamh Finnegan, Martina Ryan and Michelle Wood who all made us so welcome at the morning MPS III clinic.

A special thank you again to Professor Peter Clayton, Dr Stephanie Grundewald and their staff and Isaac Keighley and his parents who came and supported the MPS awareness stand. Below are a selection of photos from the day. **Chris Murphy & Linda Warner, Advocacy Support Officers, c.murphy@mpssociety.co.uk, l.warner@mpssociety.co.uk**

Photos left to right: Linda Warner and Chris Murphy (MPS Society); Professor Peter Clayton, Dr Maureen Cleary, Linda Warner, Chris Murphy, Jo, Carolina and Isaac Keighley; MPS Families, MPS Society staff and medical staff at Great Ormond Street.

MPS Awareness Day at Shire Pharmaceuticals

May 15th 2008 saw the MPS Society holding its second MPS Awareness Day. I had kindly been invited to the offices of Shire Pharmaceuticals to give three presentations throughout the day. As nerve wracking as it was, I was relieved to know that Catherine Little from Birmingham Children's Hospital had also been asked.

After arriving at the offices of Shire I was met by Janis Clayton and before too long Catherine and I were sitting in front of a room full of people waiting for us to start our presentations. Neither of us had time to think about our nervousness as we were introduced to our audience by Maud.

All together Catherine and I gave three presentations each during the course of the day, which we both hope raised awareness of MPS and Related Diseases and gave an insight of what we do at the MPS Society and what the team at Birmingham Children's Hospital do.

I would like to thank Janis and Maud for making us so welcome and for looking after us so well, to Kimberley for driving us around and for her selling skills and to Catherine who kept me going through the day.

Neisha Hall, Advocacy Support Officer
n.hall@mpssociety.co.uk

Northern Ireland

Fabry Workshop and MPS Conference

On 28 May 2008 at the Hilton Templepatrick, Belfast, a Fabry Workshop was held for MPS members, doctors, nurses, pharmaceutical companies and many more.

There were over 35 of us which was a great turn out and there was plenty of opportunity to learn more about the diseases and ask lots of questions of the expert panel that formed at the end of the session. Dr Fiona Stewart who is based in Northern Ireland as a Consultant in Medical Genetics at Belfast City Hospital chaired the meeting and supports our MPS Members in Northern Ireland.

Thursday 29 May 2008 was just as busy and full of information and ran for the full day with registration at 9am and then a lunch break at 12.20pm. The children that attended were entertained and cared for by three MPS Volunteers which included a trip to Adventure Island, a soft play indoor centre. I'm not sure who had more fun with slides and a ball pit to keep us very busy but everyone seemed to enjoy themselves. The conference speakers consisted of a mixture of professional speakers and families sharing their wisdom with the 60 plus people that attended the conference.

We look forward to the next conference in Northern Ireland and to provide the best support possible to those professionals and members alike. Thank you to all those speakers that gave of their time to share their knowledge. Special thanks go to Dr Fiona Stewart who worked closely with us in organising the event.

Miriam Blowers, Volunteer & Event Co-ordinator
m.blowers@mpssociety.co.uk

NI CONFERENCE

Our life with Sophie

17 September 1999 - 21 June 2007

Where to begin, to capture the life of a vibrant soul trapped in a body that is not growing or developing in the right way?

Sophie had Hurler Syndrome. She was diagnosed at our local hospital in Exeter a few days before her first birthday. A few days after her birthday the diagnosis was confirmed by Dr Ed Wraith in Manchester.

The possibility of a bone marrow transplant was raised and so within the space of a couple of weeks the expectations of a new family life in Devon, where we had moved five months previously, were shattered and the next three months were spent in an anguish of discovery and debate. We were greatly helped by doctors in Bristol and Manchester who patiently gave us as much information as we requested and answered all our questions as best they could. Eventually we decided against bone marrow transplant, for many reasons, but principally our overriding aim was to keep as normal a family life as possible and subject Sophie and all of us to as little time in hospital as possible. Also, from a practical point of view it would have been almost impossible for us to manage at that stage with other children to look after and no additional support available from family or friends as we were so new to the area.

The decision about bone marrow made, life slowly began to take on a certain normality albeit quite different to how it had been and how we had expected it to be. At that stage Sophie's two brothers were 11 and 3; family life was hectic and the initial shock and disbelief of Sophie's diagnosis was replaced with the joy of watching and helping our daughter develop and become an integral part of the family. Slowly, slowly she learned to get around; bottom shuffling at first, next walking with an aid and the magical day when she all of a sudden got up and walked on her own. Slowly she learned to communicate; first, using simple Makaton signs - 'more' was her favourite for anything involving chocolate, then she moved onto proper words and later she was able to put words together in sentences. Delightfully Sophie learned to sing - we did lots of singing.

When Sophie was just three we were asked whether we would like her to participate in a clinical trial for enzyme replacement therapy for children with MPS I. We were aware that this would not be a cure for Sophie but we hoped it might delay the onset of the inevitable loss of her skills and generally help her physical wellbeing. It was a challenge to get to Manchester from Devon each week and we could not have managed it without the enormous help of Sophie's granny, but it was worthwhile. We got to know other children with similar conditions to Sophie's and the Willink team were great fun and very supportive. Sophie loved the treatment area, particularly the double buggy for her babies; we did lots of colouring, sticking, story reading and watching teletubbies. We were also pleased that Sophie's life could contribute to part of the learning process on the long road to finding a cure for children with her condition.

Also at three Sophie started at our local pre-school as well as attending a specialised centre for children with special needs one morning a week. At pre-school she had a one to one helper but soon dispensed with her help in true Sophie fashion, she was much more interested in her own little band of friends. Megan took Sophie under her wing and would meet us at the door each morning and take Sophie off by the hand. Sophie also had a special friend in 'the' Ned, as she called him, and he always looked after her even after they both finished pre-school and started school. Milestones were reached, for instance Sophie climbing the steps unaided to go down the big slide outside and we managed to persuade her to dress up as Mary for the Nativity play. When Sophie was four her little brother Toby was born. Fantastic, she now had her own real baby to smother with blankets and care for, 'Toady' as she called him.

Interspersed with this there was of course the inevitable round of hospital appointments and various operations which were always hugely stressful but I look back on this time as the golden time with Sophie when she could walk, talk, make jokes, sing, do puzzles, laugh and generally make herself the centre of attention. This time was all too short. Even before she started school just before her fifth birthday Sophie was slowing down. She sat and rested a lot more, gazing into the distance and she would start a song but then seemed to forget the words two lines in. We had our last holiday abroad with Sophie just before she started school. We could still manage without specialist equipment at this stage; we took her Tripp Trapp chair and a buggy but she could still sit up in the bath and we could still do short walks carrying her in the backpack.

Sophie went to a small local primary school. It was quite a challenge getting school uniform to fit. We tried many different options and I was always on the lookout for T-shirts with large head openings and stretchy skirts to fit comfortably over her tummy. The staff at school were wonderful and from the beginning they relished the chance to be able to help Sophie and give her as good an experience of school as possible. She had a wonderful and dedicated full time helper and lunchtime helpers. Sophie had school lunches for the first two years and the school meals' service eventually had to puree them. When this no longer worked because swallowing became difficult I took in her own homemade pureed food each day.

The children loved Sophie and accepted her for who she was. They dressed her up and pushed her around the playground in breaktimes and made her sensory pictures and toys. She was included in every activity and had a part in every school performance. When the children had an afternoon learning to play African drums, Sophie was helped to bang on the drums, when pets were taken to school, Sophie held all of them on her lap including the snake! She certainly contributed to school life. On one occasion the headmistress had to give up on assembly because Sophie had a fit of giggles and everyone around her couldn't help but join in until the whole school was laughing and of course nobody knew why! She was the only child who had her birthday parties in school. I took in chocolate cake with Toby and Granny and there were crisps, drinks and games.

During the first term at school Sophie was finding things increasingly difficult. She could still talk but did not want to walk. After Christmas the situation changed dramatically; she lost all her sight suddenly and within the space of three months was no longer able to sit at the table unsupported or in the bath or on the loo. The house filled up with specialist equipment and she had oxygen at night and a number of medications which we kept under constant review to help keep her comfortable. She stopped speaking but we could still make her smile and we put A LOT of effort into this. It was really hard when she stopped even being able to smile - then we just had to imagine that she was smiling inwardly at our silly antics even though she could not show it. Despite the huge loss of skills Sophie still attended school for three years and was able to go to school until three weeks before she died. Sophie died peacefully in her sleep at home.

She was a beautiful, sunny, happy girl, she enriched all our lives and the lives of so many people who came into contact with her. We are sure she had a happy life, that is all we really wanted for her. **Sally Richards**, May 2008

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

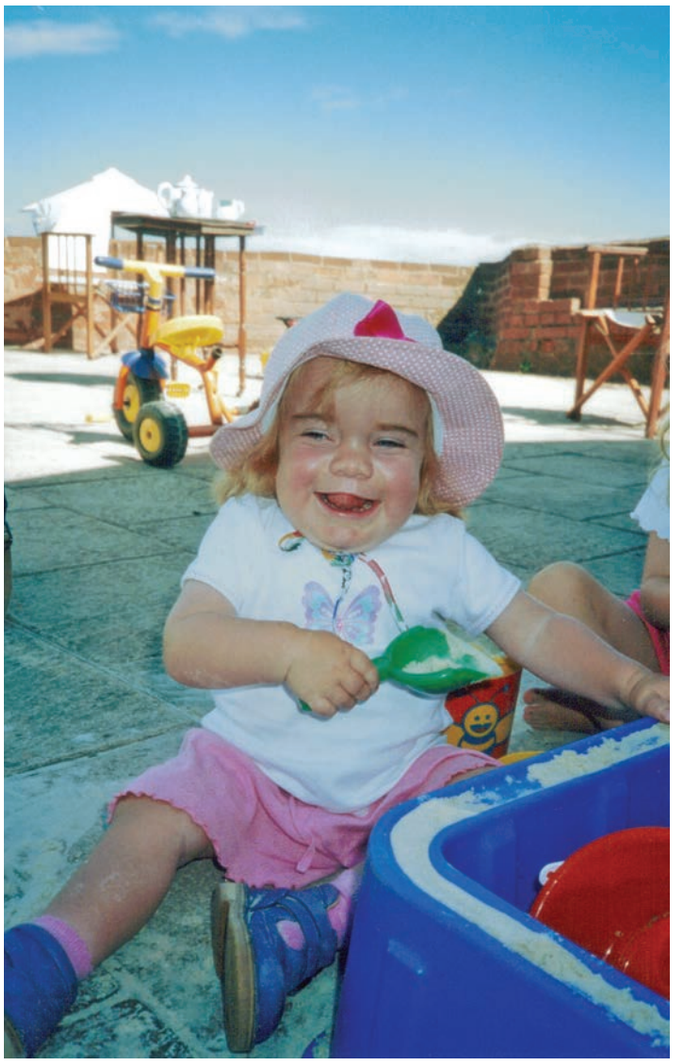
A Picture of Sophie

There once was a child
Sunshine smotes in her hair
Of ever changing colours

Wide smile; deep belly guffaw
Chocolate buttons, chocolate cake - 'more'

Thick wool cuddly cardigans
Feet planted firmly, round tummy pushed forward, considering…
Then determined, head inclined,
arms swinging… forwards
Bag clutched in her hand or hooked on her shoulder

Soft, softest skin so peachy kissable
The joy of her forever intermingled
with the dreadful sadness
Of the ephemeral nature of her being your child.



Palliative Care: A Patient's Perspective

We are the parents of four daughters. The eldest Louise died peacefully at home last August at the age of 28 from Sanfilippo disease. Her three younger sisters are now aged 26, 23 and 18.

We would like to share our experiences in caring for our Sanfilippo daughter at home, and how we believe she perceived her life with us.

There is no treatment for Sanfilippo and our aim has always been to provide as full a life as possible for Louise while also giving our three other daughters a normal childhood.

These aims may seem mutually exclusive, but we have always felt that Louise was with us for a purpose and had a contribution to make. If Louise had to go through her life with Sanfilippo disease we had a responsibility to make sure that purpose was not lost.

Anne trained to be a teacher, but has always been a full time Mum, and we could not have managed any other way. I was a teacher and had the same school holidays as Louise, which was a big help.

Whenever we went on family outings our strategy was always that I look after Louise while Anne looked after the other three girls. This worked well while Louise was robust, but we knew the prognosis and had seen more advanced symptoms of the disease at MPS conferences. We knew we had to plan for the future if we were going to keep Louise at home with us.

With hindsight, planning for a worse case scenario has been quite a good coping strategy, as we can honestly say that no stage has ever been as difficult as we imagined - even the final ones!

We decided it would be better if both of us were to be available and working flexibly around Louise's changing needs in order to keep a happy family life.

I was fortunate enough to be able to start a farming business growing trees and fruit to enable me to work from home and build a house suitable for a large family with one member becoming increasingly immobile and in need of constant care. We moved in when Louise was 18. At this stage she was still walking.

We had used respite care to enable us to take family holidays and occasional breaks with the other three girls. It seemed as if Louise always returned with an infection, and when Louise was ill life was very difficult. It just didn't seem fair on Louise or worth all the bother.

A children's hospice opened near us when Louise was 16, and we all went and stayed as a family. Louise was cared for beautifully as were the rest of us. This truly was respite for us, and everyone looked forward to the next visit. Unfortunately when Louise turned 18 we were no longer able to go as she was no longer a child.

It is worth mentioning here that hospice facilities for young adults in England are sadly lacking as the charities that try to run them cannot present themselves in a way that is as attractive as childrens' hospices. We both have a passion for changing this and hope to some day.

We realised then how much we needed help, and got funding for a carer for Louise to come into our home for a few hours a week. This grew into a full time job as Louise left school at 19 and the day centre she then attended struggled to give her the standard of care we demanded. She was losing her mobility now.

We bought Louise a car, and her carer would get her up when she awoke naturally, usually towards lunchtime, wash, dress and feed her, and then take her out for the day to see the sea, go shopping or simply a ride in the country. These were happy years and her health was stable. Louise had a lovely relationship with her carer and we were fortunate to have found someone so special.

Another perspective that hindsight gives us is how much Louise's quality of life and health improved when we stopped trying to make her fit into our routine, and let her choose her own routine. For years she never even caught a cold! Maybe this was partly due to that she was not always mixing with other people with streaming noses and coughing all over her, but I believe she was genuinely happier this way. We should have stopped forcing her to get up early for school only to spend half the day asleep in her wheelchair years before we gave up trying!

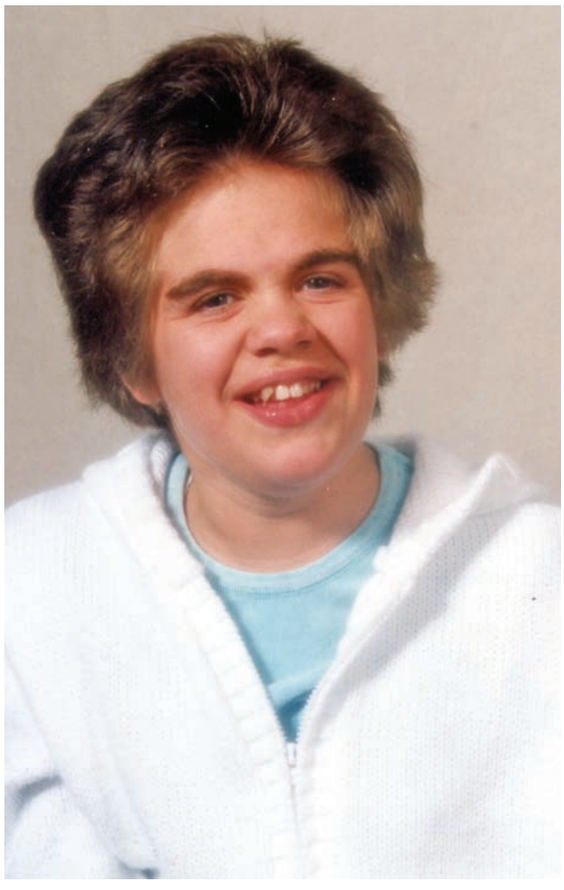
Two years later she started choking when drinking, and the ENT specialist suggested fitting a gastrostomy device to enable us to put liquids and eventually all her food directly into her stomach as the disease took its course. We are so glad that we did this while Louise was still robust and before she was in desperate need of it, as she never had any problems with it.

To avoid visits to hospital, starving Louise beforehand and waiting in line, we learned to change the gastrostomy buttons ourselves, and always made a point of learning to do any techniques necessary in Louise's care so that in an emergency we should be able to cope. This was as much for our own peace of mind as for Louise.

While talking about food and drink her dental hygiene comes to mind. Louise always hated having her teeth cleaned and would resist all attempts by biting the brush or my finger or keeping her teeth tightly gritted. Visits to the dentist were a nightmare and thankfully she never needed any treatment. We partly put this down to her drinking only water and having no sweets or chocolate from about the age of 9. This may seem cruel, but the thought of Louise having toothache and us being unable to tell what was wrong seemed worse.

At the age of 22 came her first seizure. We had been warned of the likelihood of Louise having fits and it was something we had always dreaded. Even though we had medication in the house and been told what to do, we were afraid to use it, so called the doctor. That was a difficult 10 minutes waiting for him. We resolved never to put Louise through that again if we could help it, and kept several tubes of rectal Diazepam by her bed and learned how to use them.

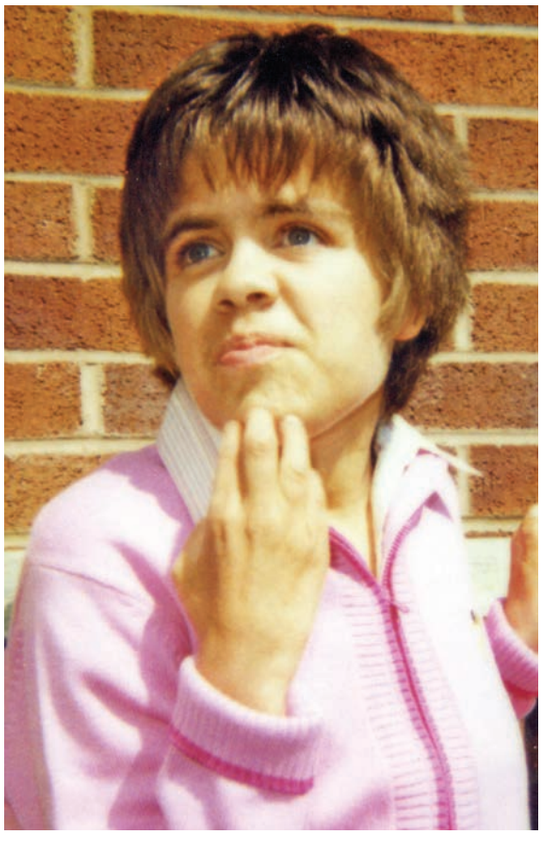
These seizures or severe, regular muscle spasms were perhaps the worst part of the disease's progression. They would occur at any time of the day or night and go on for hours. Epilim, Tegretol and Baclofen all failed to stop them, and at this point we were allocated a hospice nurse who would be our main link with the medical profession. She moved us from rectal Diazepam, which didn't work when Louise had a bowel movement during a fit, to buccal Midazolam, which was more effective, but was distressing for her as it made her cough and choke. [cont.]



Choking on her own saliva due to losing her ability to swallow made life miserable for her so she was prescribed Glycopironium tablets and Scopoderm patches to dry her mouth. This made the buccal Midazolam more effective in controlling the spasms.

In October 2004 the spasming increased dramatically. By now she was getting more support from the doctors at the adult hospice, and they decided a syringe driver constantly injecting Midazolam subcutaneously could be the answer. Their motto was 'go with what you see'. If one dose didn't stop the fitting she needed another, and so on.

At one stage she was on 160mg Midazolam per 24 hours which we are told would knock out an elephant, and she still would not rest. We were warned that this couldn't continue, and that Louise might not survive the weekend. All her sisters came home to say goodbye and we went through a lot of grief together. Louise however calmed down as if someone had turned a switch and so we in turn reduced the Midazolam. Had we not taken it upon ourselves to reduce the dose we are sure she would have died then.



This was the start of a yo-yo period in her fitting. The seizures would gradually decline and stop and we would wean her off the Midazolam and eventually remove the syringe driver. We learned how to insert the needles to change the injection site and became quite confident in guessing what dosage would keep her comfortable. We continued in this way for three years, making constant assessments of Louise's state, which meant one of us had to be with Louise constantly, as we couldn't ask someone else to make those judgements. This was quite a stressful time and we both felt the weight of responsibility.

Throughout this period Louise's quality of life deteriorated slowly. So slowly that we barely noticed it. Much of her life was spent in bed or in a moulded chair around the home, with outings in her car only on lovely days when she wasn't spasming or choking.

In early August last year we noticed Louise was not wetting her pad as much as usual and called the doctor on Monday 6th August. He poked and prodded her and said he had no idea what was going on. He came back the next day and examined her again. This time he concluded that she was suffering organ failure and would die within a few days. Having been told that before we didn't take him too seriously, and the following day took her to the seaside to enjoy the sunshine, something we don't take for granted in England! We had a lovely time.

When the doctor returned on Thursday he was even more sure Louise would survive only days and prescribed medication which would sedate her if her breathing became difficult and she became distressed over the weekend.

We called her sisters home again and spent a close family weekend together around Louise. Amazingly there was laughter as well as tears that weekend. Louise spent the time in bed in her own familiar room in the centre of the home, and we are sure she was aware of the love surrounding her.

By Sunday night Louise seemed no worse and so one of her sisters returned to York to sit an examination.

We both felt exhausted by the emotional turmoil and when Louise got us up to change her pad at 4am and again at 6.30am we felt we just couldn't keep this up. An hour later while Sally's fiancé was eating breakfast he heard Louise sigh deeply, which we now realise, were her agonal breaths. He woke us as she was unresponsive, and we soon realised she had left us. We turned her on her side as we saw mucous in her mouth but made no attempt to resuscitate her. The doctor came within 10 minutes and confirmed she was dead.

As it was summer we washed her and cooled her body in our walk-in cold store so that we could put her back into bed before her sister returned from York. We then made a small chapel of rest in the cold store and kept Louise's body there until her funeral so that anyone who wanted to could spend a few quiet moments by her.

Throughout this whole journey we wanted the very best for Louise and also wanted to have no regrets after she had gone. We feel that coping with Sanfilippo disease within our family has helped us to deal with our grief in losing Louise. Other families may find different ways of coping. At no stage did we feel we had lost control of her care. The doctors and nurses we worked with made us feel very much part of Louise's care team, and we felt able to speak out if we didn't agree with what we thought Louise would want. We do feel however that we both needed to be involved 24/7 to support each other, both physically and especially emotionally through the turmoil particularly towards the end of her life. We could not have done this alone or if one of us had had other demanding commitments.

We can honestly say that we are glad that we did what we did for Louise, and our only regret is that we weren't strong enough to sit up with her and hold her hand through her last night on earth.

Finally, we need to acknowledge the support we received from all the doctors and nurses involved in Louise's care, Christine and MPS, Louise's physiotherapist, and Janet her carer, without all of whom we would not have been able to cope.
Gordon and Anne Hill

Your news and views

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

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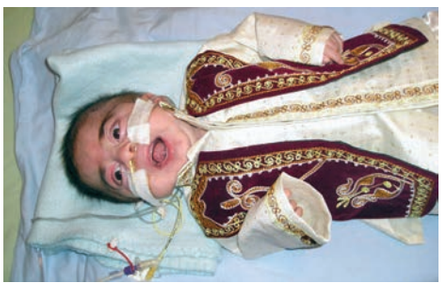
Aayan Hussain

27 April 2007 - 5 February 2008

Thank you all at the MPS Society for all the help you have given us. Words can not describe how grateful we are. **Iqbal and Neelam**



Helen and Douglas House Hospice celebrates 25th anniversary

This year as well as the MPS Society, Helen and Douglas House celebrated their 25th anniversary. To mark the end of their celebrations for their 25th year, the Helen and Douglas House Anniversary Concert was held at St.John's Smith Square on 19 March 2008. The concert was given by the London Chamber Orchestra. Both my husband and I were asked to attend.

As patron not only of Helen and Douglas House, but St.John's and the London Chamber Orchestra, Her Royal Highness The Duchess of Cornwall attended the concert along with His Royal Highness The Prince of Wales.

We were asked to bring photographic ID along with our invitations. We were also asked to make sure that we were seated in the venue by 7.15pm. There was a huge police presence around St.John's and residents were not permitted to park around the area for this special occasion. Her and His Royal Highnesses arrived under police escort and sat in front of the middle isle.

The London Chamber Orchestra conducted by Christopher Warren-Green performed pieces by Elgar, Bruch, Hess (Hess Concerto for piano and Orchestra, the first London performance of this special concerto, written in memory of HM Queen Elizabeth The Queen Mother) and Beethoven.

After the concerts, there was a drinks reception in the Vaults. It was an immensely enjoyable and a very memorable special event.

We made our way back to our coach at Victoria bus station on a high note and arrived home in the West Country early hours of the morning. **Fer Pidden**

HRH Prince Charles with Sister Frances Dominica, Founder and Trustee of Helen and Douglas House

Thank you to Dreams Come True

Matthew is our 13 year old son who suffers from MPS II. Recently we were introduced to a charity called Dreams Come True **www.dctc.org.uk** by Chris Murphy in the MPS office.

We were soon in touch with Martin at the charity who explained the process of requesting some sensory play equipment for Matthew. Following a brief discussion and the expected confirmation from our GP of Matthew's condition, we asked for a bean bag and some fibre-optic lights. As you can see from the picture on the right, Matthew is making the most of the new equipment.

The bean bag is large, waterproof, and moulds to fully support Matthew when he is playing with his twizzle sticks, lights or watching television. The lights themselves are multi-core fibre-optics that colour phase. They provide stimulation not just from a visual perspective but Matthew really enjoys the feel of them in his hands.

We would like to thank Martin and the team at Dreams Come True for the fantastic work the charity is doing. We would also like to thank everyone at the MPS Office for their continued support for Matthew, and making us and other families in our position aware of these opportunities. **Jacqui and Steve Home**

Three's not a crowd!

Liz (18) has recently joined Hugh (20) and me (48) having her infusions at home. She wants to learn to self-infuse too over the next few months. Then we shall all be independent. It's great to be able to choose when we have our infusions. We usually choose a Sunday when Hugh travels down from university. That way I can look after the storage of the drug for the three of us, check the stocks of ancillaries and arrange to be in for the deliveries. I expect Liz will do the same when she goes to university.

Careology have helped us to learn to be independent and although we still get a little nervous when carrying out the procedure we support and encourage each other and my husband, Justin, provides an excellent

You are important to us, please keep in touch.

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

Your news and views

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Lewis on a high as RAF give family £38k grant

A little boy with a rare terminal illness will soon have his dream home thanks to a £38,000 donation from the RAF.

Lewis Broadley (10) was diagnosed with the condition Sanfilippo disease (MPS III) at the tender age of four, and has lost his hearing and speech.

Now becoming unsteady on his feet, Lewis will shortly need 24 hour care and a safe environment in which to live.

For the past year his parents, family and friends have been raising funds and working towards converting the ground floor of the family's home into a special bedroom for him, complete with special sensory equipment.

The family were given planning permission for the conversion by Falkirk Council and work started earlier this year. Now a major grant from the RAF Benevolent Fund has helped the project along.

Mum Norma said: 'This has made our lives much easier and I can't thank the RAF enough, they've been fantastic.'

Lewis's dad Gordon served with the RAF when he was 17 and was involved in the clean-up operation on the Falklands Islands during his service.

He got in touch with the Soldiers, Sailors, Airmen and Families Association to see if they could help with the cost of the conversion.

The SSAFA, a registered charity which provides help and support to current or former service personnel and their families was instrumental in securing the funding from the RAF.

Andrew Wood, SSAFA deputy branch secretary, said: 'It was quite a success story getting such a significant sum and the family were delighted when the RAF came back with that amount.'

Lewis and his family are currently staying elsewhere as the work continues in their home.

'We hope to move back in the next couple of weeks,' said Norma.

'We couldn't have wished for anything better - Lewis's room looks amazing. We're really grateful to Andrew and SSAFA for helping to make this happen.'

For more information on SSAFA call 01786 860364.

Article written by James Trimble appears courtesy of the Falkirk Herald, Thursday 12 June 2008.
www.falkirkherald.co.uk

Bristol MPS Clinic

In April I attended the Bristol clinic. It had been a while since I had attended a clinic at Bristol and the thought of having to drive into the centre of Bristol was a little daunting. However, my memory didn't fail me and I found the hospital and thankfully somewhere to park. The clinic was held in the out patient department and Dr Jardine and Dr Wraith were joined by other doctors from the hospital. The clinic ran very smoothly and this was thanks to the nursing team who were assigned to the clinic for the day. It was really good to see and catch up with so many families some who I have not seen for some time.

Although this clinic was held at the Children's Hospital we hope that future clinics will be held at the Children's centre. This will hopefully afford us all accessible parking something I know is extremely difficult to find at the Children's Hospital.

Lastly, I would like to extend our thanks to Dr Wraith and Dr Jardine for their support at these clinics. **Sophie Thomas**

Northern Ireland MPS Clinic

On 30th May I attended the Northern Ireland clinic. Due to the conference being held the day before and many of us staying over in the hotel, Dr Fiona Stewart took Dr Wraith and I to the hospital where the clinic was being held. This was on the proviso that we did not make any comments about her car. We both said nothing!

As always the clinic was very busy and we even had a film crew in during the morning. However I was not too happy about being filmed especially without any prior warning, no make up and hair not looking its best! Hopeful my part will be edited out!

Once again it was lovely to catch up with so many families, some whom I have not seen for some time.

On behalf of the Society I would like to pass on our thanks to Dr Fiona Stewart, her team and Dr Wraith for another successful clinic. **Sophie Thomas, Senior Advocacy Officer, s.thomas@mpssociety.co.uk**

Photos clockwise from top right: Hannah Shannon (MPS III), Santana McDonagh (ML II), Jade McAfee (MPS III), Brooke Harvey (Fucosidosis), Clare McDonagh (ML II)

Birmingham MPS Clinic

Friday the thirteenth! 13 June, what a date to have a clinic, but we were all optimistic and positive that this date was not going to have any influence on the day.

I arrived at the hospital to the very welcoming team of Catherine, Sat, Dr Hendriksz and Suresh. The only person missing was Dr Chakrapani; but I had been told he was on call so would be popping in when he could.

The clinic went extremely well and it was lovely to meet up with familiar families, and also to meet new families. It was also very beneficial to have been allocated a room which I was able to use for families to discuss any issues they may have and needed support with.

I would like to thank all the team at Birmingham Children's hospital, Dr Chakrapani, Dr Hendriksz, Dr Vijay, Catherine Little and Sat, for making me feel so welcome again, and for their continued support. I would also like to thank Louise Simmons for arranging the clinic and getting all the information to me, and wish her well with her impending new arrival.

Neisha Hall, Advocacy Support Officer
n.hall@mpssociety.co.uk

Photos clockwise from top right: Jebran and Shabana Shoukat (MPS IV), Toby Martin (MPS III), Sultan Ali (MPS IV), Luke Edwards (MPS II), Caitlin Powell (MPS III), Dwain Caines (MPS III)

MPS III Clinic

Our Sanfilippo clinic at Great Ormond Street Hospital on 15 May 2008 happened to be on MPS Awareness Day this time so we had a full schedule. The children and families were seen by Dr Cleary and Dr Vellodi and also by Martina Ryan, our speech and language therapist and Michelle Wood our physiotherapist. We were also joined by Chris and Linda from the MPS Society.

It was my job to co-ordinate everyone and run around trying to ensure that everyone was in the right place at the right time! It was a very noisy and lively clinic where old and new families had the opportunity to meet and chat.

This was our fifth Sanfilippo clinic and families seem to like this multidisciplinary approach and the opportunity to chat to other families. We hope to continue this format twice a year. **Niamh Finnegan, Metabolic Clinical Nurse Specialist, GOSH**

Cardiff MPS Clinic

On 6 June 2008, Linda and I made our way to Cardiff on Thursday afternoon and got a good night's sleep so we were ready for a full clinic at the Cardiff University Hospital.

The clinic list was a full one and seemed to run quite smoothly. It was lovely for both Linda and myself to meet the families, some of whom we had met and some new faces to us both. Everyone made us feel really welcome and even though the waiting room was at times very busy with the different clinics going on, we still had time to spend with the families. Linda even had the time to have a deep discussion with a young man about football - and we won't be mentioning any football teams or players!

Linda and I would like to thank Dr Ed Wraith and Dr Graham Shortland for their continued support at these clinics. Also, a big thank you goes to Judy Holroyd and to Sally Davies for their support at this clinic. **Neisha Hall, Advocacy Support Officer n.hall@mpssociety.co.uk**

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

Photos clockwise from above right: Yassin and Harris Mahmood, Mrs Mahmood and Chris Murphy; Tim and Sally Summerton, Sophie Summerton, Dr Maureen Cleary, Harris Mahmood, Niamh Finnegan, Chris Murphy; Ashley Brown.

SPOTLIGHT ON FABRY

Amicus Therapeutics announces results of Phase 2 clinical trials for AT1001, a pharmacological chaperone under investigation for Fabry disease

Amicus Therapeutics, Inc. is developing orally administered compounds called pharmacological chaperones for the potential treatment of lysosomal storage disorders, including Fabry disease. These compounds are designed to selectively bind to and stabilise enzymes. It is hypothesised that stabilised enzymes are able to exit the endoplasmic reticulum (ER) and be sent to the lysosomes where they are needed to break down substrates.

Amicus recently announced preliminary results from Phase 2 clinical trials of AT1001, a pharmacological chaperone for Fabry disease. The primary objective of the Phase 2 program is to evaluate the safety and tolerability of AT1001. The studies also examine the effects of AT1001 on alpha-GAL (the enzyme deficient in Fabry disease) and GL-3 (the substrate that builds up in the lysosomes).

The Phase 2 trials of AT1001 involve five different studies. Four studies included either 12 or 24 weeks of treatment with AT1001 with an option to continue treatment in the fifth extension study. Different dose levels and different intervals for dosing (for example, taking AT1001 every day or every few days) are being tested. Twenty-six subjects with Fabry disease (17 males and 9 females) completed the primary treatment phases of the clinical trials for AT1001. Some highlights of the results to date include the following:

AT1001 was generally safe and well-tolerated at the different dose levels studied. Drug-related adverse events reported to date have generally been mild to moderate and resolved without intervention.

Thirteen of the 17 males were classified as 'responders' to AT1001 because they demonstrated significant increases in alpha-GAL levels after treatment (defined as a net increase greater than 3% of normal enzyme levels).

Five of the 9 females were classified as responders to AT1001 based on in vitro characterisation (laboratory testing) of their mutations. Since women have two copies of the GLA gene, a laboratory test was used to determine the expected response.

Treatment with AT1001 was shown to increase alpha-GAL levels in white blood cells, kidney, and skin.

Reduction in kidney substrate accumulation was assessed by measuring GL-3 levels in urine. The majority of male and female responders showed a decrease in GL-3 levels in the urine.

A majority of the responders self-reported an improvement in their Fabry symptoms.

Twenty-three subjects continue to receive AT1001 as part of an extension study. The extension study will continue while meetings are held with regulatory authorities to discuss the design of a Phase 3 clinical trial for AT1001.

In November '07, Amicus Therapeutics and Shire Human Genetic Therapies entered into a partnership to jointly develop AT1001, along with Amicus' two other pharmacological chaperones (AT2101 for Gaucher disease and AT2220 for Pompe disease). Amicus will develop these products in the United States while Shire will lead development outside the United States beginning with Phase 3 studies. Through its Patient Advocacy Department, Amicus will continue building its relationships with lysosomal storage disease patient organisations such as the UK MPS Society.

Please direct any questions to patientadvocacy@amicustherapeutics.com.

Joint LSD Patient Meeting

This is the first time adults with Fabry, Gaucher and Pompe Diseases have had a joint meeting. Dr Mehta opened the meeting to a very full audience.

The primary aim of the meeting held on 15 June 2008 at the Royal Free Hospital was to be patient focussed and in the morning we heard from Dr Derralynn Hughes who spoke on the Clinical Management of LSD's and new and emerging therapies. Ian Hedgecock gave a very personal account of living with Fabry Disease.

His recollection of how he felt around the time of diagnosis and how he developed strategies for problem solving and achieving goals were very poignant. Equally other speakers Michael, Juanita and Elizabeth gave very valuable insights into their lives with their respective diseases, and in the case of Juanita her wider family. We would like to wish Juanita's nephew a successful tour of duty and a safe return as he heads off to serve as a Para Trooper in Afghanistan.

The afternoon session was when we were split into three groups, Fabry, Gaucher and Pompe. These were workshops which concentrated on ERT, when to start, and what are the benefits.

Christine and I would like to thank the team at the Royal Free Hospital for inviting us to take part in such an informative and well organised meeting. **Neisha Hall, Advocacy Support Officer, n.hall@mpssociety.co.uk**

Hunter Outcome Survey

In the Summer of 2007, the MPS Society undertook to verify the diagnosis and death data of all known adults and children with MPS II, Hunter disease, in the UK. This was undertaken by an undergraduate student who carried out a telephone questionnaire and obtained from the Registrar General the death certificates of the affected individuals. With the informed consent of all families this data was added to the Hunter Outcome Survey (HOS) with the collaboration of the Royal Manchester Children's Hospital.

As you will see, out of 129 historical deceased patients in HOS, 106 are from the UK. This data shows us that the average age of the life expectancy of Hunter disease is 13 years and the most common causes of death were reported as pneumonia followed by respiratory and cardiac failure. From collecting the death certificates we saw that many had the diagnosis of Hunter incorrectly recorded, sometimes stating obscure definitions such as Hurler syndrome, mucosaccharidosis, gargoylism, Hunter-Hurler syndrome, mental subnormality, mongol, congenital MPS disease or not showing any indication of an MPS disease at all.

We have to conclude that for families who have lost relatives with MPS II, and who may rely on death certificates for a family history the information is at risk of being inaccurate and professional advice should be sort. **Christine Lavery, Chief Executive, c.lavery@mpssociety.co.uk**

hos hunter outcome survey

Age and Cause of Death in Patients with Hunter Syndrome (mucopolysaccharidosis type II): Data from HOS – the Hunter Outcome Survey

S Jones[1], K Aleck[2], Z Almássy[3], M Beck[4], K Burt[1], J Clarke[5], R Giugliani[6] L Lavery[7], G Malm[8], U Ramaswami[9], R Tincheva[10], JE Wraith[1] on behalf of the HOS Investigators

OBJECTIVES:

Hunter syndrome is a rare, progressive, X-linked disorder of glycosaminoglycan (GAG) metabolism. It is caused by deficiency or inactivity of the enzyme iduronate-2-sulfatase, and results in progressive accumulation of GAGs within tissues and organs throughout the body. Death occurs mainly as a result of cardiac or respiratory failure, often in the first or second decade of life, especially in those patients with the most severe form of the disease affecting the CNS. The aim of the present study was to use historical mortality data collected in HOS and by the UK Society for Mucopolysaccharide Diseases to examine the cause of death in patients with Hunter syndrome in order to improve prognosis and current patient management.

METHODS:

HOS was initiated in October 2005 as the first and only global outcome survey of the natural history of Hunter syndrome and the long-term safety and efficacy of enzyme replacement therapy with idursulfase (Elaprase™; Shire Human Genetic Therapies, Cambridge, MA, USA). The latest analysis of the HOS database was conducted on 15 May 2008. At that time, data from 550 patients were available, including 135 death records (129 historical, mainly from the UK, and 6 after entry into HOS). Death certificates for those patients who had died before the initiation of HOS and were entered retrospectively into the database were reviewed to determine the cause of death. This analysis focuses on 129 historical patients from the HOS database with available data (Fig 1).

RESULTS:

The median age at death was 13.0 years (10th–90th percentile, 6.0–26.0 years) (Fig 2). The most common causes of death were reported as pneumonia (26% of historical deceased patients), followed by respiratory failure (15%) and cardiac failure (13%) (Fig 3). Kaplan–Meier analysis of the data in HOS revealed a difference in the age of death for patients with and without cognitive problems (median survival, 11.7 versus 13.9 years, respectively; p = 0.027 [log-rank test]) (Fig 4).

CONCLUSIONS:

By collecting mortality data, HOS is providing important information on the natural history of Hunter syndrome. The present analysis confirms that cardiac and airway problems are significant causes of death. The reporting of pneumonia as the most common cause of death reflects the final event in a multisystemic disorder.

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10 Genetic Institute, Children's Hospital, Sofia, Bulgaria

Acknowledgments for HOS data, 15 May 2008

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Sioux Falls, SD: Laura Keppen
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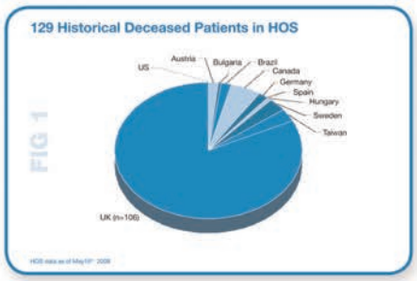
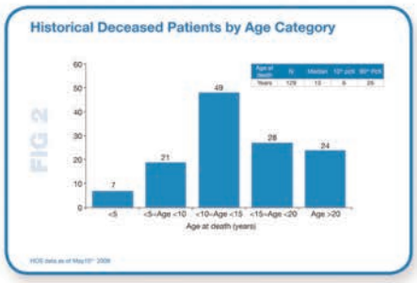
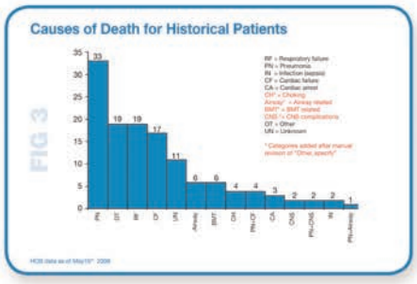
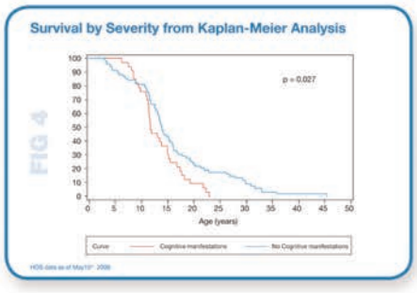








10th International Symposium on Mucopolysaccharide and Related Diseases, Vancouver, Canada, June 26-29, 2008.

SPOTLIGHT ON MORQUIO

Introducing the 'Tipping the Lens' Project

Individuals are telling their stories
A small team of individuals have decided to 'Tip the Lens' and take control of defining what it means to live with Morquio (MPS IV). Tipping the lens away from X-rays and MRI scans, medical examinations and being told about how it is going to be by professionals. The Tipping Lens Project has given people a copy of the viewpoint book and single shot cameras so that they can join this unique opportunity to tell their story using their own words and images, saying how it really is living in their body, their viewpoint of their world; importantly what makes them, them. Participants only have to share with the project what they are happy to. It is their chance to show people what their world is really about, what they would like to change in the world (if anything) and what they really value in their life. The project is a participatory piece of social research designed to empower people living 'it' to communicate what their ordinary everyday life is like.

Facilitating the Project
The MPS Society is working in collaboration with postgraduate students at the University of Bath. One of the researchers is myself - Sarah Long and I live with Morquio (MPS IV). Having worked in the disability rights movement mainly in the not-for-profit and public sector for over ten years, I'm currently in my first year of a PhD at the University of Bath in the field of Sociology and Disability Studies. I really value individuals working with me on this project as I feel it's important that we share our experiences, say what it is really like, and also importantly have some fun along the way! My co-facilitator is Erin Abshire, a Masters student at the University of Bath who is interested in Participatory Photography projects and who has previously worked with young people in the US and Palestine. In the MPS office working on the project is Sue Cotterell.

Coming to the Morquio Conference
We have the opportunity to take to this conference a display using some of the photographs and words created by the team to illustrate what it's truly like to live with our impairment. There will also be opportunities for others to share their viewpoint at the event. Afterwards we are exploring producing a DVD and book to tell peoples stories. I aim to use this narrative work within my academic studies and produce a specific academic paper. Our aim is to assure that at all stages of this project that when we use individual's words and images we do so in a way that is comfortable for them and true to the context. **Sarah Long**

Expert Meeting on Morquio Disease (MPS IVA)

Friday 29 - Saturday 30 August 2008

Hilton Hotel, Northampton

NEW SPEAKER ANNOUCMENT FOR MORQUIO CONFERENCE

'Enzyme replacement therapy for Morquio A: Translation to the clinic'
Dr Emil Kakkis, Chief Medical Officer, Biomarin Pharmaceutical Inc.

We are delighted that Dr Emil Kakkis and some of his team are coming to the Morquio Expert Meeting to explain in more detail the Enzyme Replacement Therapy for Morquio A and the upcoming clinical trials.

Please book in early to avoid disappointment!
Visit www.mpssociety.co.uk or phone 0845 389 9901

BIOMARIN™

BioMarin Announces Program for ERT for Treatment of MPS IVA, Morquio A Syndrome

Novato, Calif, June 5, 2008 - BioMarin Pharmaceutical Inc. today announced its program for its third enzyme replacement therapy (ERT) for the treatment of mucopolysaccharidosis IVA (MPS IVA), or Morquio A Syndrome. BioMarin plans to initiate a Phase 1/2 clinical trial in the first quarter of 2009.

'With two MPS drugs on the market, we plan to leverage our clinical, manufacturing and regulatory expertise to efficiently develop a treatment for Morquio patients,' said Emil Kakkis, M.D., Ph.D., Chief Medical Officer of BioMarin. 'Our planned program includes a clinical assessment study to measure the depth and breadth of disease as well as a separate GALNS Phase 1/2 study in Morquio Type A patients.'

'Preliminary studies are promising and indicate that our drug candidate binds naturally to bone matrix and can adequately reach the growth cartilage after IV infusion. The skeletal system disease is a primary concern in the treatment of this disease.'

The company has successfully developed and manufactures two FDA-approved enzyme replacement therapies for the treatment of MPS I and MPS VI. Naglazyme® (galsulfase) for MPS VI is wholly developed and commercialised by BioMarin. Aldurazyme® (laronidase) for MPS I is manufactured by BioMarin and marketed by Genzyme Corporation.

Additional details of the MPS IVA program, along with an overview of BioMarin's product portfolio, advancements in the research and development pipeline and other ongoing programs will be provided today at BioMarin's R&D Day program in New York City. For general inquiries, please email Morquio@bmrn.com.

About MPS IVA

Mucopolysaccharidosis IVA (MPS IVA, also known as Morquio A Syndrome) is a disease characterised by deficient activity of N-acetylgalactosamine-6-sulfatase (GALNS) causing excessive lysosomal storage of keratan sulfate (KS). This excessive storage causes a systemic skeletal dysplasia, short stature, and joint abnormalities, which limit mobility and endurance. Malformation of the thorax impairs respiratory function, and odontoid hypoplasia and ligamentous laxity cause cervical spinal instability and potentially cord compression. Other symptoms may include hearing loss, corneal clouding, and heart valvular disease. Initial symptoms often become evident in the first five years of life. Depending on severity of the disease, age of diagnosis will vary.

The rate of incidence of MPS IVA is as yet unconfirmed, but estimates vary between 1 in 200,000 live births to 1 in 300,000 live births. Approximately 370 patients worldwide are currently registered in The International Morquio Organisation (IMO) survey. An estimated 1,100 patients in the developed world have MPS VI and an estimated 3,000 patients in the developed world have MPS I.

Genzyme 5th International Symposium on Lysosomal Storage Disorders

Christine and I were very fortunate to have been invited to attend the Fifth Symposium on Lysosomal Storage Disorders in Paris, 10 - 12 April 2008, hosted by Genzyme. The program looked a full and interesting one, and we were both looking forward to hearing the speakers and their presentations.

The presentations and speakers were informative and interesting and reflected in the packed conference room. The conference covered all aspects of MPS and related diseases, treatments and future hopes for treatments.

All in all it was a great opportunity for all who attended to meet professionals who we come into contact with.

I would like to thank Genzyme for inviting Christine and myself to such a well organised and informative conference. **Neisha Hall, Advocacy Support Officer, n.hall@mpssociety.co.uk**

Shire 8th International Symposium on Lysosomal Storage Disorders

Christine and I found ourselves returning to Paris after kindly being invited to the 8th International Symposium on Lysosomal Storage Disorders, 18-19 April 2008, hosted by Shire.

The program had been sent to us prior to our departure and I was looking forward to hearing many of the speakers. It was a full program and I was personally interested in the speakers who were going to be talking about the current state of the management of LSD's, and the Novel TTX and future perspectives which included pharmacological chaperones as potential therapeutics for conformational disease, oral therapies and the MPS III dog model.

It was a very informative conference and it was lovely to be able to catch up with many of the professionals I speak to and work with.

I would like to thank Shire for inviting Christine and myself to such a well organised and well attended conference. **Neisha Hall, Advocacy Support Officer, n.hall@mpssociety.co.uk**

ITEMS AVAILABLE!

We have some items which are looking for a new home. If anyone would like to use the following please let the Society know. All the items are in full working order and in good condition:

Small Ferguson 12" portable colour TV and remote

Panasonic portable CD/Radio/Cassette player (quite large model) with remote

Epson desk top scanner

Small Royale vacuum cleaner

Office chair with arms
(a little worn, but OK)

1 x floor standard lamp in curvy black metal

1 x table lamp to match above - both working but need shades

If you are interested in any of these items please phone **0845 389 9901** or email **mps@mpssociety.co.uk**

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

New 0845 number for families

Families caring for sons/daughters with severe learning disabilities can now receive individual telephone support around understanding and managing challenging behaviour for the cost of a local call.

Some children (and adults) with severe learning disabilities typically display behaviour which may put themselves or others at risk, or which may prevent the use of community facilities or an ordinary home life. This behaviour may be in the form of aggression, self injury, stereotyped behaviour or disruptive and destructive behaviours.

Whilst anyone may at times display challenging behaviour, the work of the Challenging Behaviour Foundation is aimed at helping those with severe learning disabilities. Severe learning disability (sometimes referred to as severe intellectual disability or severe mental handicap) is a developmental disability and refers to individuals who have either no speech or limited communication and require support with daily living skills such as dressing and eating.

Family carers experiencing these issues can now access individual information and support from the Challenging Behaviour Foundation Family Support Worker on 0845 602 7885.

Originally started by a parent to provide help and information to other parents, the Challenging Behaviour Foundation's expertise is now recognized by a growing number of learning disability professionals. Email support is also available, and families and professionals are invited to contact **info@thecbf.org.uk** to find out more about the information, resources and support available from the Challenging Behaviour Foundation, which specialises in challenging behaviour associated with severe learning disabilities. All information, resources and support free to family carers.

For more information contact:
The Challenging Behaviour Foundation, The Old Courthouse, New Road Avenue, Chatham, Kent ME4 6BE, email info@thecbf.org.uk or telephone 01634 838739 (general enquiries) 0845 602 7885 (Family Support Worker)

The Challenging Behaviour Foundation

Email: info@thecbf.org.uk

www.challengingbehaviour.org.uk

General Enquiries: Tel. 01634 838739

Family Support Worker: Tel. 0845 602 7885

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

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

Lone Parents of Disabled Children and Signing On

You may have seen recent publicity about lone parents made to sign on at the Job Centre once their youngest child is 12 years old.

This proposal is due to come into force from October 2008. The matter has now been clarified and, if you are a lone parent with a disabled child on middle or high rate Disability Living Allowance (DLA) care component, you will be exempt from the proposals and will not have to sign on.

I thought other members of the MPS Society might find the following information useful:

If you are in receipt of Disabled Living Allowance then you can apply for a Cinema Exhibitors Association Card which allows you to take a carer for free. There is an administration fee but since the card is valid for three years it soon pays for itself. For an application form or further information visit **www.ceacard.co.uk.**

Also, our local Corn Exchange and theatre both allow a carer to go free to concerts and shows etc. Worth asking at yours if they do a disabled concession when you book. These are just a couple of things that I have stumbled across. Perhaps other members have got some other handy tips they can share?

Sue Jenkins (Fabry)

INFORMATION EXCHANGE

HM The Queen presents disabled drivers with keys to driving freedom as Motability celebrates 30 years on the road

Motability
The leading car scheme for disabled people

Friday 13th June - Motability, the UK's leading car scheme for disabled people, celebrates 30 years of providing independent mobility to disabled people with a presentation at Royal Hospital Chelsea led by Motability's Chief Patron, Her Majesty the Queen.

The event celebrates the Motability Scheme's success since 1978 in providing over two million cars, scooters and powered wheelchairs to disabled people and their families through a unique partnership between the charitable sector, Government, leading banks, motor and insurance industries. A range of vintage vehicles will be on display, reflecting the limited choice of vehicles available to disabled people before the introduction of the Motability Scheme. These include a motorised bath-chair (developed for disabled servicemen from the First World War), an Argson trike and an example of a blue trike, introduced by the National Health Service in 1948.

Lord Sterling comments: "We are delighted that Motability's Chief Patron, Her Majesty the Queen and} His Royal Highness the Duke of Edinburgh, have joined us for our 30th anniversary of providing disabled people with access to motoring and mobility. Not only has Motability brought greater freedom to disabled people in the running of their everyday lives, it has also brought benefits to their families. I am personally very glad to use this occasion to honour those who need help with disabilities sustained whilst serving in the Armed Forces and the Emergency Services."

"We are proud of the progress this unique Scheme has made over three decades. Getting on for half a million disabled people and their families now enjoy the choice of more than 3,000 vehicles from major manufacturers, and a worry-free package which includes insurance, road tax, servicing and breakdown cover. This has been accomplished over the years by the work of thousands of unsung heroes, a number of whom are present here today. As the late Lord Goodman often said: 'Motability is a unique partnership between the Government and the private sector'."

Commemorating this milestone on the Scheme's journey, HM The Queen, accompanied by HRH The Duke of Edinburgh, will present six Motability customers with keys to the cars they have chosen. The event will mark the Motability Scheme's role in supporting people with disabilities and especially on this occasion, those who have become disabled whilst serving in the Armed Forces and Emergency Services. Captain Peter Norton, who was awarded the George Cross for bravery, when heading the Army bomb disposal unit will receive the keys to his car from The Queen. Two other ex-service personnel and a former member of the Fire Brigade will also receive their keys together with a 17-year-old motor sports racer who has recently joined the Scheme, and a 61-year-old retired psychiatric nurse, who has been a Motability customer since its inception.

The guests attending the presentation include two of Motability's founding patrons, Baroness Thatcher and Lord Jenkin of Roding, as well as Lord Alf Morris of Manchester AO QSO, the first Minister for Disabled People. Anne McGuire MP, the current Minister for Disabled People, will be present as will General Sir Richard Dannatt and other Chiefs of Staff. They will be welcomed by the Chairman of the Scheme, Lord Sterling GCVO CBE, who was co-founder with the late Lord Goodman, and General The Lord Michael Walker GCB CMG CBE DL, Governor of the Royal Hospital.

The Motability Scheme is the UK's leading car scheme for disabled people. It provides affordable, convenient, trouble-free motoring to some 480,000 disabled customers and their families. Powered wheelchairs and scooters can also be financed using the Motability Scheme.

The Scheme is available to anyone who is receiving the Higher Rate Mobility Component of the Disability Living Allowance or the War Pensioners' Mobility Supplement. Customers simply transfer their mobility allowance to Motability to lease or purchase a car, powered wheelchair or scooter.

For those customers choosing a car with no advance payment, through the Contract Hire Scheme, their mobility allowance will cover the total cost of the vehicle, including insurance, tyre and windscreen replacement, servicing, breakdown cover and road tax. Over 300 models of car are currently available to lease with no advance payment.

As part of the Contract Hire package, a range of popular adaptations are available at no additional cost, such as push-pull hand controls and left-foot accelerators.

The Scheme is directed and overseen by Motability, a national charity, which also provides financial assistance to customers, who would otherwise be unable to afford the mobility solution they need.

More information about the Motability Scheme can be obtained by telephoning **0845 456 4566** or by visiting **www.motability.co.uk**.

Patient Engagement takes off in Scotland

I would like to take this opportunity to introduce myself as a new member of the Genetic Interest Group, an alliance of charities that support people affected by genetic conditions. My name is Claire Cotterill and I will be working as the Patient Engagement Project Officer for Scotland.

To tell you a little more about myself, I have had an interest in genetic disorders from a young age as both my sister and I have alpha-1 anti-trypsin deficiency. Before joining GIG I worked as a research scientist looking at the biology of Multiple Sclerosis. In recent years I have moved into patient advocacy working to support people with mental health problems to get their views and experiences across in challenging situations.

What is Patient Engagement?
Patient engagement, also known as patient/service user involvement, is simply a commitment by organisations such as the NHS, to talk to patients, listen to their views and use this information to improve patient experience in general. The Scottish Government developed a policy called Patient Focus Public Involvement in 2000. This policy stated that the NHS would strive to become more 'patient-focused' and identified the following key aims:

1. A service where people are respected, treated as individuals and involved in their own care.

2. A service where individuals, groups and communities are involved in improving the quality of care, in influencing priorities and in planning services.

3. A service designed for and involving users.

Although these principles are not new, my post was created by the Scottish Government, as part of the Calman Review of Genetics in Relation to Healthcare in Scotland, to breathe life into these commitments.

What will we be doing?
All patient engagement projects will be organised and run as a form of collective advocacy. This means that the issues we try to address with decision makers, the health service and the media are determined by patients and represent a group view rather than an individual one. Some initial suggestions have been:

Creating a 'virtual panel' of patient representatives
A virtual panel is made up of individuals who have agreed to being contacted by email to share their views on future health related issues or changes in policy and practice.

Creating a UK wide online forum
The forum will function as a hub for discussion around a variety of issues that impact on people living with genetic conditions. It is also hoped that it will link up people affected by the same or similar genetic conditions across the UK, and facilitate the sharing of useful information.

Why should you get involved?
Patient engagement is the way forward if you want to influence and mould the kind of health service you receive and tell the Government what benefits and agendas really matter to you.

For example:
Do you want better support networks locally?
Would you like to meet scientists and researchers and question them about treatments and developments?
Would you like to lobby the Government to improve the rights of carers?

Would you like to share your expertise in managing your own condition with other patients or health care professionals?

Finally, Scotland is unique in its vast rural geography and for some communities, remoteness. Getting involved can reduce the sense of isolation that a lot of families with rare conditions experience. GIG will provide support and training to help you get the most out of being involved and the experience gained as a patient representative can be useful experience for future jobs and career moves.

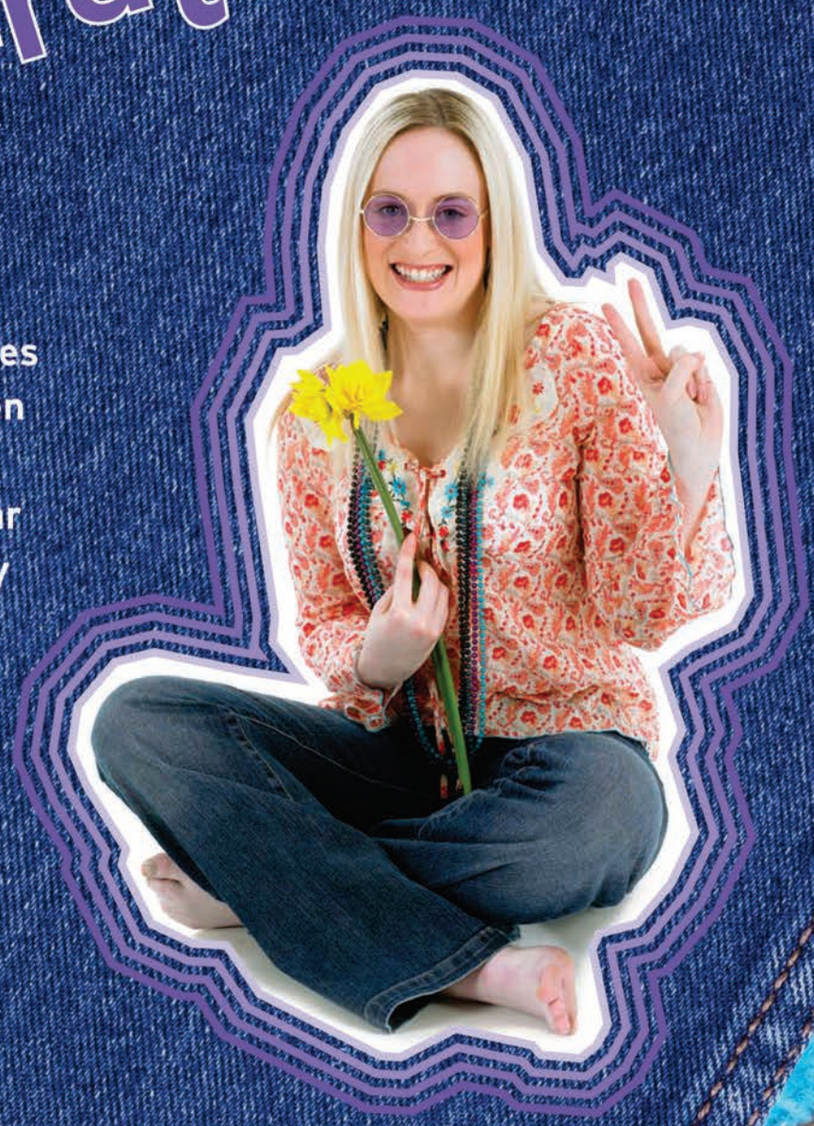
Please feel free to contact me by email: claire@gig.org.uk, phone 0131 651 4805 or 07508 503100, or write to me at GIG in Scotland, University of Edinburgh, St John's Land, Holyrood Road, Edinburgh, EH8 8AQ. For more information about patient engagement in the rest of the UK contact 0207 704 3141.

Changing the world for children with genetic disorders

Jeans for Genes Day

Friday 3rd October

Talkin' 'bout my Gene-ration



Register for Jeans for Genes Day 2008 and help children across the UK affected by genetic disorders. This year it's all about raising money 'Through the Generations' and we'll give you loads of fundraising ideas inspired by everything from punk rock to flower power!

Register for your fundraising pack today

www.jeansforgenes.com

0800 980 4800

Jeans for Genes Reg. Charity No. 1062206

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