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MPS

Care Today, Hope Tomorrow

Spring 2008

Society for Mucopolysaccharide Diseases

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

What is the Society for Mucopolysaccharide Diseases?

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.

What are the aims of the MPS Society?

To act as a support network for those affected by MPS and Related Diseases

To bring about more public awareness of MPS and Related Diseases

To promote and support research into MPS and Related Diseases

How does the Society achieve these aims?

Advocacy Support
Provides help to individuals and families with disability benefits, housing and home adaptations, special educational needs, respite care, specialist equipment and palliative care plans

Telephone Helpline
Includes out of hours listening service

MPS Befriending Network
Puts individuals suffering from MPS and their families in touch with each other

Support to Individuals with MPS
Empowers individuals to gain independent living skills, healthcare support, further education, mobility and accessing their local community

Regional Clinics, Information Days & Conferences
Facilitates eleven regional MPS clinics throughout the UK and information days and conferences in Scotland and Northern Ireland

National & International Conferences
Holds annual conferences and offers individuals and families the opportunity to learn from professionals and each other

Sibling Workshops
Organises specialist activities for siblings who live with or have lived with a brother or sister suffering from an MPS or Related Disease

Information Resources
Publishes specialist disease booklets and other resources

Quarterly Magazine
Imparts information on disease management, research and members' news

Bereavement Support
Supports individual families bereaved through MPS and the opportunity to plant a tree in the Childhood Wood

Research & Treatment
Funds research that may lead to therapy and treatment for MPS and Related Diseases as well as furthering clinical management for affected children and adults

CARE TODAY HOPE, TOMORROW

Cover photograph: Bethany Allen (MPS III)

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FSC

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Newsletter Deadlines

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

Become a Friend of MPS

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CHIEF EXECUTIVE'S REPORT

I hope you'll allow me on this occasion to let me use the Chief Executive's Report to address you from a personal perspective. I want first and foremost to thank every one of you, MPS families, the MPS Societies, doctors, nurses, scientists, volunteers and the pharma industry around the world who have taken the time to offer their condolences in so many ways following the sudden and unexpected death in January of my husband, Robin.

Many of you knew Robin personally and have your own personal recollections of time spent with him. As a founding Trustee of the MPS Society, Robin's sense of responsibility towards the work of the Society never diminished over the past 25 years. Commitment to the welfare of children and young adults with MPS, their brothers and sisters as well as the parents and partners was in Robin's mind the driving purpose for the MPS Society's existence.

Over the past weeks, so many of you have shared your own special memories of Robin. These have meant a lot to our family. From the bottom of our hearts I, and our children, Andrew, Benjamin and Lucy, thank you for adding to our own memories of a remarkable husband of thirty-seven years and a wonderful father and grandfather.

Robin would feel very humbled but proud to know that over £7,000 was received in donations in his memory by the Society. Robin took a keen interest in the development of the MPS newsletter into the professional and patient-orientated MPS Magazine of today received by over 1000 MPS families and friends of MPS as well as doctors and scientists. It has therefore been agreed that these donations will fund the production and distribution of this and the next two issues of the MPS Magazine.

Finally, I would like to thank the entire MPS staff team and the Society's Board of Trustees for their continued support as I make the considerable adjustment of learning to live without Robin.

Christine Lavery
Chief Executive

Robin Lavery OBE
6 March 1944 - 9 January 2008

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News 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 8-9 February 2008.

Governance

The Trustees considered the draft accounts for the year ending October 2007 and these were approved unanimously. The Chief Executive outlined to Trustees the position in relation to the income and expenditure budget for the first four months of the financial year. Trustees were advised that new money had been secured for the Northern Ireland Fabry workshop and Northern Ireland MPS conference, the Scottish conference and the International Symposium in Vancouver, Canada. The budget figures for Alton Towers Weekend were approved.

Risk Management and Health and Safety

Prior to the Trustees meeting, the Chief Executive and the HR & Information Officer undertook a detailed review of the Risk Register and prepared recommendations for Trustees. Significant discussion took place and all but four recommendations were agreed. The Chief Executive was asked to incorporate the Trustees recommendations into these four risk areas. The Trustees received an update on all the Society's health and safety procedures and were advised of the Society's fire risk assessment, health and safety risk assessment, and fire emergency plan.

Strategic Plan

A review of the 2006-2007 second year key priorities and the draft 2007-2008 third year priorities were discussed. The third year key priorities were agreed unanimously and are published in this MPS magazine.

Personnel

Trustees were advised that the Chief Executive's appraisal had taken place and that all staff appraisals would be completed by the end of February. As had been previously agreed, the Society will be adopting a new appraisal system for 2009 following consultation with the staff.

Advocacy Support

Since 1 January 2008, the team have undergone training in Disability Living Allowance and pharmacological chaperone treatment for Fabry disease. Trustees were advised that training on genetic testing and new born screening and paediatric Fabry disease will teak place in March. A site visit to the Willink Genetics laboratory and Royal Manchester Children's Hospital is also planned.

The CEO updated Trustees on Enzyme Replacement Therapy. The CEO confirmed that following the support of the MPS Society a young baby with MPS II in Grampian NHS Trust has been awarded ERT on appeal. The Society is continuing to support the parents of two MPS II boys and two adults with MPS I and MPS II respectively that are fighting for ERT in Scotland. In Wales the Society is supporting the parents of an MPS II boy to achieve ERT.

Trustees were updated on the situation regarding Metabolic Networks and it was agreed that the Society working with the other patient organisations for lysosomal storage diseases should make representations to the Department of Health.

Policies

The Society's updated Reserves Policy was considered and it was agreed that the formula for arriving at the reserves sum should be appended. The policy was agreed subject to the changes recommended.

MPS Research Grants

The CEO, on behalf of Bryan Winchester, presented an update on each of the five major MPS research projects funded by the MPS Society. A small grant application from Dr Maria McCarry, Community Paediatrician at the University of Glasgow was considered for screening for MPS diseases in Scotland. A grant of £2000 was awarded subject to the Society receiving a copy of the LREC (ethics application) and subsequent approval notice.

Farewell to Steve...

Sadly it is time for me to move on from the MPS Society. I have enjoyed my time working for the Society and would like to say a fond farewell to all the families that I have worked alongside during my time here.

I have certainly learned much from the challenges that we have faced together and have great memories from the events that I have attended. I definitely intend to keep in touch with the team and you may even see me from time to time at some of the family days. I wish everyone well for the future, it has been a privilege to work with you.
Steve Cotterell

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New Members

Kristi Lyon has recently been in contact with the Society. Kristi has a diagnosis of Fabry Disease. Kristi is 32 years old. Her two children, Declan aged 11 years, and Kaiden aged 18 months, have also been diagnosed. The family live in East Anglia.

Ms Nowland has recently been in contact with the Society. Dominic has a diagnosis of Morquio Disease. Dominic is 5 years old and the family live in the North of England.

Mrs Blignaut has recently been in contact with the Society. Luke has a diagnosis of Hurler Disease. Luke is 2 years old and the family live in Yorkshire.

If you would like help, guidance or information from the MPS Society's advocacy team please do call us on 0845 389 9901

Deaths

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

Miles Bradborne who suffered from Hurler Disease and who died on 11 December 2007 aged 4 years.

Bridget McDonagh who suffered from ML II, I-Cell Disease and who died on 19 December 2007 aged 3 years.

Luke Chappell who suffered from Sanfilippo Disease and who died on 21 December 2007 aged 18 years.

Aaron Craig who suffered from Sanfilippo Disease and who died on 29 January 2008 aged 14 years.

Aayan Hussain who suffered from ML II, I-Cell Disease and who died on 5 February 2008 aged 9 months.

Births

Congratulations to David and Jo on the safe arrival of Poppy Gosling, born 27 December 2007. A sister for Ollie and Sophie.

Introducing Linda...

My name is Linda Warner and I am a new member of the team at the MPS Society. I started work on 4th February 2008 as an Advocacy Officer within the Advocacy Support Team. I am really enjoying my new role and felt at home within the team straight away.

My career history has mainly been within the public sector; National Probation Service and NHS, however, I did spend five years working for a large pharmaceutical company.

I am married with two grown up children, a King Charles Cavalier dog, and a pair of Cockatiel birds we have had for 18 months but still remain unnamed!

In my spare time I enjoy reading and gardening and watching Watford Football Club. I also work as a Voluntary Counsellor for the South West Hertfordshire Bereavement Service and Peace Hospice.

During my first few weeks, I have been fortunate enough to meet some of you already, but in the meantime if you would like to contact me please do so, either by phone or email: l.warner@mpssociety.co.uk

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EVENTS CALENDAR

2008

CONFERENCE EVENTS

Wednesday 28 May	Northern Ireland Fabry Workshop
Thursday 29 May	Northern Ireland Conference
Friday 13 June	Scottish Conference
Tuesday 24 – Monday 30 June	10th International Symposium Vancouver, Canada
Friday 29 – Saturday 30 August	Morquio Conference

CLINICS

Friday 11 April	Manchester BMT Clinic
Friday 18 April	Manchester BMT Clinic
Thursday 15 May	Great Ormond Street MPS III Clinic
Friday 30 May	Northern Ireland MPS Clinic
Friday 13 June	Birmingham MPS Clinic
Tuesday 1 July	Bristol MPS Clinic
Friday 4 July	Manchester BMT Clinic
Friday 11 July	Manchester BMT Clinic
Tuesday 5 August	Bristol MPS Clinic
Friday 10 August	Manchester BMT Clinic
Friday 17 October	Manchester BMT Clinic
Tuesday 4 November	Bristol MPS Clinic
Friday 21 November	Birmingham MPS Clinic

REGIONAL EVENTS

Saturday 3 – Sunday 4 May	Alton Towers Family Weekend & AGM
Wednesday 14 May	Reception at the Palace of Westminster
Thursday 15 May	MPS Awareness Day
Saturday 14 June	Ollie G Ball
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
Saturday 29 – Sunday 30 Nov	MPS Adult Weekend

*Please note that the correct date for the Childhood Wood Remembrance Day is Sunday 13 July 2008. There was an error on the programme that was sent out with the last MPS Magazine in January.

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MPS CLINICS

Newcastle MPS Clinic

After a restful night sleep, following a four hour drive to Newcastle I was ready for the Newcastle MPS Clinic on 24 January 2008 - the thing was, were the staff of the Royal Victoria Infirmary (the RVI) and the members attending the clinic ready for me?!

The clinic ran to time and went very smoothly, and it was lovely to meet up with familiar families and to put a face to a voice in some cases. I was very grateful to have a separate consulting room, which gave families and individuals the opportunity, if they wished, to discuss any issues they had.

I would like to thank Dr George Rylance and Dr Ed Wraith for their continued support at this clinic, without which we would not be able to continue with the Newcastle MPS Clinic.

I would also like to thank the staff at the RVI who accommodate us so well, and on a personal note I would like to wish Trish Martin every success - you will be missed. **Neisha Hall**

Manchester BMT Clinic

I had received the clinic list prior to travelling to Manchester for the BMT Clinic for 25 January 2008, so I was prepared for a very busy day!

Jean had done a fantastic job in getting refreshments for the children, which helped enormously - as tempting as the box of goodies was, I refrained from raiding it!

The waiting room at times, was a hive of activity, but the children behaved perfectly and waited very patiently. It was lovely to see all the children and their parents again - the cuddles I received from a certain young man (yes, that's you Aiden) were very welcome and helped me through the busy day!

I would like to thank Dr Ed Wraith for his continued help and support with this clinic, for Jean Mercer for just about EVERYTHING, and to Dot who kept me sane during such a busy day. **Neisha Hall**

Photos clockwise from top right: Oliver Gosling (MPS I BMT), James Fair (MPS II), Luke Chapman (MPS III) and his brother, Jeanette and Andrew Whiteside (ML III), Callum Pollock and Jordan Mount (both MPS I BMT), Matthew Ingram (MPS I BMT)

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Dartford Family Day

As we hadn't seen many MPS families since the conference in June we were really pleased to be having a get together as that is the only time we feel anything like a 'normal' family. We are the Gremo family and we have Nathan who has Sanfilippo and is 16 years old.

We set off in plenty of time to reach Dartford knowing that the motorway was closed for the day but as it turned out, not soon enough. When we hit the crawling traffic we still had 10 miles to go! We were nevertheless not put off and stuck with it. We were only an hour late. We did manage to arrive at the same time as the food which was lucky!

From the minute we arrived we thoroughly enjoyed ourselves. After being greeted by Chris Murphy (MPS Advocacy Officer) we sat down to a delicious buffet with a large choice of dishes, and especially tasty chocolate pudding. We met some new families as well as families we already knew and, as usual at MPS gatherings, everyone was friendly and had a story to share.

There was a very patient and entertaining conjuror who appealed to children of all ages. Nathan was so chilled out that he ended up lying spralled out on the floor, which as we all know is not usual for a Sanfilippo person!

I have only one complaint about the day and that is that it ended all too soon. Hopefully we will be able to get together again before too long. Thank you to everyone involved in organising it for us. **Janet Gremo**

Photos clockwise from top right: Willow Reed Barnes (MPS I) and her brother, Toby Brooks (MPS I), Nathan Gremo (MPS III), Kamal Hoteit (MPS IV)

MPS Clinic at Birmingham Children's Hospital - On 22nd February we had a very busy day, meeting many new families as well as some familiar faces. We hope that all families found the day useful and informative. I would like to pass on our thanks to all the doctors and nurses involved for inviting the MPS Society to come along. **Steve Cotterell**

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MPS Awareness Day

The Society is celebrating MPS Awareness Day on 15 May 2008. This is a day devoted to raising awareness for 23 rare, genetic diseases known as Mucopolysaccharide (MPS) & Related Diseases.

During last year's MPS Awareness Day we held a children's party at MPS House. MPS children and their siblings enjoyed playing on the big red play bus!

How do I get involved?

Help us spread the word about MPS and raise funds to provide vital services.

Spreading the word...

Use MPS Awareness Day to tell everyone you know about MPS. If you suffer from MPS why not tell your story, talk about your job, or school, your family life, local issues or campaigns that you may be involved in, pursuits you enjoy or sports you are passionate about.

If you can't support MPS yourself, maybe you can approach your place of work. The Society is eager to explore more ways of working with our supporters in the corporate sector.

Raising funds...

Does your workplace, friends, relatives or neighbours support a chosen charity? Why not tell them about MPS? Or are you organising a fundraising event?

Email fundraising@mpssociety.co.uk to tell us what you are doing and to order your fundraising pack. Tell your supporters where the money is going and what it will support.

Try to get some coverage in your local media by giving them a call or writing them a letter. Local press like to feature inspirational stories so let them know about your event. Make use of your local amenities, for example, pubs, restaurants and shops as they are great places for promoting awareness. Remember to check whether you need permission from anyone to use their venue. Ask small companies to donate gifts as they will benefit from the publicity and supporting worthwhile causes.

Do you have your own place in an event and haven't told us or are you still looking for a charity to support? Set up your own fundraising web page. For collection boxes, stickers, balloons and other support materials please contact us. You can also call us for tips on organising an event or advice on how to publicise it locally. For all activities remember to check the legal insurance requirements and health and safety regulations.

Photos below and top left appear courtesy of The Buckinghamshire Examiner www.buckinghamshireexaminer.co.uk

MPS Awareness Luncheon at Hampden House

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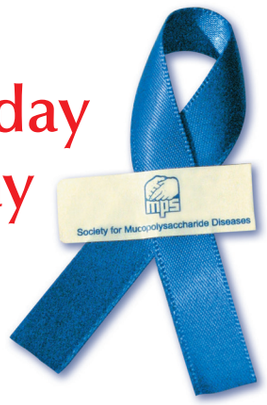
Thursday 15 May 2008



You can help support MPS Awareness Day by ordering some of our great promotional goods:

MPS Trolley Key Ring

Also useful if you're at the gym but don't have a spare £1 for your locker!

MPS Awareness Ribbon

The latest fashion must-have to promote MPS awareness

MPS Dog Tags

A fashionable way of supporting a good cause. Our dog tags feature our web address and slogan Care Today, Hope Tomorrow

MPS Awareness Wrist Band

Show your support to MPS with our trendy awareness band inscribed with web address and slogan

To order your MPS Awareness Day promotional goods please complete the reply slip below and return it to the MPS Society

To make a Donation

☐ I wish to make a general donation of £ .....

Gift Aid

Contributions to charities are eligible for tax relief. This means that if you pay income tax or capital gains tax, and you make a donation during the year, the Society can claim tax on this donation. To make this possible just tick the box and complete this section.

I wish for all contributions I may make to the Society for Mucopolysaccharide Diseases to be treated as Gift Aid donations.

Signature .....

Date ..... Postcode .....

If your circumstances change, please let us know.

To Order Goods

MPS Awareness Ribbon	49p	Qty .....
MPS Trolley Key Ring	£1	Qty .....
MPS Awareness Wrist Band	£2	Qty .....
MPS Dog Tags	£2	Qty .....
	TOTAL	**£.....**

☐ Please send me a free MPS Fundraising Pack

Address and payment details

Mr/Mrs/Miss/Other (please delete as appropriate).....

Name .....

Address .....

.... Postcode .....

☐ I enclose a cheque/postal order made payable to the MPS Society (UK Sterling only)

☐ I wish to pay/donate by (please delete as appropriate)
Mastercard/Visa/Visa Electron/Maestro/Solo

Card No .....

Last three digits on signature strip .....

Valid From ..... / ..... Expiry Date ..... / .....

Issue No ..... Name on Card .....

Numbers in address .....

Numbers in postcode .....

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Ollie G make it Special,

but where was the Special one?

Ben, MPS I, Hurler-Scheie, Sarah, his big sister, Tracey, Ben and Sarah's mam, recently had an amazing experience, to me, Ben and Sarah's dad, I had all my Christmases rolled into one. 27th October 2007 was my Christmas Day this year, my best day since Ben was born, but what happened?

For the record, I slept extremely badly on the Thursday and Friday night... I was just too excited (big kid)!

Well, we had known all about this day for a couple of months but were very cautious about telling Ben as October can sometimes be a problem month for Ben. He was diagnosed in October and had severe problems last year which resulted in him needing a shunt (book to follow). So when we got the letter we decided to not tell Ben until we actually reached our destination.

So, as we approached the day, we had to plan very carefully. We had clothes to buy, hotel to book, drinks to buy and then the cost of petrol, at least £70 return. Then came the next bit of good news, we had been selected to receive a donation from the Ollie G clay pigeon shoot. This would take care of all our hotel, food and petrol and even a little bit left over for Sarah and Ben's special clothes.

Finally the big day arrived. We set off at 8.30 a.m. on 26th October (half-term Friday) to begin the first part of our epic journey. We had arranged to meet relatives at the new Wembley Stadium sometime in the early afternoon. Depending on traffic and fatigue we would let them know when we were close. The journey was slow due to road works but Ben and Sarah were amused by the DVD of Happy Feet and Sarah amused herself by being travel sick, not a past-time I'd recommend. Thanks to the wonderful British motorway system and several cups of coffee, we arrived at Wembley and met up with the relatives.

Tracey had enquired if the FA were offering Stadium tours of Wembley but this had not yet been organised by the FA.

With a donation from the Ollie G clay pigeon shoot we would even have had enough money to buy a drink each at Wembley. Unfortunately we missed out on that privilege. After a brief game of football, which I can claim to be the first Conlin to score a goal at Wembley (but not on the hallowed turf, just outside in the car park), we piled back in the car and headed for Richmond.

At Richmond we stopped for a coffee and another game of football, have you spotted the theme yet?

Then a really fun game of "Hunt for Ben's missing Glasses Lens". This is a great game whereby you find a huge area of green land, i.e. Richmond Green. Ben then proceeds to throw himself to the floor several times for a laugh and to get muddy. You then leave the field in the fading light (dusk) only to discover that Ben has lost a lens from his glasses. After much swearing and agonising, all the participants head back to the large area of grass to hunt for the missing lens. The winner gets a pat on the back.

Tracey was the lucky winner of this particular game and, lens found, we finally headed back to the car and finally the hotel.

The next morning we awoke and after a hurried breakfast set off in the car back to Richmond to catch the tube. We left the tube at Fulham Broadway and walked for about 200 metres, until we saw our destination... STAMFORD BRIDGE, home of Chelsea Football Club, ……oh my goodness.

After a very expensive trip round the Chelsea Mega Store we began to walk round the stadium to the East Reception Area for our 12.15 p.m. appointment (have you guessed yet?).

Ben, being totally unaware, does not want to walk around the stadium, he just wants to play football. Nothing could persuade him to get a move on. Bribery of "if we hurry we might meet John Terry" were ignored, as were the threats of "Get a move on or I will kick you up the bum."

We eventually arrived at the East Reception Area and were introduced to Joan, our helper for the day. She tried to tell us what was going to happen, but I could not hear a word of what she had to say, I was too excited (big kid). It was at this point, with the journey safely completed, that we broke the amazing news to Ben…

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"Ben, you're going to be a Chelsea Mascot for today against Manchester City."
"YES!" said Ben followed by a punch into the air.

We were given an itinerary which I eventually understood and then the short wait for everything to happen. We watched and waited in the reception area as the press and numerous other people walked by. Then I spotted Peter Bonetti (ex-Chelsea goalkeeper) and Ron "Chopper" Harris (Captain of the 1970 FA Cup Final team)... oh my goodness. Then the bad news, being a lady, Tracey would not be allowed into the Chelsea changing room, just Sarah, Ben and I.

Still waiting and then players began to drift by, we saw them all walking within feet of us. Then more waiting.

Eventually, Frank came up and asked if we were ready. Was I ready? What a ridiculous question. I'd waited 37 years for this. Of course I was ready.

We were led into the inner sanctum at Stamford Bridge, the home changing room… OH MY GOODNESS.

"Help yourself to autographs", said Frank and we did. We could not move fast enough to say "hello" to all the players, we got photographs, autographs and handshakes. I was star struck and hardly able to speak, Sarah was also star struck. Ben took it all in his stride as if it was an everyday occurrence.

Sadly, the experience was over much too soon and we were ushered back to the reception area and back to Tracey to show off our much prized possessions. [continued over]

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After a brief chat we were then taken to the WAGS (wives and girlfriends) area of Stamford Bridge to enjoy a spot of lunch. On the way we met Neil Barnett (Chelsea TV, who suggested a possible interview for Sarah and Ben for Chelsea TV - unfortunately this never materialised).

I introduced myself to Roy Bentley - Chelsea Captain in 1955 when we won the first division for the one and only time, and Bobby Tambling, all time top scorer for Chelsea. We also saw Frank Lampard's girlfriend and gran and Joe Cole's mother. "This is the life", I thought, and "I could get used to this type of environment".

Then the big moment. 2.40 p.m. Time for Sarah and Ben to get changed into their Chelsea Football strips and finally be escorted round the pitch to the tunnel.

Tracey and I took our places pitch-side and I got my video camera ready. Sarah and Ben were taken into the tunnel, the last we were to see of them for 5 minutes.

Then the familiar music started, the flags began to wave and Neil Barnett, Chelsea TV, began to read out the names of the teams to much booing (Manchester City) and much cheering (Chelsea).

Video rolling, the first people out of the tunnel were Mike Riley (referee) and his two assistants followed by the Manchester City Captain and finally Frank Lampard holding the hands of two very excited children, Sarah and Ben. Ben has MPS and obviously had trouble with the stairs onto the pitch so Frank Lampard had to lift him up the stairs onto the pitch, (the lucky little boy). We saw Sarah and Ben march across the pitch to line up with the teams.

I stood in awe as I watched my two children, dressed in Chelsea kit running onto the pitch at Stamford Bridge. The two teams then lined up. As is the custom nowadays, the whole Manchester City team shook hands with the whole of the Chelsea team which on this occasion included Sarah and Ben.

Chelsea FC player Branislav Ivanovic, Sarah and Ben Conlin

The two teams did a final warm up and Ben thought he had to leave the pitch, I watched helplessly as Ben dragged Sarah before the Matthew Harding stand.

Luckily, Sarah thought quickly before Ben could get too far and she brought Ben back before Frank Lampard stepped in and guided Sarah and Ben to the centre-circle and their meeting with Mike Riley (referee). Mike bent and asked Ben his name and which position he played when he played football.

"Defender", replied Ben.
"Do you know what I do to defenders?", asked Mike.
"No", answered Ben.
Mike produced his yellow card from his top pocket and said,
"I book them."

Another quick photograph in the centre before Frank Lampard asked Ben and Sarah to leave the pitch.

We were then escorted to our seats behind Sven Goran Ericson and the Manchester City staff and substitutes.

What made it all the more enjoyable was that Chelsea won 6-0, their biggest score in years. It was obviously down to the good luck that Sarah and Ben brought.
At the end of the match we waited for ages before leaving, the day had been so special I did not want it to end, but it had to and we slowly made our way out of the Stadium.

A quick tube ride and then the stressful car journey home, all six hours of it. We eventually arrived at 1.00 a.m. and emptied the car before collapsing into bed.

The next morning we woke up, watched "Match of the Day" on BBC1 and Football First on Sky to catch any pictures of Sarah and Ben we could, then we watched our own video of Sarah and Ben coming onto the pitch. It was like a dream, but it really happened.

To this point we have watched the video about 20 times, we have shown it to every relative we can grab hold of and also Ben's class have watched it several times as we lent them the video.

27th October was a truly special day and we would like to thank the following people and organisations: Chelsea Football Club, Joan at Chelsea for being so helpful and looking after us so well, the Chelsea players for being so friendly and for winning 6-0, Ollie G Events for helping with the expenses.

Peter Conlin
Ben's and Sarah's dad
37 Years a Chelsea Fan

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Our visit to 'Highgrove'

The Prince of Wales and the Duchess of Cornwall invited 10 children from Ty Hafan Children's Hospice to their home, Highgrove House in Gloucestershire, on 20th December '07. The Prince is Patron of the hospice and the couple provided their guests with food, drinks, a magician and carols played by the royal Harpist.

Georgia (MPS III, Sanfilippo disease) was invited, so I thought I'd share the experience with you…

Georgia came out of hospital on Wednesday 19 December, so I wasn't too sure how our visit to 'Highgrove' on the 20th would work out. She slept well on the Wednesday night, so on Thursday morning, I decided to go for it and we all got ready, in our best clothes!

It was a beautiful day, a little cold, but the sun was shining. It took only 45 minutes to get there. Highgrove is a stunning place and on our arrival at the gates, our car was checked over by the police and our invitations were also checked, to make sure they were valid! We were greeted by staff and offered wine, coffee or soft drinks. I had the wine!

The room we were in was beautiful, a huge Christmas tree and grand piano. Ten children and their families were invited from Ty Hafan Children's Hospice, and also some of the staff. Everyone looked lovely and even though the dress code was formal, the whole atmosphere was very relaxed and informal. There were lots of photographers and film crew there.

We sat in a sunny spot by the window, as Georgia was still quite weak and not 100%. Food and canapés were being passed round. Then, Charles and Camillla came in; they were filmed chatting to some of the families on the other side of the room.

We stayed sitting and eventually Camilla came over to us. She started talking to Georgia, who was still sitting and I explained that Georgia would look at her if she was on her level, so Camilla got down on her knees! I never expected that! It was like she was bowing down to Georgia! Of course, we've always said that Georgia is a very special princess!

Camilla was genuinely warm and friendly, talking to Georgia, face to face, on her level. Georgia responded beautifully, 'talking' back to her and smiling.

Then, Charles made his way over to us, he pulled up a chair and had a good old chat! He and Jon had a lot in common - horses and farming! I was amazed how down to earth he was, talking about William and Harry like we'd been friends for years. He talked to Georgia too, she must've found him quite funny, as she was laughing.

Each family was given a 'goody' bag, from the Highgrove shop, some lovely items, worth quite a bit of money, and a bottle of champagne included.

All in all, we had a lovely day and it really was a pleasure to meet Charles and Camilla. As you can see from the photo, Georgia was the perfect princess, incredibly poised and on her best behaviour!
Louise Lewis

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Battling for Bethany

This is a message of hope to all the families that are trying to obtain a buggy or a wheelchair for their child via a not so helpful wheelchair services. I am sure that there are many of you who can relate to this problem.

We have found a solution thanks to advice from Neisha, our MPS Advocacy Officer and Whizzkidz. Now I try to do battle with everyone with help from the MPS Society for any equipment needed for my daughter Bethany, for example, social services, wheelchair services and the local council, depending on what you are fighting to obtain.

Now I have heard some of you have good services. Well, you are the lottery winners because where I live we don't have such a good service, and I believe they are using the good nature of other charities as a means for you to get the equipment you need but so they don't have to pay for it.

That's another fight I am going to take on soon. However, for the buggy we wanted for Bethany and the lack of help from wheelchair services we had no choice and Neisha put us in touch with Whizzkidz. We completed the application form and were then told there was a lengthy waiting list. Neisha then rang them and soon after we were given an appointment to see some buggies.

I knew in my own mind what buggy I wanted for Bethany and I said to my wife that as we had come this far, we shouldn't settle for anything else. The representative came out and asked if we had anything particular in mind and I replied that we would like what we had put on the application form. Unfortunately, he said they hadn't got that one. He led us into a room full of buggies and chairs and there was one similar to the one I had requested but it was more expensive and had better features.

From the moment we entered that room they were fantastic. Their Occupational Therapist knew about Sanfilippo Disease whereas our own Occupational Therapist knows nothing. I asked if there was a tray we could buy. He told us not to worry and that they would add one to the order. We came out of that room feeling so up beat and positive from the help offered. They said they would be in touch in about twelve months time to re-assess Bethany and change the chair if need be. All we had to do was insure the buggy against damage and breakdown which wasn't expensive, and they supplied a list of insurance companies to ring. Their attitude was so different to wheelchair services. Wheelchair services are supposed to help and guide you but all you do is battle. Up the revolution for the disabled! Thank you to Whizzkidz and the MPS Society. **The Allen Family**

Do you need support from the MPS Advocacy Team?

Please remember that should you wish to speak with a member of the advocacy team do not hesitate to pick up the phone or email if you find it easier. Please bear in mind that at the moment we are a small team covering the entire UK, however we will always return calls and respond to messages as quickly as possible.

advocacy@mpssociety.co.uk
or **0845 389 9901**

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Me and My Hobby

Hello, my name is Jasmine, I am fourteen years of age and I am going to tell you about my hobby and why it is so important to me. I have a sister Bethany (pictured left) who is 10 years old and she has Sanfilippo disease, and a brother Tom, age 6, who isn't affected.

I go dancing and I have done this ever since I was four. I have been to three different dance schools (one being a stage school). The types of dancing I do are: Modern, Acro, Lyrical, Tap, Ballet and Song and dance. I dance three times a week (though this varies when I have festivals and shows coming up). We have two festivals every year in May and September. I do feel like I am stopping my family from doing things sometimes such as going on holidays. But they do support me throughout it all and I really do appreciate it! My mum sews the sequins on all my costumes and it takes her forever. She doesn't have to do it but she does, so thanks mum! On a Saturday I go up and help teach the younger ones from 10am - 2pm. When this opportunity came up I was thrilled!

I really do enjoy it. My friends say things to me like "We're going downtown, swimming or to the cinema. Do you want to come? I'm sure your dance teacher wouldn't mind you missing it this once". Every time I give them the same answer "No!". My teacher obviously chose me for a reason and I don't want to let her down. This is like a proper job and in a couple of years time when I am working this will have a good impact on me. Every year we have a presentation and since I have been at this dance school I have won a trophy every year.

I won the 'happy trophy' when I was younger for always smiling, the 'most improved' trophy twice, 'most promising' twice and the 'best in lyrical'. At my last festival I also came second in the championships with my acro duet (we had won it in the acro section to get into the championships). This was a big surprise, our first time doing it and out of the whole duet/trio section there was a good 24 of us!

As well as enabling me to make new friends, Mum, Dad, Tom and even Beth have too, which I think is great! We're now all really close and do things together outside of dancing too. We're all like one big family! Hopefully one day I could make something out of this.

I have written this to share with you how important it is for me to have a hobby and if I had to give it up for what ever reason I don't know what I would do. It's just great for me to be able to get out of the house for an hour or so a few days a week, and to be doing something I really enjoy. **Jasmine Allen**

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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Princess Royal Attends Carers' Rights Day Conference

Devizes, 7 December 2007

I am a member of Carers Support West Wiltshire. Every year they organise a string of events, including pampering sessions like massage, manicure, a day at a retreat, etc as well as informative meetings. They also mark the Carers' Rights Day with a local event.

Last year they joined forces with other districts of Wiltshire and brought about this exciting conference in Devizes Corn Exchange, where the Princess Royal was the guest speaker.

I was privileged to be invited and to attend. Weeks in advance all our personal details had to be submitted for police checks and we had to bring a separate form of identification as well as the invitation.

After arrival, registration and coffee, everyone was seated for HRH's arrival. House-keeping and Royal Protocol etc (things like if she puts her hand out, then we would shake hands) was explained while we waited.

The Princess is president of her own trust, The Princess Royal Trust, which supports hundreds of carers support organisations all over the country. There was a real buzz in the hall before her arrival and an expectant hush fell around the place when she came in and officially opened the conference entitled *I'm Still Standing*.

She told us that it was a pleasure for her to be able to join us and was delighted to see so many carers able to make it there since she knew how much planning was involved to get out of the house just for a morning!

Her speech was very informed. She talked about respite care and how it was badly understood and its availability being very patchy. She said that many people preferred to be cared for in their own homes and that home care was very difficult to find.

She said: "You live in a largely rural area and that is an even bigger challenge. Many people do not see themselves as carers under a heading of any sort, and that again makes it difficult for us, but we are getting slightly better at responding to the individual needs."

"I hope that the phrase *I'm Still Standing* is a phrase we won't have to hear much more in the future."

The Princess listened to the personal experiences of a carer looking after his wife and Down Syndrome daughter.

The Princess came to each table where we were seated. We all got up. She shook hands with each person and asked about their own circumstances. She came across as being very caring, informal and friendly and she made everyone feel at ease. She also dotted her conversation with her experiences and comments like "I know how older people can be very independent. Could we get my grandmother to have a chair lift!" She left the Conference around 11:30 amidst great applause.

Next were four speakers. Their speeches were very powerful and moving and highlighted the plight of carers. They really got the point across very forcefully just how much carers do!

Young carers ended the morning session with a sketch on what life is like for them and what they wanted to see happen. Overriding message from all Carers was simply; please listen and treat us with the respect we deserve.

We all had a very nice buffet lunch. The afternoon session began with the responses from the statutory agencies to the carers' stories, non-executive Chair of the PCT, and the Director of the Department of Community Services. As we all know, passionate speeches don't necessarily mean action!

Next, we formed working groups according to our relevant positions. (Parent carer, etc.) Each group had at least one representative from the statutory agencies. We were asked to identify what major issues faced us as a carer, what we found most helpful and what was unhelpful.

The discussion groups were very lively and everyone would like to have had more time. The emerging overall issues seemed to be: respite, allowances, and attitudes to carers, lack of information and the endless assessments and paperwork!

A lot of work went into organising the day and it was a huge success. It brought together many carers with varying needs and difficulties and gave them a platform to launch their stories and have their voices heard. I believe it also empowered them. If we unite, then we should be a force to be reckoned with! The dilemma is who's going to look after our charges while we fight and protest with our last drop of energy? **Fer Pidden (mother of Natalie, MPS III. Fer is pictured right with the Princess Royal. Photo appears courtesy of Devizes Gazette and Herald).**

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A Day Fishing With Tom And Ben

Hello! My name is Carl Rogers. I am 17 years old and have Hunter Disease, MPS II. I live at home in Hertfordshire with my mum, dad and two sisters. My mum is called Debbie and my dad is called John, my oldest sister is called Hayley, she is 19. My younger sister is called Lauren and she is 15. We also have a pet dog called Riley. He is a lab retriever who is four years old.

My interests are fishing and golf. I enjoy fishing because it is relaxing and very enjoyable. I like golf (pitch and putt) because it is enjoyable but also challenging.

Normally I go fishing with my dad, who taught me everything I know about catching small fish, so I thought it was time I moved on and caught something bigger. On Sunday 10th February I went fishing with my sister's boyfriend Tom and his friend Ben. We went fishing at a lake in Nazeing (Essex). When we got there I helped Tom carry the fishing equipment to the swim and helped set up ready for a nice day of fishing. At the start of the day it was cold but towards the afternoon it seemed to warm up.

As soon as we were all set up, we cast out our line. The first fish I caught was a 15 ½ pound common carp. I was over the moon as it is the biggest fish I have ever caught! As the fish was so big I had difficulties gripping the fish but Tom and Ben showed me a good way of gripping the fish which made it easy when I had some photos taken.

After about 45 minutes I managed to hook a 12 pound common carp. Yet again I was happy with the fish I had caught but my luck didn't stop there and about half an hour later I caught a four pound common carp. Tom and Ben were very helpful and they were very good teachers. All fish were cared for and returned safely.

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Planners say yes to family's home plea

Jeff and Jo Lloyd, parents of Amy seven and Scott four, who both have Sanfilippo Disease have asked that we share their story with you all. They hope that their 'success', will inspire other families who have battled for so long in a similar situation.

Jeff and Jo's story was featured in the Torbay Herald (7.2.08), and headlines are as follows:

'The parents of two terminally ill children have won their long battle to build a special care unit at their cramped family home'.

Jeff said: "You can't describe the winning feeling - we are so pleased."

Jeff and Jo Lloyd, from Dawlish, desperately needed a unit to care for Amy and Scott, and the alterations will provide bed space and a specially adapted bathroom for their severely disabled children.

Their story began three years ago when Jeff and Jo noticed Amy was struggling with her learning. Although it is just one symptom of Sanfilippo, the Lloyds were unaware such a condition existed. Amy underwent tests at Torbay Hospital and was later diagnosed with Sanfilippo Disease. It came as a huge shock, but there was also a one-in-four chance Scott had the same disease. Following tests, it was confirmed one-year-old Scott also carried the disease.

Jo recalled the huge shock and how their lives were thrown into complete turmoil, she said: "We went into a dream-like state. You don't want to believe it."

Jeff, at the time a self-employed plumber, was forced to give up work to care for Amy and Scott. Now both parents work part-time but are committed to looking after their children.

After investigations into the possibility of moving to a more suitable home failed, Jeff and Jo then became aware of how long things can take.

Jeff said "You do not realise how hard you have to fight… it's a constant battle".

The Lloyd's decided to extend their property and contacted Neil Goldsworthy of First Class Independent Living, which specialises in adapting property for people with limited mobility.

The process then included submitting the planning application to Teignbridge Council's development control committee meeting. Initially planning officers refused because it was 'unsympathetic' in relation to the home, it impacted on neighbouring amenities and the scale was 'inappropriate'. At the meeting, Mr Goldsworthy implored councillors to OK the plan. They unanimously voted in his favour with Dawlish mayor councillor Rosalind Prowse saying: "You can't give back life or health. Think not about planning, but your conscience. Can you really rob these people of what is left of their lives?"

The final outcome was 24 councillors voting in favour of the planning application.

Jo would also like to add, that it has not been easy for the family, but Neil has worked wonders for us. We were so pleased councillors supported us.

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Invitation to take part in the GSCC's survey of people who use domiciliary care services

All 'domiciliary care workers' - those paid to come into people's homes to help with things like washing or dressing - will soon need to register with a government body, the General Social Care Council (GSCC). To help make sure that registration works as well as possible for people who have care services in their home, the GSCC wants to find out their views about what aspects of registration are important and valuable to them.

If you are:
A person who uses domiciliary care services; or
A family member or friend who represents someone who uses domiciliary care services…
… we would like to invite you to take part in the GSCC's survey.

What will the survey involve?
An interviewer will arrange to meet you in your home or other suitable place
They will ask you about your views of registration of those working in homecare
It will last about 15-20 minutes
You will be able to have a friend or family member with you, if you would like to do so
You will receive a £10 gift voucher to thank you for your contribution

All participation will be strictly confidential. Your name will not be linked with any of the information you give, and your details will not be passed on to anyone else.

If you would like to take part in this survey, please let Stephen Hodgkins at the GSCC know on
Email: stephen.hodgkins@gscc.org.uk Tel: 07951050153

The GSCC will place your details on a list and participants will be selected at random and called by a researcher working for the GSCC to find a suitable time for you to take part in the survey.

The GSCC would hope to speak to everybody who agrees to take part but due to the timescales it may not be possible to interview everybody.

Thank you in advance for taking part in this research.

Yours sincerely

Lisa Watch
Head of External Affairs
General Social Care Council, Goldings House, 2 Hay's Lane, London SE1 2HB, www.gscc.org.uk

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.

Solitude

A sense of freedom, of being at one with oneself
A situation of being alone but not lonely
Is a good place to be, a fortunate place to be
Because it means being comfortable in one's own skin
Having a strong sense of self, enjoying one's own company
Solitude gives space, time for reflection
On oneself and others, on what one is doing
And what is to be done, time to catch up
Or time to relax, time for music or just silence
Time to write or paint, doing creative things
Make plans for the future but to enjoy what is here now
Solitude is not hiding from life, not hiding from people.
It is an enjoyment to live life to the full
To be with people, especially family and friends
And solitude is to be savoured.

Shauna Gosling September 2007

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Strategic Plan 2005 - 2010

Policy Priorities and New Initiatives 2007 - 2008

These are the key priorities and new initiatives reflecting the vision and objectives of the third year of the MPS Society's five year strategic plan.

Children and Adults First

Continue to work with key professionals identifying the strengths and weaknesses of diagnostic practices in the UK, particularly in the context of the proposed Network for Metabolic Diseases.

To identify new initiatives to involve the Society's membership in having a voice in identifying and influencing best practice in areas of social care, respite and palliative care and clinical management.

To undertake an evaluation of clinical and homecare services where a child or adult is receiving enzyme replacement therapy for MPS I, MPS II, MPS VI and Fabry disease. Ensure the scope of services offered meets the needs of the stakeholders and upholds the principle of being person-centred.

Whilst continuing to develop and update our range of educational materials and information resources, to initiate as part of the quarterly MPS Magazine, a children's newsletter suited to a range of ages, disabilities and siblings.

Finalise the development of the MPS website ensuring all aspects of the Society's work are fully illustrated and ensure that the content meets the needs of our membership and serves as a communication tool for breaking news.

Advocating for Children and Adults

Continue to increase the number of regional social activities and family events offered to members with particular emphasis on greater involvement of bereaved members.

Continue to offer encouragement and opportunities to adults affected by MPS and related diseases to develop independent living skills and have a voice in the decision-making of the Society.

Develop an MPS interpreting scheme for families where English is not their first language using the skills of volunteers including those within the membership of the Society.

Continue to develop our relationships with the expert regional centres in Scotland, Northern Ireland and Wales providing support at their MPS clinics as appropriate.

Meeting Needs

Continue to promote the Department of Health's guidance on national specialised commissioning for Lysosomal Storage Diseases in England to ensure equitable access to diagnosis, clinical management and therapies are upheld.

Continue to uphold members' rights to equitable access to diagnosis, Clinical Management and therapies in Scotland, Wales and Northern Ireland.

Continue to deliver on equitable UK-wide specialised person-centred individual advocacy service with emphasis on developing the support afforded to children and adults in the palliative care stage of their diseases.

Develop and implement a support strategy for MPS families facing and following bereavement in consultation with the members.

Continue to offer training opportunities for providers of health, educational and social care through MPS information days and regional conferences.

Organise disease-specific and bereaved members exchange summits to inform the Society of the needs and expectations of its members.

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MPS Awareness Day
15 May 2008

To find out ways in which you can help email **fundraising@mpssociety.co.uk** or phone **0845 389 9901**

Making a Difference

To continue to support four innovative research projects with a particular emphasis on the Blood Brain Barrier and the use of stem cells that may lead to the development of improved clinical management and new therapies.

Continue to monitor developments relating to a Network for Metabolic Disease and ensure decisions made reflect the needs of the Lysosomal Storage Disease patients.

Using new software, update the MPS Registry allowing for dissemination of results nationally and internationally in the interests of the MPS Community worldwide.

Continue to develop the skill base of the MPS Society's staff team through the implementation of an in-house training programme using the expertise of clinicians, social care providers and the pharma industry.

Host two multi-national expert meetings on MPS IV and the use of Palidronate for ML II/ ML III involving stakeholders and professionals from the MPS community.

Fundraising

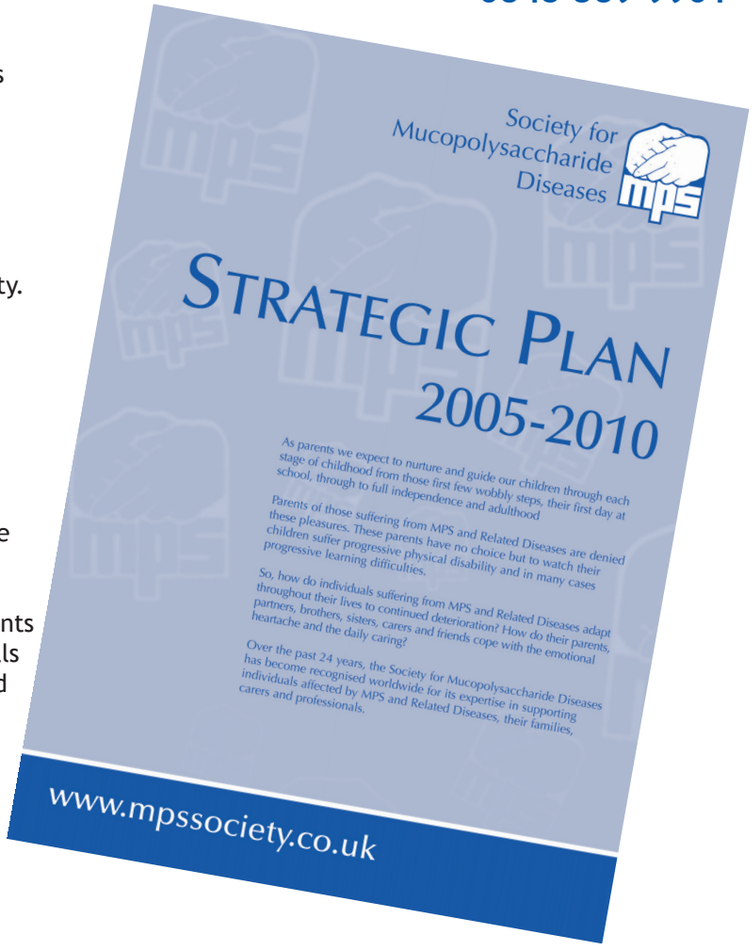
Continue to work with the consultant fundraiser to identify new opportunities for funding.

In collaboration with three Jeans for Genes partner charities implement the five year agreement signed in March 2006 leading to increased awareness and income from the Jeans for Genes Charity.

Working in collaboration with Countrywide Special Events to organise two clay pigeon shoots and two Ollie G Balls between 2008 and 2011 to raise funds for research and special activities for children and adults with MPS.

To maximise fundraising and awareness opportunities in the Society's 25th anniversary year, 2007 – 2008, including hosting a reception at the Palace of Westminster.

If you would like a copy of our strategic plan, please do get in touch. **0845 389 9901**



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Anderson-Fabry Disease in Heterozygous Females

Olaf A Bodamer
Division of Biochemical and Pediatric Genetics, University Children's Hospital Vienna, Austria

Anderson-Fabry disease (AFD; α-galactosidase A [α-gal A] deficiency; OMIM #301500) is an x-linked inborn error of glycosphingolipid metabolism that was first described independently by Fabry and Anderson in a 13-year-old male and a 39-year-old male, respectively, with both patients presenting with symptoms of angiokeratomata and proteinuria [1-13]. Its reported incidence ranges from one in 40,000 – 117,000 live male births [3-4], although a recent report from Northern Italy suggests that the incidence may be as high as one in 3100 live male births [5]. α-gal A deficiency leads to the progressive lysosomal accumulation and storage of globotriaosylceramide (Gb3), digalactosylceramide, and blood-group B, B1, and P1 glycolipids, predominantly in the vascular endothelium, smooth-muscle cells, epithelial cells of the cornea, renal glomeruli and tubules, and ganglion cells of the periphera nervous system [3]. Although storage of these glycolipids may play an important role, the precise pathophysiological mechanism that leads to the clinical phenotype of AFD is not known.

AFD is a devastating, progressive, multi-system disorder that is characterised clinically by:
Acroparasthesia [6]
Hypohidrosis [6]
Gastrointestinal symptoms [7]
Angiokeratoma [3]
Cornea verticillata [3]
Proteinuria with subsequent renal impairment [8]
Cardiomyopathy and conduction abnormalities [9]
Stroke [10]
Many additional but less frequent clinical signs and symptoms [3]

Onset of clinical symptoms may be as early as 6.7±3.4 years of age in males and 7.8±4.5 years in females [6, 11, 12]. Presenting symptoms may be limited to acroparasthesia, delaying diagnosis considerably [6, 11, 12].

The gene coding for α-gal A is located on Xq22.1 and many different, mostly unique, mutations have been identified in patients with AFD [13]. It is estimated that approximately 3-10% of patients with AFD have novel mutations [13]. Diagnosis of AFD in males is based on an analysis of α-gal A activity in leukocytes or plasma, or an analysis of plasma or urinary Gb3 isoforms [3, 14, 15] with molecular analysis serving as the gold standard. However, in females α-gal A analysis may show activity levels within the normal range and diagnosis of AFD may subsequently only be reached through molecular analysis. The development of recombinant enzyme preparations such as agalsidase alfa has improved the long-term prognosis for AFD patients considerably [16, 17]. Different groups have demonstrated the efficacy of biweekly enzyme replacement therapy (ERT) for stabilization or improvement of renal function, and improvement of acroparasthesia, hypohidrosis, and cardiac function [16-19]. Early and prompt initiation of ERT is known to prevent many AFD-associated disease complications and may contribute to the maintenance of an adequate quality of life. Consequently, early diagnosis (preferably within the newborn period) is preferable and current screening strategies employ fluorometric or tandem mass spectrometry techniques [5, 20].

Clinical phenotypes of AFD in Heterozygous Females
For many years it was thought that heterozygous AFD females showed only a few, if any, clinical symptoms. Where more significant clinical signs were found, it was thought that skewed inactivation of the non-mutant X chromosome was the most likely explanation, as this has been demonstrated in other X-linked inborn errors of metabolism [21]. However, an increasing number of individual case reports concerning the clinical manifestations of AFD involving female AFD patients and large surveys among patients with AFD have uncovered an unusually high incidence of affected females [3, 7, 10, 22, 23].

Only the most pertinent reports can be included in this review, although there is a substantial number of individual case reports on female AFD among the biomedical literature databases (for example at www.ncbi.nih.gov). An overview of the current knowledge about clinical phenotypes in heterozygous females is given in Table 1, which includes comparisons with the data available for male AFD patients.

Biochemistry
α-gal A activity in AFD females is within normal limits in a significant proportion of individuals, including those who have a severe clinical phenotype (Table 2) [7, 24, 25].

If a diagnosis of AFD cannot be reached in a symptomatic female using a combination of enzyme analysis and measurement of urinary storage product excretion, molecular analysis should be initiated. This may be straightforward in familial AFD but rather difficult in sporadic cases.

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Table 1. Manifestations of Anderson-Fabry disease in heterozygous females from selected publications.

Manifestation	Number of affected individuals with Fabry disease		Ref.
	Females	Males	
Reduced α-gal A activity	63/119	151/152	[7]
	2/14	13/13	[24]
Hypohidrosis	2/7	15/21	[10]
	4/38	27/29	[24]
	25/42	NR	[28]
Angiokeratoma	82/165	129/165	[7]
	1/7	6/21	[10]
	5/38	27/29	[26]
	11/20	NR	[27]
Acroparesthesia	4/7	7/21	[10]
	18/20	NR	[27]
	26/40	NR	[28]
	34/44	NR	[32]
Proteinuria	5/7	9/21	[10]
	8/38	NR	[27]
Renal impairment	11/20	NR	[27]
	21/36	NR	[28]
	51/202	55/251	[29]
End-stage renal failure	2/60	NR	[22]
	5/40	NR	[28]
	3/256	23/251	[32]
Cardiomyopathy	2/38	3/29	[26]
	11/20	NR	[27]
Gastrointestinal symptoms	4/38	26/29	[26]
	12/20	NR	[27]
White matter lesions on MRI	4/14	NR	[24]
Cebero-vascular complications	45/165	NR	[7]
(i.e. stroke, transient ischemic attack)	7/289*	21/432*	[10]
	2/38	NR	[22]
	13/60	NR	[27]
	8/36	NR	[28]
Reduced vestibular function	4/8	11/13	[31]
Hearing loss	14/38	NR	[28]
Cornea verticillata	2/7	5/21	[10]
	23/32	32/34	[32]
Posterior lens opacities	0/32	4/34	[32]
Retinal vascular tortuosity	6/32	26/34	[32]
Fatigue	24/51	NR	[28]

α-gal: α-galactosidase; MRI: magnetic resonance imaging; NR: not reported. *Population of patients with cryptogenic stroke.

Table 2. Levels of plasma α-galactosidase activity and urinary globotriaosylceramide in patients with AFD and control subjects.

	Plasma α-gal A activity (nmol/h/mL)	Urinary globotriaosylceramide (mg/mmol Cr)
AFD females [25]	0.32-7.50 (n=122)	0.02-0.37 (n=82)
Classic AFD males [25]	0.00-0.91 (n=86)	0.12-2.80 (n=41)
AFD males who carry a mild mutation (N215S) [25]	0.25-0.99 (n=7)	0.01-0.11 (n=7)
Healthy controls [25]	4.1-17.4 (n=100)	0.01-0.03 (n=38)

α-gal: α-galactosidase; AFD: Anderson-Fabry disease; Cr: creatinine.

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Neuronopathic Pain, Hypohydrosis, and Angiokeratoma
Acroparasthesia of fingers and toes may be severe and triggered by heat, fever, exercise, or stress [3]. Its impact on quality of life is significant as even the most potent analgesia may not be sufficient to completely alleviate the pain in some patients [3]. Up to 90% of females with AFD have or have had acroparasthesia and symptoms may start as early as 4 years of age [7, 22, 26, 27]. Angiokeratoma, which is thought to be a pathognomonic sign of AFD in males, is found in up to 55% of AFD females (Table 1) [27].

Renal involvement
Early signs of renal impairment, such as proteinuria and reduced glomerular filtration rate can be identified in up to 58% of females with AFD [10, 27, 28]. Progression to end-stage renal failure is only reported in up to 12.5% of females (Table 1) [22, 28, 29].

Cardiac manifestations
Signs of cardiac involvement including septal hypertrophy, cardiomyopathy, cardiac valve disease, and arrhythmias with the need for pacemaker insertion have been reported in up to 65% of AFD females (Table 1) [7, 9, 22, 23, 26, 27, 30].

Neurological involvement
Rolfs et al. identified seven out of 289 females (2.4%) with otherwise unexplained cryptogenic stroke who had a mutation within the gene coding for α-gal A compared with 21 out of 432 males (4.9%) [10]. Owing to these findings, AFD should be considered in all patients with unexplained stroke episodes even when no other signs of AFD are present. In general, stroke, transient ischemic attack, and other cerebrovascular complications have been reported in 5-21% of females with AFD (Table 1) [7, 22, 26, 31, 32].

Life expectancy
Life expectancy appears to be significantly reduced in both males and females with AFD, mostly owing to disease-related complications such as end-stage renal failure, cerebrovascular events, and cardiac involvement [7, 22]. Whether or not life expectancy in patients with AFD will improve following early diagnosis and initiation of ERT remains to be the subject of randomised long-term studies, which may not be justified ethically.

Conclusion
Heterozygous females with AFD may be as frequently and as severely affected as hemizygous AFD male patients despite the known X-chromosomal inheritance [33]. Therefore, heterozygous AFD females should not be considered as simple carriers but rather as affected patients and should be followed and treated accordingly [34]. These findings are unexpected considering the random nature of X-chromosome inactivation and the fact that enzyme deficiency underlies AFD. At present, the true underlying pathomechanism for disease in AFD females is unknown. It can be speculated that additional factors, such as tissue-specific expression, gene-gene interaction, or semi-dominant effects of the α-gal A gene, play a role [35-36]. However, it is obvious that a diagnosis of AFD in females is difficult to reach, and one has to include AFD in the differential diagnosis irrespective of the inheritance pattern.

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References

1. Fabry J. Ein Beitrag Zur Kenntnis der Purpura haemorrhagica nodularis (Purpura papulosa hemorrhagica Hebrae). Arch Dem Syph 1898; 43: 187-200.

2. Anderson W. A case of angiokeratoma. Br J Dermatol 1898; 10: 113.

3. Desnick RJ, Ioannou YA, Eng CM. ?-Galactosidase A deficiency; Fabry disease. In: Scriver CR, Beaudet AL, Sly WS et al. (editors). The Metabolic and Molecular Basis of Inherited Disease 8th ed. New York, NY; McGraw-Hill, 2001:3733-74.

4. Meikle PJ, Hopwood JJ, Clague AE et al. Prevalence of lysosomal storage disorders. JAMA 1999; 281: 249-54.

5. Spada M, Pagliardini S, Yasuda M et al. High incidence of later-onset Fabry disease revealed by new born screening. Am J Hum Genet 2006; 79: 31-40.

6. Ries M, Gupta S, Moore DF et al. Pediatric Fabry disease. Pediatrics 2005; 115: e344-55.

7. Mehta A, Ricci R, Widmer U et al. Fabry disease defined: baseline clinical manifestations of 366 patients in the Fabry Outcome Survey. Eur J Clin Invest 2004; 34: 236-42.

8. Kleinert J, Lorenz M, Hauser AC et al. Measurement of renal function in patients with Fabry disease. Acta Paediatr Suppl 2005; 94: 19-23.

9. Shah JS, Hughes DA, Sachdev B et al. Prevalence and clinical significance of cardiac arrhythmia in Anderson-Fabry disease. Am J Cardiol 2005; 96: 842-6.

10. Rolfs A, Bottcher T, Zschiesche M et al. Prevalence of Fabry disease in patients with cryptogenic stroke; a prospective study. Lancet 2005; 366: 1794-6.

11. Bodamer OA, Ratschmann R, Paschke E et al. Recurrent acroparasthesia during febrile infections. Lancet 2004; 363: 1698.

12. Ramaswami U, Whybra C, Parini R et al. Clinical manifestations of Fabry disease in children: data from the Fabry Outcome Survey. Acta Paediatr 2006; 95: 86-92.

13. Schafer E, Baron K, Widmer U et al. Thirty four novel mutations of the GLA gene in 121 patients with Fabry disease. Hum Mutat 2005; 25: 412.

14. Fauler G, Rechberger GN, Devrnja D et al. Rapid determination of urinary globotriaosylceramide isoform profiles by electrospray ionization mass spectrometry using stearoyl-d35-globotriaoslceramide as internal standard. Rapid Commun Mass Spectrom 2005; 19: 1499-506.

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Fabry International Network Conference & General Meeting

9 - 10 February 2008

I was fortunate to have been invited to attend the Fabry International Network (FIN) meeting which was held this year in Amsterdam. The meeting was being held over two half days which enabled people from all over the world to arrive in good time for the start.

There were representatives from eighteen countries, including myself and Dr Waldek from the UK, attending and representatives from three pharmaceutical companies, Shire, Genzyme and Amicus.

The presentations given by members of the FIN Board were informative and gave me a better insight into what goes on. Following that came presentations from Prof Dr J Aerts, Dr G Linthorst, Dr Waldek, and Dr Raas Rothschild. Following each presentation there was an opportunity for discussion and an open forum for debating.

Over the two half days we were all split into small groups where we held discussions on various topics, one of which was "Key Activities, Challenges and Successes; What have been the main activities of your group/organisation in relation to Fabry; What have been your key successes/strategies that have worked and what are the main challenges you face?"

What I found most interesting and saddening too was that not all countries get the ERT funded by their NHS or equivalent. Many countries run out of money towards the end of the financial year so there are no funds for treatment therefore the individuals go without. Although some individuals in the UK struggle to get ERT they know that if they get awarded it they do not have the worry of funds running out at the end of a financial year.

It was a pleasure to attend the FIN Meeting and great to meet so many professionals from around the world. It was also great to see the pharmaceutical companies working together in teams!

A very big thank you to Ed, Marlene, Rune, Kees and all on the FIN Board for allowing me the opportunity to take part in such a successful meeting. **Neisha Hall**

15. Young E, Mills K, Morris P et al. Is globotriaosylceramide a useful biomarker in Fabry disease? Acta Paediatr Suppl 2005; 94: 51-4.

16. Schiffmann R, Murray GJ, Treco D et al. Infusion of alpha-galactosidase A reduces tissue globotriaosylceramide storage in patients with Fabry disease. Proc Natl Acad Sci USA 2000; 97: 365-70.

17. Eng CM, Guffon N, Wilcox WR et al. Safety and efficacy of recombinant human alpha-galactosidase A – replacement therapy in Fabry's disease. N Engl J Med 2001; 345: 9-16.

18. Schiffmann R, Floeter MK, Dambrosia JK et al. Enzyme replacement therapy improves peripheral nerve and sweat function in Fabry disease. Muscle Nerve 2003; 28: 703-10.

19. Weidemann F, Breunig F, Beer M et al. Improvement of cardiac function during enzyme replacement therapy in patients with Fabry disease: a prospective strain rate imaging study. Circulation 2003; 108: 1299-301.

20. Gelb MH, Turecek F, Scott CR et al. Direct multiplex assay of enzymes in dried blood spots by tandem mass spectrometry for the new born screening of lysosomal storage disorders. J Inherit Metab Dis 2006; 29: 397-404.

21. Tuschl K, Gal A, Paschke E et al. Mucopolysaccharidosis type II in females: case report and review of literature. Pediatr Neurol 2005; 32: 270-2.

22. MacDermot KD, Holmes A, Miners AH. Anderson-Fabry disease: clinical manifestations and impact of disease in a cohort of 60 obligate carrier females. J Med Genet 2001; 38: 769-75.

23. Chimenti C, Pieroni M, Morganta E et al. Prevalence of Fabry disease in female patients with late-onset hypertrophic cardiomyopathy. Circulation 2004; 110: 1047-53.

24. Fellgiebel A, Muller MJ, Mazanek M et al. White matter lesion severity in male and female patients with Fabry disease. Neurology 2005; 65: 600-2.

25. Winchester B, Young E. Biochemical and genetic diagnosis of Fabry disease. In: Mehta A, Beck M, Sunder-Plassmann G. editors. Fabry Disease: Perspectives from 5 Years of FOS. Oxford, UK: Oxford PharmaGenesis Ltd. 2006; 163-81.

26. Galanos J, Nicholls K, Grigg L et al. Clinical features of Fabry disease in Australian patients. Intern Med J 2002; 32: 575-84.

27. Whybra C, Kampmann C, Willers I et al. Anderson-Fabry disease: clinical manifestations of disease in female heterozygotes. J Inherit Metab Dis 2001; 24: 715-24.

28. Wang RY, Lelis A, Mirocha J et al. Heterozygous Fabry women are not just carriers, but have a significant burden of disease and impaired quality of life. Genet Med 2007; 9: 34-45.

29. Palla A, Widmer U, Straumann D. Head-impulse testing in Fabry disease; vestibular function in male and female patients. Acta Paediatr Suppl 2003; 92: 38-42.

30. Kampmann C, Baehner F, Whybra C et al. Cardiac manifestations of Anderson-Fabry disease in heterozygous females. J Am Coll Cardiol 2002; 40: 1668-74.

31. Fellgiebel A, Muller MJ, Ginsberg L. CNS manifestations of Fabry's disease. Lancet Neurol 2006; 5: 791-5.

32. Mehta A, Ginsberg L et al. Natural history of the cerebro-vascular complications of Fabry disease. Acta Paediatr Suppl 2005; 94: 24-7.

33. Eng CM, Fletcher J, Wilcox WR et al. Fabry disease: baseline medical characteristics of a cohort of 1765 males and females in the Fabry Registry. J Inherit Metab Dis 2007; 30: 184-92.

34. Eng CM, Germain DP, Banikazemi M et al. Fabry disease: guidelines for the evaluation and management of multi-organ system involvement. Genet Med 2006; 8: 539-48.

35. Maier EM, Osterrider S, Whybra C et al. Disease manifestations and X inactivation in heterozygous females with Fabry disease. Acta Paediatr Suppl 2006; 95: 30-8.

36. Stromberger C, Bodamer OA. Fabry disease defined. Euro J Clin Invest 2004; 34: 449.

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‘Focus on Fabry’ goes live

Shire’s first Fabry disease internet resource launched on European Rare Disease Day...

Shire Human Genetic Therapies (Shire HGT), a business unit of Shire plc, recently announced the launch of ‘Focus on Fabry’ (www.focusonfabry.com), to coincide with the first European Rare Disease Day. ‘Focus on Fabry’ is Shire’s first website dedicated to raising awareness and understanding of Fabry disease among healthcare professionals and the general public. ‘Focus on Fabry’ has been initiated by Shire HGT, who will also be funding its ongoing maintenance and development.

Fabry disease is a rare genetic disorder that can cause a wide range of symptoms such as pain, gastrointestinal complaints, skin rashes (angiokeratomas), renal failure, and can lead to an increased risk of heart disease and stroke.[1] Due to the varied nature of the symptoms and the slow progressive course of the disease, there is an average delay between the onset of symptoms and diagnosis of 12 years. [2] Life expectancy is generally reduced by 20 years in men and 15 years in women with Fabry disease, compared with the normal population. [3,4].

‘Focus on Fabry’ has been developed by Shire HGT to provide information to both the general public and healthcare professionals on the causes, signs and symptoms, diagnosis, and management of the disease as well as providing details of other sources of support. It is hoped that ‘Focus on Fabry’ will be an invaluable resource for healthcare professionals interested in learning about the disease and for members of the general public who may be directly or indirectly affected by this disorder.

“Fabry disease is rare: the symptoms are very varied and similar to a number of other conditions – as a result, patients often go undiagnosed for many years,” explains Dr Patrick Deegan of the Lysosomal Disorders Unit at Addenbrooke’s Hospital, Cambridge. “Raising awareness of the disease will help to ensure that patients are diagnosed as early as possible so that they can receive appropriate care. ‘Focus on Fabry’ is a very useful and informative resource towards this goal.”

The launch of ‘Focus on Fabry’ coincides with European Rare Disease Day – a day devoted to raising awareness about rare diseases among decision makers, healthcare professionals and the general public. European Rare Disease Day aims to improve equality in access to care and treatments, increase funding for research and care, and bring all stakeholders together to improve the lives of individuals living with rare diseases. [5]

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References

1. Hughes DA, Ramaswami U, Elliott, P, Deegan P, Lee P, Waldek S, et al. Guidelines for the diagnosis and management of Anderson-Fabry disease. Department of Health, 2005.

2. Beck M. Demographics of FOS - the Fabry Outcome Survey. In: Mehta A, et al (eds). Fabry disease: perspectives from five years of FOS, 2006; pp. 155-161.

3. MacDermot KD, Holmes A, Miners AH. Anderson-Fabry disease: clinical manifestations and impact of disease in a cohort of 98 hemizygous males. Journal of Medical Genetics 2001; 38: 750-760.

4. MacDermot KD, Holmes A, Miners AH. Anderson-Fabry disease: clinical manifestations and impact of disease in a cohort of 60 obligate carrier females. Journal of Medical Genetics 2001; 38: 769-775.

5. European Rare Disease Day, available at http://www.rarediseaseday.org (accessed on 14th February 2008).

6. Mehta A, Ricci R, Widmer U, Dehout F, Garcia de Lorenzo A, Kampmann C, et al. Fabry disease defined: baseline clinical manifestations of 366 patients in the Fabry Outcome Survey. European Journal of Clinical Investigation 2004; 34: 236-242.

7. Meikle PJ, Hopwood JJ, Clague AE, Carey WF. Prevalence of Lysosomal Storage Disorders. Journal of the American Medical Association 1999; 281: 249-254.

Lysosomal Storage Disorders Study Day - Birmingham Children’s Hospital

On 11th December 2007 I attended a study day at Birmingham Children’s Hospital. The day was to inform medical teams about how to recognise Lysosomal Storage Disorders in both children and adults, the causes of these conditions, the early signs and symptoms and the treatments available.

Specialists in areas such as Neurology, Ophthalmology, Radiology, Bio-Chemistry and Anaesthetics gave an overview of their involvement and how they manage these conditions.

Presentations were also given from the specialist nursing team explaining Enzyme Replacement Therapy, as well as the other areas of support that they offer.
I explained the role and services of the MPS Society. The day was very successful and it was certainly useful to meet all the professionals, other days are to be planned for 2008.
I would like to thank the team at Birmingham Children’s Hospital for inviting the MPS Society.
Steve Cotterell

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The Lysosomal Storage Disorders Unit

Royal Free Hospital, London

The Royal Free Hospital Lysosomal storage disorders unit (LSDU) is based within the Royal Free Hampstead NHS Trust located in Northwest London and is also part of the Royal Free and University College Medical School.
Our unit was first established over 25 years ago to care for patients with Gaucher disease (a lysosomal storage disorder resulting in problems with blood, bones, liver and spleen). The National Specialist Commissioning Advisory Group (NSCAG) first designated the Royal Free Hospital as one of the specialist centres treating Gaucher disease in 1997. This remit has since been extended to include other Lysosomal Storage Disorders (LSD) for which therapy is available.

The centre now offers comprehensive care for patients with a number of LSDs including Gaucher, Anderson-Fabry and Pompe disease. We care for over 250 patients from throughout the British Isles. Our core team consists of two consultants, Dr. Atul Mehta and Dr. Derralynn Hughes, a team of specialist nurses lead by Linda Richfield and a number of laboratory and research personnel. The unit undertakes a wide range of research and development; including basic laboratory research and clinical trials of new therapies for LSDs. The molecular and enzyme laboratory provide diagnostic services on site.

The centre benefits clinically and academically from its location within the context of a major teaching hospital. The rare disorders treated at our unit can affect various organ systems and require the input and specialist knowledge of a number of medical and surgical specialities. It is our intention to be able to provide as much access as possible to these specialists in one centre. To this end we have assembled a group of physicians and nurses with accumulated experience of lysosomal disorders. We understand that a patient's pathway to diagnosis may have taken many years and been frustrated by miss-diagnosis and misinterpretation of signs and symptoms along the way, we therefore aim to minimize waiting times for screening, assessment and treatment once a patient has finally been referred to our centre. We see each patient as an individual with specific needs and requirements and tailor our assessment and investigations to that individual.
Most of our patients receive their enzyme replacement therapy at home and we are very pleased that we have recently been able to extend this convenience to our adult patients with Pompe disease. We also try to assist our patients in communication with other physicians or outside bodies who do not have knowledge or understanding of their condition.

The LSDU at the Royal Free works closely with the UK MPS Society in the care and support of patients with Anderson-Fabry disease and with the other relevant patient support and advocacy groups. We regularly host patient meetings which are open to patients and families attending other specialist centres. There is a meeting planned for later this year and information will be circulated when the final programme has been drafted.

Direct line/fax: 0207 472 6409

www.royalfree.nhs.uk/lsdu/

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SPOTLIGHT ON...

Home treatment from Careology

The breadth and depth of care required to support individuals and families who are living with rare genetic disorders is large indeed and whilst Careology would never claim to be able to meet all these needs we are determined to offer as much support as possible from a practical perspective.

Much of the Careology team are seasoned and experienced healthcare professionals who have chosen to build an expertise in this and many other complex therapeutic areas.

Our ethos is to use this expertise to provide a highly specialist clinical service for children and adults who need support with the administration of specialist products at home.

We are particularly proud of our work which was led by the team at Great Ormond Street to help to make the administration of Elaprase to Hunters children at home a reality. Careology's involvement started with Great Ormond Street when we were asked to provide research nurses to work with several children who were taking part in the Elaprase trial. This enabled us to develop relationships with the children and their families and to gain experience to help us understand the complex nature of this disorder.

Elaprase was licensed in February 2007 and approval was quickly given to allow these children to receive their treatment at home.

From Careology's perspective this prompted a detailed planning process to ensure that the transition from hospital to home treatment would take place smoothly. We needed to ensure that supply arrangements were in place to obtain the Elaprase and that our pharmacy team, patient service coordinators, home delivery drivers and specialist nurses were all fully aware of the requirements of each individual family.

We wanted to provide a truly personalised service to each family and this was helped immensely because our nurses already knew the families from their involvement in the clinical trial at Great Ormond Street.

In addition to this with our experience of working with children in the home setting, we were able to consider various equipment options and processes that would ensure the treatment would be given safely and with ease.

Our first requirement was to ensure that we had an infusion pump that would enable the children to move around with their medication whilst it was being administered. We have learnt that this is an important step with home infusions for children! Our lead Paediatric nurse was able to source a pump that was light and would comfortably fit into a backpack with the infusion bag.

We also understood that it would be important to provide home visits that would not interfere with schooling or family life. We do provide home visits after school; however it was important also to ensure that the initial visits took place whilst support could be provided from the Great Ormond Street team should any advice be required.

After the first two visits, provided all had gone well, these time constraints were relaxed and infusions took place outside of normal clinic hours.

There is no getting away from the fact that these are long infusions whether they take place at hospital or home but our job is to look for other approaches that may enable infusions to take place whilst not interfering with school commitments or other social activities. This was achieved by understanding each family's needs on an individual basis and looking for alternative solutions.

It is a real tribute to some families that they have now become totally independent and are administering their own infusions at home.

As home treatment begins we have a training programme that is designed to suit each individual family. If a family does become independent, we still have regular communication through our patient service coordinators and our specialist nurses are only a phone call away to offer support. We will also provide six monthly reviews where a nurse will visit to discuss any matters that may have occurred and answer any questions that the family may have. In addition, we do provide an on call service that will be answered by a nurse who is familiar with the therapy so that we are able to offer useful and effective advice.

Careology is now supporting children and adults in their own homes who are receiving many different enzyme replacement therapies. It has been a great experience and has enabled us to develop our service to meet the individual needs of the child and the family in addition to working closely with the highly specialist referral centre teams.

We are constantly looking to develop our teams expertise and as we become involved with each new therapeutic area and each new group of families we discover that we learn more and more about how to provide a truly individual service to help support each family.

On behalf of the whole of the Careology team I would like to congratulate the MPS Society on the celebration of their 25th year as a highly valued patient association.

Dominic Moreland dmoreland@careology.co.uk

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Amicus Therapeutics Announces Positive Results From

Phase 2 Clinical Trials of Amigal™ for Fabry Disease

Amicus Therapeutics Inc. - a biopharmaceutical company developing small-molecule, orally administered pharmacological chaperones for the treatment of human genetic diseases, recently announced positive results from its recently completed Phase 2 clinical trials of Amigal™ (migalastat hydrochloride) for Fabry disease.

As of November 2007, Amigal is being developed in partnership with Shire Human Genetic Therapies (HGT), a business unit of Shire plc, which is focused on genetic diseases. The results will be discussed as a part of an "R&D Day" meeting.

"The completion of these trials is a major milestone for Amicus and these clinical results represent an important proof of concept for the pharmacological chaperone platform technology." said John F. Crowley, President and CEO of Amicus Therapeutics. "We look forward to advancing our program in Fabry as well as Gaucher, Pompe and other important therapeutic targets utilizing this new approach to the treatment of a wide range of human genetic diseases."

Summary of Study Results
The primary objective of the Phase 2 trials was to evaluate the safety and tolerability of treatment with Amigal. The secondary objective was to evaluate certain pharmacodynamic measures of treatment, including effects on alpha-GAL (the target enzyme deficient in Fabry patients) and levels of GL-3 (the substrate that builds up in the cells of patients) in cells and tissues affected by the disease. An additional objective was the preliminary assessment of cardiac and renal function.

The four open-label, multi-national Phase 2 trials of Amigal enrolled 18 men and 9 women with Fabry disease between the ages of 17 and 65. The four studies examined various dose levels and frequencies of Amigal administration and had 12 or 24 week primary treatment arms with an optional treatment extension.

Twenty-six patients completed the primary treatment arms and all entered the optional treatment extension. The 26 patients had 21 different missense genetic mutations that cause Fabry disease. The mutations represented the full spectrum of Fabry patients, including those with both early-onset and late-onset forms of the disease. Twenty-three patients are currently being treated with Amigal under the treatment extension, including 8 who have been treated for more than a year and 4 who have been treated for almost two years.

"The positive results of these first trials of a pharmacological chaperone in Fabry disease are impressive," said Raphael Schiffmann, M.D., Lead Investigator at the Metabolic Neurology Branch of the National Institute of Neurological Disorders and Stroke (NINDS), a part of the National Institutes of Health, and a principal investigator in one of the Amigal clinical trials. "I believe this technology has the potential to be an important new treatment option for many Fabry disease patients."

The key findings in the Phase 2 studies were:
Amigal was generally safe and well-tolerated at all doses evaluated. No drug-related serious adverse events were reported during the primary treatment arm, and none have been reported during the treatment extension.

Twenty-four out of 26 patients demonstrated an increase in alpha-GAL as measured in white blood cells, kidney, and skin.

Alpha-GAL increases were seen in patients with both low levels of residual enzyme activity (<3%) at baseline as well as patients presenting with higher baseline levels (>3%).

Kidney GL-3 levels as measured in urine or biopsies were decreased in patients who demonstrated greater increases in levels of alpha-GAL.

Renal and cardiac function results were encouraging, including those seen in patients treated for nearly two years.

Patient responses were consistent with the results of in vitro testing of Fabry mutations, thus improving the ability to select likely responders for future studies.

"These data demonstrate that Amigal has a meaningful effect on a range of genetic mutations in Fabry disease and thus has the potential to treat a significant portion of the Fabry patient population" said William Wilcox, M.D., Ph.D., Director of the Metabolic Disorders Clinic at Cedars-Sinai Medical Center and a principal investigator in one of the Amigal clinical trials.

Amicus expects that the results will be presented again at the American College of Medical Genetics (ACMG) Annual Meeting on March 12-16, 2008, in Phoenix, Arizona.

Based on the results of these Phase 2 trials, Amicus and Shire plan to meet with US and European regulatory authorities to discuss the design of a Phase 3 clinical trial for Amigal. [cont. bottom of next page]

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Vivendy Therapeutics Ltd. to develop an Enzyme Replacement Therapy for MPS IVA (Morquio Disease)

Vivendy Therapeutics Ltd. is a pharmaceutical start up company that is developing an enzyme replacement therapy (ERT) for Morbus Morquio (Mucopolysaccharidosis (MPS) IVA), a rare lysosomal storage disease. Since it is a rare disease, it applies to orphan drug-status criteria in the United States, Europe and Japan.

The new CEO, Dr. Roland Toder states: "With no drug therapy available, Morquio patients and their families experience prolonged and significant suffering. The ERT provides Morquio patients with the enzyme they are naturally lacking and therefore represents the first true therapy for this rare disease. We are pleased that we were able to attract such world class investors to our company at this important time in our development. We are confident that with their support, we will build on our promising preclinical results to advance the clinical development program of an enzyme replacement therapy for MPS IV."

Today, there is no therapy for MPS IVA in place, therefore the development of Morbus Morquio ERT addresses unmet medical needs with a high chance of success due to the fact that Vivendy Therapeutics is applying the established strategy of ERT with the Morquio specific enzyme. Dr. Christoph Heinzen, Founder and Technical Director of Vivendy Therapeutics said: "We developed a specific recombinant human enzyme tailored to meet the particular requirements for Morbus Morquio - enhancing the efficacy of the therapy significantly."

Dr. Gerhard Ries, General Partner at BioMedInvest and member of Vivendy's Board of Directors is excited about Vivendy's ERT project: "We are delighted to be associated with this impressive team working on such an unbearable disease for which there is no cure. We strongly believe that Vivendy's ERT approach has a high chance of success."

Dr. Annegret de Baey-Diepolder, Partner at TVM Capital and member of Vivendy's Board of Directors highlights: "The lean company structure addresses the need for increased capital efficiency in early stage development situations."

Dr. Jörg Neerman, Partner at LSP and member of Vivendy's Board of Directors adds "According to a new regulation within the EU an early access for MPS IVA patients to the new treatment can be provided. This would allow returns on investment earlier and before the total cost of product development has been expended."

About Vivendy Therapeutics Ltd.
Vivendy Therapeutics was founded in March 2006, as a spin-off of Inotech Biotechnologies AG. The company's mission is the development of an enzyme replacement therapy (ERT) for Morbus Morquio (Mucopolysaccharidosis MPS IVA), a rare lysosomal storage disease, based on a gene deficiency with an incidence of 1 to 200 000 live births. MPS IVA represents 5% of the lysosomal storage disorders (LSDs).

Dr Roland Toder Chief Executive Officer
14 January 2008

[cont. from page before] **About Amicus Therapeutics**
Amicus Therapeutics is a biopharmaceutical company developing novel, oral therapeutics known as pharmacological chaperones for the treatment of a range of human genetic diseases. Pharmacological chaperone technology involves the use of small molecules that selectively bind to and stabilize proteins in cells, leading to improved protein folding and trafficking, and increased activity. Amicus is initially targeting lysosomal storage disorders, which are severe, chronic genetic diseases with unmet medical needs. Amicus has completed Phase 2 clinical trials of Amigal for the treatment of Fabry disease and is conducting Phase 2 clinical trials of Plicera™ for the treatment of Gaucher disease.

The Company has completed Phase 1 clinical trials of AT2220 for the treatment of Pompe disease.

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Therapy Prospects for MPS IIIA Demonstrated with Animal Models

John Hopwood

Lysosomal Diseases Research Unit, Women's and Children's Hospital, Adelaide, Australia

Professor Hopwood started by stating his goal for the last 25 years and then presented his paper 'Early diagnosis and effective therapy for Lysosomal Storage Disease Patients'.

At present, there is no safe and effective treatment for lysosomal storage disorders (LSDs) that affect the brain. Mucopolysaccharidosis type IIIA (Sanfilippo A Disease) results from a lack of functional heparin-N-sulphatase (HS) and other secondarily-stored compounds subsequently accumulate, primarily within the central nervous system (CNS), resulting in progressive mental deterioration and early death. Clinical presentation varies but a usual pattern results in:

Age of clinical presentation 2-7 years

Frequent and severe temper tantrums; hyperactivity

Death usually between 15 and 35 years

It is estimated the MPS III A, B, C, D incidence is 1:55,000.

Naturally occurring MPS IIIA mouse and dog models were used to evaluate the effect of repeated injections of recombinant human SGSH (rhSGSH) into the cerebrospinal fluid via the cisterna magna (CM) of the mouse and dog CNS pathology and behavioural function in the mouse. The cisterna magna is between the cerebellum and medulla in the brain. It is part of the subarachnoid space, through which the cerebrospinal fluid that surrounds the brain moves.

Studies using these animal models sought to determine the effectiveness of intra-cerebral spinal fluid of human SGSH on brain pathology as a potential treatment. Unpublished mouse studies by Hemsley et al 2007 have demonstrated a dose-dependent reduction of heparin sulphate storage in the brain and spinal cord, reduced lysosomal vesicle formation in various cell types, decreases in the number of activated neuroglia and fewer axonal spheroids were observed in several brain regions. The biochemical changes were accompanied by improved behaviour particularly in mice treated more frequently. In a similar approach by Hemsley et al, again unpublished, with MPS IIIA Huntsman dogs from New Zealand, human SGSH was administered into the cerebral spinal fluid of MPS IIIA dogs via the cisterna magna. All treated MPS IIIA dogs had high concentration of human SGSH throughout the brain and spinal cord. Storage of heparin sulphate derived oligosaccharides was significantly reduced in many brain regions.

These studies are continuing to evaluate the effect of enzyme dose and frequency on the efficiency to clear lysosomal storage deep into the brain of both animal models and to prevent the development of clinical disease pathology.

Professor Hopwood concluded by saying that their preliminary results suggest that this method of enzyme delivery may present an immediately applicable short to medium term means of reducing the neurological pathology associated with MPS IIIA and other lysosomal storage diseases affecting the brain. He also outlined the limitations to therapies for MPS III; the need to define the nature and time of irreversible neuronal pathology; the transfer of therapeutic enzyme/agent into the brain; and identification of biomarkers to directly monitor therapy and storage of disease.

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Celebrating

Jeans for Genes Day 2007

Famous landmarks and famous people were among the fundraisers for last year's Jeans for Genes Day on Friday 5th October. Statues across the country did their bit in encouraging people to get involved in 'denimising the nation' and raising money for the charity.

Dylan Thomas in Swansea is denimised

In Morecambe in Lancashire Eric Morecambe was a wonderful sight in the morning sunshine in a pair of jeans created by fashion designer and Lancastrian, Wayne Hemmingway. He'd borrowed an original pair of Eric's glasses from his son, Gary, to create the bleached design and the jeans were then tailormade by local artist, Jane Anderson.

In Swansea, Dylan Thomas was somewhat warmer than usual on the seafront, in his denim scarf and customised jeans. Together with one of his famous characters, Captain Cat, the poet inspired the whole city to offer its support to Jeans for Genes Day. Not to be outdone, Cardiff 'denimised' their local hero, the former rugby union star, Gareth Edwards.

You can see photographs of more statues around the UK which were 'denimised' for the Day, and supporters having fun and fundraising, by visiting the website **www.jeansforgenes.com/yourphotos.**

In Manchester, construction consultancy Currie and Brown combined their favourite family recipes to produce a cook book that they then sold in aid of Jeans for Genes. On the Day they served up some of the dishes for lunch and so far they've raised close to £400 - and counting!

Kathryn and Julie took their buckets, their cowboy hats and their best smiles and headed off to Sainsbury's at Fosse Park in Leicester. Kathryn is a member of the Fragile X Society, one of Jeans for Genes' guest charities in 2007. The pair spent several hours collecting with Kathryn's brother, Howard, who has Fragile X syndrome, an inherited learning disability. They all had a wonderful day and raised £200.

Amongst the younger fundraisers on Jeans for Genes Day were the toddlers at Rainbow Nursery in Birstall in Leicestershire. They are big fans of Fifi and the Flowertots and they dressed as their favourite characters from the children's TV series on a walk to Watermead Park. But they didn't stop there! They managed to dress Woolley, a life size statue of a mammoth, in a giant pair of jeans!

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And perhaps one of the most unusual Jeans for Genes Day events was held by staff at Adept Scientific in Letchworth in Hertfordshire. As well as having a cake sale they all dressed as Jon Bon Jovi!

In the West Country, Toby Martin, who has Hunter syndrome, showed local Jeans for Genes supporters how effective his enzyme replacement therapy has been. On Jeans for Genes Day in 2006, he appeared on local television in a wheelchair. In 2007 he was pictured playing football.

JEANS FOR GENES APPEAL

Jeans for Genes Day 2008 takes place on Friday 3rd October, and there's plenty of fundraising going on in the meantime! A team of ten marathon runners will be aiming to raise more than £20,000 when they take to the streets of London on Sunday April 13th. If you'd like to help Jeans for Genes to cheer them on, you can contact Amanda Sinke, the events manager on 020 7163 6901.

Jeans for Genes is aiming to raise £3 million in 2008. Proceeds from the Day will benefit members of the MPS Society, and other families living with genetic disorders. If you'd like to get involved in Jeans for Genes Day on Friday 3rd October visit **www.jeansforgenes.com/donate**

Photo top left: Sainsbury's staff get in the spirit at the Oadby store in Leicestershire. Photo bottom right: Children at Rainbow Nursery in Leicester dress as Fifi and put giant jeans on Woolley the Mammoth

3 October 2008

www.jeansforgenes.com

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Brains for Brains Second European Workshop

Frankfurt, Germany, 7 - 9 March 2008

Brains for Brains (B4B) is a research consortium of distinguished scientists, leaders in basic and applied neurotechnology and neurology grouped together to create a co-ordinated effort towards the comprehension of pathophysiology processes of the neurodegenerative lysosomal storage diseases (LSDs), the implementation of knowledge on the blood brain barrier (BBB) and the development of new molecular and/or biochemical strategies to overcome the BBB and treat neurological diseases caused by storage in the lysosomes.

The aim of the workshop was to discuss research achievements at a clinical and basic science level in the field of neurodegenerative lysosomal storage diseases and the blood brain barrier. This aim was enriched by discussions on how B4B might collaborate with the European Union to stimulate interest in research on LSDs and the BBB and to consider collaborations with the international patient organisations and the pharma industry.

Christine Lavery and Tanya Collin-Histead (Chief Executive of the Gaucher Association) represented the international LSD patient association community and made a joint presentation on 'Advocate Associations and B4B: A Collaborative Effort'. Christine was able to describe the natural history and longevity studies for MPS I and MPS II carried out by the Society in partnership with the Willink Genetics Centre at the Royal Manchester Children's Hospital and share information on research initiatives currently funded by the MPS Society.

Photo above: Dr Maurizio Scarpa, Photo below: Christine with Dr Helen Michaelkakkis and Tanya Collin-Histead

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Information for Carers

Carers' legal rights in employment

The Work and Families Act 2006 gives certain rights to families to request flexible working such as changing hours or working from home. From April 2007 the act extended the right for carers of a disabled adult to also request flexible working. This applies to all parents, carers, guardians, foster parents and holders of a residence order.

This means that employees can apply to make a permanent change to their terms and conditions. However, only one request can be made per year and although employers must consider any requests for flexible working they do not have to agree them if there is a good business reason why an application is refused. In these circumstances employees have the right to appeal. Your employee should have a policy regarding flexible working and may have their own application form to complete if you are wanting to make a request.

The act defines a carer as someone who cares for, or expects to care for, a husband, wife or partner, a relative such as a child, uncle, sister, parent in law, son in law or grandparent, or someone who falls into neither category but lives at the same address.

Under the Employment Relations Act 1999 employees gained the right to 'reasonable time off' to deal with any unexpected situations that arise in relation to their caring or parental roles. There are a number of different ways in which leave can be granted these are; carers leave, compassionate leave, borrowing or buying leave or taking a career break. At the discretion of the employer, time off can be paid.

Situations where leave might be taken include:

A disruption or breakdown in care arrangements;

If a dependent falls ill or has been assaulted or in an accident including when the victim is hurt or upset rather than physically injured;

To deal with an incident involving a child during school hours;

To make longer term arrangements for a dependent who is ill or injured.

Parental Leave

If you are a parent with parental responsibility who is named on your child's birth certificate or adoption certificate you have a statutory right to take parental leave if you have a child under five years of age or a disabled child up to 18 years of age.

If you have one year's continuous service and you're an employee with a contract of employment you are entitled to 13 weeks of parental leave for each child under five years or 18 weeks for each disabled child up to 18 years.

Most employers will have their own set up for employees wishing to take parental leave, however if this is not the case the following applies.

Leave can only be taken in blocks of full weeks. Odd days should be taken as holiday or requests should be made to your employer to work flexibly. However, if your child has a disability, days rather than weeks can be used to take into account the possible need for hospital appointments.

Only four weeks leave can be taken for any one child and you must give your employer 21 days notice. Your employer can also postpone any leave for up to 6 months if it will cause a disruption to the business. You are also able to carry over any unused leave to a new job but you cannot take this leave until you have been in their employment for 1 year.

Carers Allowance

If you care for or look after someone who is disabled you may be able to claim Carers Allowance. You do not have to be related to the person.

To qualify you must be over 16 years of age and spend 35 hours or more a week caring for a disabled person. The disabled person must be in receipt of Attendance Allowance, Disability Living Allowance or Constant Attendance Allowance. However, you will not be entitled to receive Carers Allowance if you are in full time education with over 21 hours of supervised study or earn £95 or over a week, less reductions. It is also important to note that if you are in receipt of other benefits and make a claim for Carers Allowance, this may affect them or the total amount you are awarded.

For more information or to apply online you can either look on the website www.direct.gov.co.uk or apply to Carers Allowance Unit, Palatine House, Lancaster Road, Preston, PR1 1HB, telephone: 01253 856123 or email: customer-services@dwp.gsi.gov.uk.

You may also wish to contact Carers Line
Tel: 0808 808 7777
Carers Line is Carers UK's free advice line for carers. It is open on Wednesday and Thursday each week between 10-12 pm and 2-4 pm. www.carers.gov.uk

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React's Holiday Homes

All families can apply for a week's holiday at one of our mobile homes around the UK. We have two in Scotland, two on the Isle of Wight, and one mobile home in each of the following: Northern Ireland, Lincolnshire, South East Wales and Blackpool.

Our three-bedroom holiday homes are located in family-friendly parks and can accommodate six people in comfort. The homes are self-catering and site-fees are included.

Please note that unfortunately our mobile homes are not wheelchair adapted. However if our holidays are unsuitable for your child we may be able to contribute towards an equivalent holiday if you send us relevant information and prices.

Applying is exactly the same process as for other awards; just write React Holiday under 'Purpose of Application'. Our season runs from March/April to the end of October, and you can call us to check availability or reserve a particular week until your application is processed (note the longest we can do this for is 14 days).

For more information, please visit
www.reactcharity.org or phone **020 8940 2575**

Safespaces®

Safespaces began in the North of England in the 1990's. Since then, the company has developed to provide flexible alternatives to room padding for those with autism, epilepsy, challenging behaviour and other special needs.

Safespace rooms can be described as a 'room within a room' and can fill the whole of a room or fit in part of a larger room. Because of the Safespace's unique design, no additional building work is required and it allows for the continued use of existing facilities, so windows, radiators, plug sockets and lights can all be used normally to create a pleasant and cosy environment.

For more information:
Safespaces (Cornholme) Ltd
11 Cleveland Street
Todmorden
Lancashire
OL14 8LZ

Tel/Fax: 01706 816274
Email: info@safespaces.co.uk
www.safespaces.co.uk

Buggy Cover available!

We have very kindly been donated a cover for a buggy. This cover can act as a rain cover or as a sun shield. It is very flexible and we have been informed it will fit onto most buggies, as it clips on at the side. If anyone is interested in this item or would like to find out more please contact the MPS Office. There is no charge for the cover.

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POSTAGE

Please ensure that you use the correct postage. As a charity we do not collect undelivered items due to the expense incurred.

All figures shown here reflect the new pricing system, which is based on size as well as weight.

LETTER FORMAT
C5+ or under. Up to 100g.
Less than 5mm thick.
Max weight 100g
Max dimensions 240 x 165 x 5mm
For example: Most letters, bills, statements, greetings cards, some brochures and catalogues

LARGE LETTER FORMAT
B4 or under. Up 750g.
Less than 25mm thick.
Max weight 750g
Max dimensions 353 x 250 x 25mm
For example: Letters containing unfolded A4 paper, most brochures, CDs and DVDs

Any questions please visit **www.royalmail.com**

'Donate As You Spend' MasterCard

Donate As You Spend is an organisation founded to help smaller charities raise funds in ways only previously available to the largest charities. MPS are one of the first charities to take advantage of their new MasterCard.

Donate As You Spend Donations to MPS
£10 (plus Gift Aid £2.80) to MPS when you first use your card, 25p (plus 7p Gift Aid) to MPS for every £100 you spend on your card (N.B. balance transfers and cash withdrawals do not generate donations).
plus
A **voucher** for a FREE HOTEL BREAK FOR 2 with a choice of 200 hotels (subject to availability) once you activate and spend on your card three times!
Exclusive offers and discounts with your account statements
- Accepted in over 24 million outlets worldwide
- No interest to pay for up to 50 days
- 0% interest on Balance Transfers for 6 months
- 24/7 access to cash at over 56,000 ATMs in Britain alone
- Typical APR 16.9% variable

Ask us for an application leaflet now.
Subject to status. Applicants for credit must be aged 18 or over and UK resident. The DAYS MasterCard is issued by Sygma Bank UK, Equipoint, Coventry Road, Birmingham B25 8FE

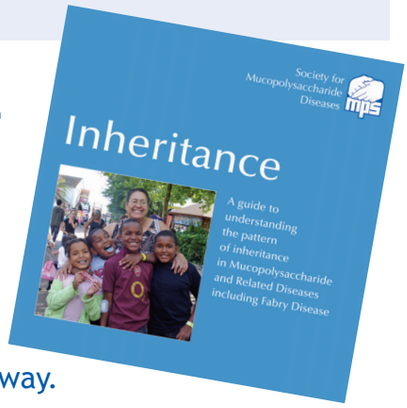
Rotating ELAP car seat and three-wheeled wheelchair available

One of our member families has a three-wheeled wheelchair, plus rain covers that came from America from babyjogger.com. It has been well used, is good over rough ground, but it doesn't fold down very small so is best transported in an estate type vehicle. The family also have a rotating ELAP car seat that can be fitted instead of the front seat in a Vauxhall Zafira. Although it has to be fitted professionally, it is brand new and saved a lot of back strain! For more information please contact the MPS Office.

Do you have a buggy which you could donate to a boy in Turkey suffering from Hunter Disease?

He is 1.3 metres tall. We are looking for a buggy that is fully reclinable as this little boy is unable to sit up.
We can arrange delivery.
If you think you can help, please phone the MPS Office on **0845 389 9901**.

New booklet out now!



Explains the Pattern of Inheritance in MPS and Related Diseases, including Fabry, in a simple, uncomplicated way.

The booklet costs **£2.00** (UK price inc. postage and packaging). To order please phone 0845 389 9901 or visit **www.mpssociety.co.uk**

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Society for
Mucopolysaccharide
Diseases

Become a Friend of MPS

Would you like to show your support by becoming a Friend of MPS? We would welcome relatives, friends, overseas MPS families, professionals or indeed anyone interested in the work of the Society or the field of MPS & Related Diseases. This would encourage us, help us plan for the future and bring about more public awareness for this group of rare, genetic, life-limiting diseases.

What are the benefits of becoming a Friend of MPS?

Membership number and card
Quarterly colour MPS magazine
Quarterly colour fundraising newsletter
Annual report and accounts
Regular publication updates
Information on and preferential rates at MPS events
Priority ordering of MPS Christmas cards

Phone 0845 389 9901
or download a form from
www.mpssociety.co.uk