MPS

Summer 2011

What's Inside...

Stories and experiences shared by our members

Clinical trial update, international news and a round up of clinics and MPS events

Society for Mucopolysaccharide Diseases

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

The MPS Society

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

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

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

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

MPS & Related Diseases

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

These multi-organ storage diseases cause progressive physical disability and in many cases, severe degenerative mental deterioration resulting in death in childhood.

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

Where does your money go?

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

Access to clinical management and palliative care

MPS Regional Specialist clinics

Support with disability benefits

Paving a child's way in accessing education

Upholding rights in employment

Advising on home adaptations

Bereavement support

Front cover photo: Emily Bradshaw (MPS I H).
Read Emily's story in our Members' News section

Society for Mucopolysaccharide Diseases

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Magazine Deadlines

Autumn	1 Sep 2011	Winter	1 Dec 2011
Spring	1 Mar 2012	Summer	1 Jun 2012

Friend of MPS

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

The articles in this magazine do not necessarily reflect the opinions of the MPS Society or its Management Committee. The MPS Society reserves the right to edit content as necessary. Products advertised in this newsletter are not necessarily endorsed by the Society.

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IN THIS ISSUE...

Summer 2011

Editor's Note: As you will read in the Chief Executive's Report overleaf, this edition of the MPS Magazine is being posted out earlier than usual as I will shortly be off on maternity leave. The fundraising magazine will posted out separately very soon. In the next MPS Magazine, Autumn 2011, we will be including the MPS Children's Newsletter as an insert in the middle pages of the MPS Magazine. This is to reduce printing costs but without economising on the support we provide to MPS children and their siblings. Thank you! **Antonia Anderson**

Once you have read this MPS Magazine, please pass it on to your family, friends and colleagues. *Help us spread the word about MPS and related diseases and the work we do.* ***www.mpssociety.co.uk***

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Chief Executive's Report

If this MPS Magazine feels a little lighter this time it is not that we are economising on our efforts communicating with our members. The reason is logistical, Antonia Anderson, the MPS Communications Officer is expecting a baby at the beginning of July and although Antonia plans to work up to her due date, just in case the baby has other ideas, we are belt and braces getting the Magazine out to you a little earlier. Whilst talking of babies, Sophie Thomas, the MPS Senior Advocacy Officer, is also expecting a baby at the end of June. I am sure you would want to join with me in wishing both Antonia and Sophie well for the safe arrival of their babies. Photos in the next MPS Magazine!

And there is more news from the MPS Office. I am delighted to welcome Steve Cotterell to the post of MPS Advocacy Officer. Now many of you may well think I know that name and probably you do! Steve was an Advocacy Officer with the MPS Society and left his post some three years ago to follow his dream of a career in catering and the food industry. Steve is an amazing cook and has thoroughly enjoyed his little venture but has decided his heart lies in the social care sector. With the departure of Tina Bough a couple of months ago, Steve successfully applied for the vacant post. Steve's wife, Sue Cotterell, has been with the Society over four years and been my PA for two of them serving the Society and I very well indeed. Sue is due to return from maternity leave in July after the birth of her and Steve's baby daughter Celeste, last November. Sue will be returning to the Society to take up the post of Trust and Corporate Fundraising Officer and Saskia Santos who covered Sue's maternity leave as my PA will be staying in the post.

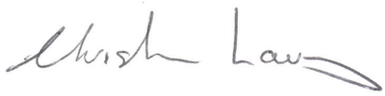
As the MPS Conference Weekend draws ever closer we have record numbers of families and professionals booked. We are also delighted that many of the thirty five delegates at the scientific MPS III Workshop being held immediately preceding the Conference will be joining us for the Weekend. This will certainly add an international flavour to our MPS Conference and provide families with a unique opportunity to talk directly with so many scientists working at the coal face seeking out new therapies for MPS diseases. We have a wonderful children's and vulnerable adult's care and social programme planned and a special Gala dinner as MPS enters its 30th Anniversary year.

There has been considerable activity on the Fabry front with two patient workshops hosted by the MPS Society held in London and Cambridge in May. For those in the North of England Dr Steven Waldek and his team are holding a Fabry patient meeting at Salford Royal Hospital on 18 September.

By the time you receive the MPS Magazine our MPS Awareness Day will have taken place at Whipsnade Zoo on Sunday 15 May. Many families have booked to celebrate this special day in the company of many amazing creatures. Keeping MPS and related diseases at the top of the agenda politically and from a societal perspective is a high priority for the Society but we can only do it with your help. If you want help with telling your story to the media we are here to help and support you. Whilst May 15 focusses our minds, awareness of MPS, Fabry and other related lysosomal storage diseases is a year-round task.

The de-designation of the paediatric LSD service at Addenbrookes Hospital, Cambridge, took place on 31 March 2011. We recognise that this has been an uncertain time for over 30 of our member families. As I write we hope that we will have more news as to where Consultant Paediatrician, Dr Uma Ramaswami from the de-designated LSD service at Cambridge will be moving to. We understand from all the communications from families affected that, despite there being excellent paediatric LSD services at Great Ormond Street, Birmingham Children's and Manchester Children's Hospital, many of you have expressed no desire for your child or children to have to get used to new doctors.

In these tough economic times the MPS Society is working hard to minimise the impact of losing funds through no longer being part of the Jeans for Genes Day this coming October 2011 and forever. Having devoted an incredible amount of the Society's time to ensure the success of 15 annual Jeans for Genes Days we did not want this any more than the two other partner charities affected, Great Ormond Street Children's Hospital (GOSH) and the Primary Immunodeficiency Association (PIA). The trademark holders, Chronic Granulomatous Disease Research Trust (CGDRT) clearly plan to continue with Jeans for Genes day this coming October and beyond so please make all your family members, friends and work colleagues aware that any funds raised or donated from now on through the trademark Jeans for Genes will not benefit the members of the MPS Society, GOSH or PIA. On a positive note thank you to the many energetic families who are using their time, skills and enthusiasm to raise funds or make donations. Without all your support the Society would not be able to offer the high level of support and advocacy that we do.



***Christine Lavery** Chief Executive*

Summer 2011

MPS GOVERNANCE

Dear Members
You will be aware that MPS is a registered charity and regulated by the Charity Commission. MPS is currently structured as an unincorporated association, governed by a set of Rules dating back to 25 March 1983.

As an unincorporated association, MPS has no legal identity of its own. Instead it acts through its individual trustees and it is those trustees who must enter into contractual arrangements on behalf of MPS.

The trustees of MPS have taken legal advice and, for a number of reasons, have decided to restructure MPS. Therefore, it is proposed that a new company limited by guarantee will be set up, with objects similar to those of MPS, and that this new company will take over the assets and undertaking of MPS, and continue its work going forward. The technical name for this process is incorporation.

The day to day work of MPS will not change and, to the outside world, it will be business as usual. The Charity Commission will be fully engaged in the process. The Charity Commission actually encourages the trustees of charities like MPS, which are currently unincorporated and which employ staff, own property and enter into contracts, to consider incorporation. Taking this step makes it easier for MPS to do business and, importantly, provides the trustees with the benefit of limited liability protection.

What will this mean for you as a member?
The process of incorporation provides MPS with a useful opportunity to review its rules and policies and ensure that the documents in place, governing its management and administration, are up to date, comply with recent changes in the law and best practice and reflect how MPS works in practice.

In relation to membership, it is proposed that a two-tier membership structure will be created. The formal, company law members, will be the trustees from time to time and it will be those persons who are expected to attend general meetings and vote on issues relating to MPS' governance.

The second tier of membership will be for the MPS Life Members. This tier of membership will be reserved exclusively for:
1. those affected by Mucopolysaccharide and related diseases;
2. the parents or, subject to the agreement of the Trustees, the carers of affected children and adults. (This will include bereaved parents or carers);
3. the partners of affected adults; and
4. any other persons, chosen by the Trustees, for their contribution to the work of MPS.

MPS Life Members will be those with an interest in the work of MPS and those who have been directly affected by it. MPS Life Members will be invited to attend conferences and meetings, and represent the organisation and be involved in its work. However, MPS Life Members will not be asked to vote on issues relating to MPS' governance. In recent years, very few members, barely enough to be quorate, have participated in the formal business of the AGM due to the nature of the MPS and related diseases, the caring responsibilities of parents and partners and distances involved in travelling. It is therefore suggested that the new structure is one which reflects the needs and interests of the majority of MPS members and the beneficiaries.

At the AGM this year, you will be asked to pass a resolution which will allow the current MPS, the unincorporated association, to dissolve in due course, following the transfer of its assets and business to the newly established company and registered charity. The following resolution will be put to the members: Pursuant to Rule 14 of the Rules of the Society for Mucopolysaccharide Diseases (the 'Society'), the Members of the Society hereby resolve that, following the transfer of its assets and business to the new company limited by guarantee of the same name, the Society shall be dissolved and removed from the Charity Commission's Register.

If you have any questions in advance of the AGM, please do contact in the first instance the Society's CEO, ***Christine Lavery, c.lavery@mpssociety.co.uk***

Could you be our next MPS Trustee?

Could you be our next MPS Trustee? We are actively identifying potential new Trustees to offer their skills to help guide the work of the Society for Mucopolysaccharide Diseases. One of the skill bases we are particularly looking for is a legal background. If you would like to know more about becoming a Trustee of our growing charity and help to make a difference please email **c.lavery@mpssociety.co.uk**

MPS Annual Report and Accounts

The MPS Society annual report and accounts for 2010 are available to download from our website, ***www.mpssociety.co.uk****. If you require a hard copy, please request this by emailing accounts@mpssociety.co.uk. Please note, for this we will charge a fee of £3 to cover costs.*

News from the MPS office

Re-introducing Steve Cotterell...

Hello everyone I'm back! After three years away I have returned to the MPS Society as an Advocacy Officer. I am looking forward to catching up with some familiar faces as well as meeting new members. I remember my time here well and I am keen to get back to it!

This time my main areas of responsibility are to support those affected by MPS III, MLD, AGU, Winchester, Sly, Sialic Acid Disease, Gangliosidosis and Geleo Physic Dysplasia. So, if there is something that you need or would like some support do contact me.
Steve Cotterell, ***steve.cotterell@mpssociety.co.uk***

NO MORE RESEARCH AND SUPPORT MONEY FROM JEANS FOR GENES

On 31st March 2011 the partnership agreement between the MPS Society, the Primary Immunodeficiency Association (PIA), Great Ormond Street Children's Charity (GOSHCC) and the Chronic Granulomatous Disease Research Trust (CGDRT) came to an end. The partnership has been together for 15 years and it was immensely disappointing that the trademark holders CGDRT have withdrawn the use of the Jeans for Genes trademark and plan to run Jeans for Genes for themselves.

The Trademark holder, CGDRT, initially offered MPS, PIA and GOSHCC a three year transition agreement that meant we would each receive a small percentage of the income raised from the 2011, 2012 and 2013 campaigns but at the eleventh hour just as the agreement was about to be signed CGDRT, the trademark holder, withdrew the offer of the transition agreement.

This means MPS has to find new avenues to raise significant funds for research and advocacy support. Not an easy task in this financial climate. If, as an MPS family or supporter you, your child's school, business or friends have raised funds for Jeans for Genes in the past you may like to tell them that ***MPS DOES NOT BENEFIT FROM JEANS FOR GENES ANY MORE***. Clearly we need to find new sources to raise the funds we used to receive from Jeans for Genes so if you, your friends, your business or your child's school wants to support MPS please let us know.

They may also be reassured to know that compared to Jeans for Genes, where in excess of 75p in the pound has been expended in administrative costs, the MPS Society spends 15p in the pound administering the Society for our members. Please spread the word and think and act for MPS.

Jeans for Genes Database
All four of the partner charities have been given a copy of the Jeans for Genes database of people and organisations that have supported the cause over the last fifteen years. To make use of this data, the law requires that where this data is not in the public domain, each charity, MPS, PIA, GOSHCC and CGDRT must validate its use by writing to each individual. It is not permissable to use email or telephone to validate the database.

A validation email was sent to everyone on the Jeans for Genes database prior to the break up of the partnership on 31 March 2011. This email provided an opt in by the recipient agreeing to be contacted in the future by any or all of the ex Jeans for Genes partner charities, MPS, GOSH, PIA and the trademark holders CGDRT. If you, your family, friends and colleagues did not receive the opt in email or did not agree to be contacted by the Chronic Granlulomatous Disease Research Trust (GDDRT) or anyone in the name of Jeans for Genes and are subsequently contacted by by the Chronic Granulomatous Disease Research Trust (GDDRT) or anyone in the name of Jeans for Genes please do let MPS know.

Advocacy work in Southern Ireland

On 9th April I was delighted to attend an Irish MPS Society family get-together in Maynooth. This was my first chance to meet with families in Southern Ireland in a social setting, so I was more than happy to make the trip to Maynooth and spend some time discussing my role. We had a lovely lunch and there was lots of time to chat and get to know each other. The children were entertained by 'Silly Sally' (a very energetic clown!) which gave all the adults time to relax and ask questions about the type of support that can be provided through the MPS Advocacy Support Service. Since this event I have been contacted by several families seeking support and advice.

Soon after this event Christine Lavery and I spent a week in Southern Ireland. The purpose of this trip was to meet with medical professionals and the Irish MPS Society to discuss the progress of the Southern Ireland service, to meet families enrolled on clinical trials and to hold 'meet and greet' evenings across Ireland. This was an excellent week! We drove 800 miles, saw the sights of Dublin, Cork and Galway, met many lovely families, and importantly identified many areas of support. Please look out for an extended article on this in the next magazine.

Alison Wilson
a.wilson@mpssociety.co.uk

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

New service offered by the MPS Society to those affected by Metachromatic Leukodystrophy

The Society for Mucopolysaccharide Diseases continues to welcome members with Metachromatic Leukodystrophy (MLD). We invite anyone who is affected by this disease, or professionals working with those affected, to contact us.

We have an Advocacy Support Officer who supports individuals and their families affected by MLD and so this does not affect the current service already provided by the MPS Advocacy Team to those affected by MPS and Related Diseases.

We have developed a Guide to Understanding MLD fact sheet which is available from the MPS office, or soon to be downloadable from the MPS website, plus a range of other information resources which cover issues related to those affected and their carers.

For further information please contact the MPS Advocacy Team by phone on **0845 389 9901** or email **advocacy@mpssociety.co.uk**

WHAT'S ON!

CONFERENCE EVENTS

MPS National Weekend Conference
24 - 26 June 2011

International Symposium, The Netherlands
28 June - 1 July 2012

SPECIAL EVENTS

Sibling Week
25 - 29 July 2011

Lapland Christmas Visit
11 - 14 December 2011

MPS REGIONAL CLINICS

Birmingham clinic: 17 June, 18 November
Bone Marrow Transplant clinic (under 6's): 29 July, 14 October
Bone Marrow Transplant clinic (over 6's): 22 July, 21 October
Bone Marrow Transplant Teenage Transition clinic: 17 June
MPS III Clinic: 10 June

MPS 2011 Annual General Meeting

The 2011 Annual General Meeting of the Society for Mucopolysaccharide Diseases will be held at the Hilton Northampton Hotel at 7.30pm on Friday 24 June 2011.

If you are interested in becoming a Trustee of the MPS Society please contact the MPS office. We would particularly like to hear from any MPS Society members living in Northern Ireland as well as other parts of the UK.

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Please let us know if you're not able to attend an event for any reason

The MPS Society is delighted to offer subsidised places at a number of events throughout the year. If you book a place for yourself, and/or your family, but later find that you are unable to attend the event after all, please do let us know. Sometimes we are able to offer your place(s) to other members and it means that MPS staff organising and attending the event aren't left waiting for you to arrive. We completely understand that sometimes it is unavoidable and last minute emergencies do crop up, but we would be very grateful if you could let us know by phoning the MPS office, out of hours number, or the contact number given to you on the event information sheet. Thank you!

ANNOUNCEMENTS

New Members

Mr and Mrs Vickers have recently been in contact with the Society. Leon has a diagnosis of Sanfilippo disease. Leon is three years old and the family live in the North East.

Mr and Mrs Desai have recently been in contact with the Society. Their little girl has been diagnosed with Morquio disease and is 8 years old.

Mr and Mrs Brooker have recently been in contact with the Society. Lily has a diagnosis of Sanfilippo disease. Lily is four years old and the family live in Kent.

Births

Congratulations to **Joanna and Alex Wilson-Smale** on the safe arrival of their daughter, Abigail Rose, weighing 6lb 6oz, on Friday 15th April 2011.

Congratulations to the **Silcock family**. Bobby (MPS III) is now a big brother! Thomas Robert Patrick Gill (Tommy) was born on 17th March 2011 weighing 8lb exactly. Bobby is loving his new role!

Become a Friend of MPS

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

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

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

Dear Friends of MPS

Our Friend of MPS subscription is an annual subscription. Every quarter, our Friends receive the MPS Magazine and Fundraising Magazine plus a range of other information on the services we provide and events we are organising.

When your Friend of MPS subscription is due for renewal, we will enclose a renewal form in your magazine mailout. If we do not hear back from you, we give you one more opportunity to renew your Friend subscription as we realise that you may have forgotten or your personal circumstances may have changed. Friends of MPS who do not renew their annual subscription are removed from our database as Friends, and are instead placed on our Fundraiser database. This means that you will only receive our Fundraising Magazine. You will no longer receive the MPS Magazine, nor information about events and support activities that we offer.

So, please don't forget to renew your Friend of MPS subscription when the time comes. These subcriptions are also vital in contributing to the funds we receive to continue our work. ***We need our Friends!***

In Memory

Jasmin May Heap

2nd October 1998 - 1st March 2011

Our precious daughter Jasmin (pictured left) gained her angel wings at the tender age of 12 years, taken from us by Sanfilippo syndrome.

Jasmin is our only, most precious child and we love and miss her so much. We are totally broken hearted and lost without her. We have the greatest memories of our brave, funny and smiley princess, which we shall treasure forever. 'Life is fragile - embrace it.'

Below is the poem that I (Ria, proud Mummy) wrote and read at her funeral.

With all our hearts, Mummy & Daddy - **Dave and Ria Heap**

Jasmin May Heap entered this world with great pace
5 weeks early, she brightened earth with her beautiful face

Her surprise arrival for us was a huge jolt
She hit our hearts like a raging thunderbolt

Our new role as parents began with speed
With Jazzy at the forefront taking the lead

Jaz lived like it was a race, she never slowed down
Forever with a beaming smile, but never a frown

Our vision was always filled with her wide grin
Our ears heard her constant laughter and giggling

Happiness and love oozed from our precious Jasmin
She laughed when people cried, like it was their sin

Our little girl radiated love and was beauty personified
She gave us so much pleasure, from her birth until she died

Our darling Jasmin passed away with a smile on her face
Her last breath in our arms taken with elegance and grace

Jasmin lived life so literally and gave us unconditional love
We now don't have her physically but feel her affection from above

The time is all too soon to say our Goodbye and bid her farewell
Our love for Jasmin May will never fade, she'll always make our hearts swell

Jasmin left us with a wealth of memories to remember her by
As now she's gained her angel wings and flies free in the sky.

Photos below: Left - Jasmin and Daddy, Right - Jasmin and Mummy

In support of Rare Disease Day in February 2011, Christine Lavery, the MPS Society's Chief Executive, was interviewed for the Independent. Here we reproduce the article...

Creating the road ahead

Lysosomal storage diseases (LSDs) are a group of approximately 50 rare inherited metabolic disorders that are the result of defects in the function of the lysosome, a specific organelle in the body's cells.

Mucopolysaccharide (MPS) and related diseases are a sub-group of lysosomal storage diseases – and their most common form. As Christine Lavery, Chief Executive of the MPS Society, explains, each MPS disease is caused by a missing or deficient enzyme. As a result the body is unable to breakdown storage material causing progressive physical disability, and in some diseases progressive neurological deterioration.

Their treatment brings many challenges – fought every day by organisations such as the MPS Society and researchers and clinicians on the front line. "The main challenge of LSDs is establishing sufficient awareness of these conditions so that, despite their rarity, they can be appropriately considered and diagnosed," explains Dr Derralynn Hughes, senior lecturer and honorary consultant haematologist at the Royal Free Hospital NHS Foundation Trust, London.

"It can take many years and visits to many different specialists to get the diagnosis - patients are necessarily treated in specialist centres which means that there needs to be very good communication with local doctors, who may have no experience of the condition, so that problems while they are at home can be managed quickly and correctly," says Dr Hughes. "Treatment is available for some, but not all conditions. This is either because of their rarity or because the manifestations are difficult to treat – for instance, they occur in a part of the body which is not easily accessible to the current modalities of therapy, such as enzyme."

Paving the way

"While the MPS Society has forged ahead with health and social care for LSDs, the hope now is that this model could be translational to other rare diseases", says Lavery. But there's still more work to be done. "It's vitally important to raise awareness – and the goal that still has to be worked towards is better practical support for families," she says. "My son Simon was born in 1974 and diagnosed with Hunter disease, an MPS condition, and died at the age of seven. We set up the Society to talk to other parents – while there is more information available than previously, people will be always want to be in touch with others in the same situation."

Transition to adult care

The MPS Society prides itself on its advocacy service, which, beyond campaigning, provides full-service support for its members, encompassing both psychological and practical support. One of the areas lacking in MPS care, says Lavery, is transitional care. "Paediatric services tend to be second to none but adult services have a number of challenges – the parents who have cared for children and know their disease and needs intimately are then confronted by a number of different problems in care as the child gets older," she points out. Issues faced range from problems with accessibility and administrative delays to time constraints and less specialist knowledge. These problems can be distressing, as well as disruptive to care. "In a progressive disease, it's crucial to be able to refer back to a previous notes and scans, for instance", Christine Lavery points out.

"We need to promote research into new, effective and affordable therapies for the currently untreatable manifestations of the conditions, such as those affecting predominantly the brain," says Dr Hughes. "We need to raise awareness of rare conditions in general so that diagnoses are made in a timely way."

Originally distributed by Mediaplanet within The Independent on 28th February 2011.

Thank you to all our supporters!

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

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

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

Fundraising resources available from MPS

Fundraising packs; School fundraising packs; Fundraising fact sheets; Sample press release; Sponsorship forms; Become a Friend of MPS; Merchandise Order Form; Publication Order Form; T-shirts, posters, balloons, plus more...

Please visit **www.mpssociety.co.uk**

MPS Awareness Day
15 May 2011

Each year the Society celebrates International MPS Awareness Day on 15 May. This is a day devoted to raising awareness of MPS and Related Diseases. In 2011 we're asking all our members, Friends and supporters to do something, big or small, to mark MPS Awareness Day.

The MPS Society held its own Awareness Day celebrations at Whipsnade Zoo on Sunday 15 May. We were delighted that over 150 children and adults were able to join us on this day!

MPS families were invited to arrive at 10.30am to collect their programmes for the day, zoo map and fun quiz before heading off into the Zoo for a couple of hours looking at the animals.

We all met up again in the party room at 12.30pm to listen to speeches by Barry Wilson, Chairman of MPS Trustees, and James Garthwaite, father of Tom and Louis who both have MPS.

Both Barry and James gave wonderful speeches explaining the significance of MPS Awareness Day and highlighting the work done by the Society over the last 30 years. James and his family very kindly cut our Awareness Day celebration cake and lunch was then served!

Lunchtime gave the opportunity for families to chat, swap stories, meet old friends and make new ones. We were delighted to welcome so many of you, some who have attended many MPS events, and others who have come to us only recently.

There was plenty of noise, fun and popping of balloons as the parents chatted and kids ran around (and the parents chased after them!).

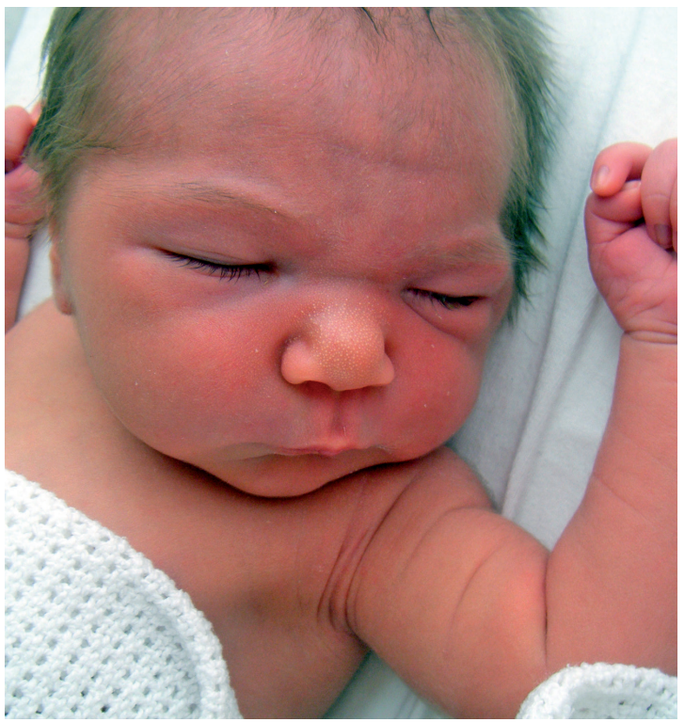
As lunch drew to a close, many families chose to have a bit more of a wander around the Zoo to see the rest of the animals. The more energetic amongst you chose to walk and I think others chose the car safari option!

We hope you had a fantastic time and thank you for joining in our awareness celebrations for this year! Here are a selection of our photos from the day. Please do send in your photos or a story about your family's day out to newsletter@mpssociety.co.uk so that we can publish them in the next edition!

We would like to take this opportunity to thank Shire, Genzyme and BioMarin for their support of this event.

Antonia Anderson a.anderson@mpssociety.co.uk

Miya's story



I am originally from Canada and my husband is British. I came to England to live in 2008, when I married my husband Chris. When we discovered our pregnancy in late 2009, we were ecstatic! Having had a stillborn child in 2000 (of a heart condition unrelated to MPS) I was nervous about the pregnancy but everything progressed smoothly. Miya was born on 07-07-10 in Ipswich at 38 weeks gestation. She was born with her face presenting, so I wasn't alarmed at the slightly "squished" look to her face, particularly her nose.

Miya began showing signs something wasn't right at about two weeks of age. She wouldn't stop crying and she suffered terribly from "silent reflux". Her breathing was noisy and I felt something wasn't "right" about the way she looked. Her face, particularly her eyes, forehead, and nose, struck me as being slightly... off. Her head was quite large and he neck was quite short. Her blue eyes were beginning to turn a cloudy grey colour. She was hitting her developmental milestones, but slightly later than other children. She was very floppy and her joints seemed to crack excessively. I took her to our GP about her noisy breathing and he advised she was young and her respiratory system was immature and just to keep an eye on it.

Miya suffers from talipes in her right foot, and our Orthopaedic doctor at Southampton Hospital felt that perhaps the talipes was a symptom of something greater and referred us to a paediatrician for testing. He was the only medical professional / child professional we saw (including health visitors, GPs, infant cranial oesteopath) who had the courage to say they thought something else was going on with Miya's health. (We later asked some of these medical professionals if they thought something was "wrong" and they advised yes, they thought they did, but didn't want to "hurt our feelings" or "overstep boundaries").

Miya was diagnosed with MPS I on 28 Feb 2011. She promptly saw Dr. Vellodi at GOSH and is now one week into weekly ERT treatments. Her dad and I have both had our blood tested for tissue typing for a BMT, but that was only last week so no results are back yet. Miya will be undergoing a BMT, but she has 12 scheduled ERTs to go through first. We're not sure if she will have to go through more or if she'll then be ready for her BMT.

Photo top left: Miya at just a few day's old. Photo above: Miya receiving ERT at Great Ormond Street aged 8 months

On these pages our members share their personal experiences of life with an MPS or related disease...

Chris and I, as parents, are trying to stay strong. Chris is a serving soldier in the British Army, having served so far for nearly 12 years. The army has been tremendously helpful in making sure Chris has time off to drive us to GOSH for our weekly visits, and ensuring that we are posted in the South West so we can be close to GOSH. My family are all in Canada, and Chris' family are all in Cumbria so we are a little "island" of sorts. The MPS Society have been very helpful in sending us supportive information and offering to put us in touch with other families who are touched by MPS.

I know it's not a long story; we've only been dealing with the specific diagnosis a short while. However, if our story can help someone else or bring awareness to MPS in some way, we would be glad to share it with you.
Sarah, Chris and Miya Hulse

The Lever family's experience of support from the MPS Society

In 2006 our four year old daughter, Aaryanna, was diagnosed with MPS I Hurler Scheie. A light went out in our lives the day we got that diagnosis. It was replaced with overwhelming fear for us and all our futures. Five years of Enzyme Replacement Therapy (ERT) have now elapsed and our daughter is thriving. Our futures are once again bright and filled with hopes and dreams. We are normal.

Our journey was horrendous. Our despair was all-consuming. Part of our recovery, and a very large part, is attributable to the MPS Society. They provided us with the information, support and help we needed during our darkest days. Their service is so comprehensive, understanding, professional and caring. We know we matter to them. We feel it. We no longer fear MPS. We have welcomed it and accepted it into our family. We did this with the love and help of the MPS Society. MPS is with us and our daughter for life. The MPS Society is also with us for life.
Helen Lever

Photo above: Miya at 6 months old

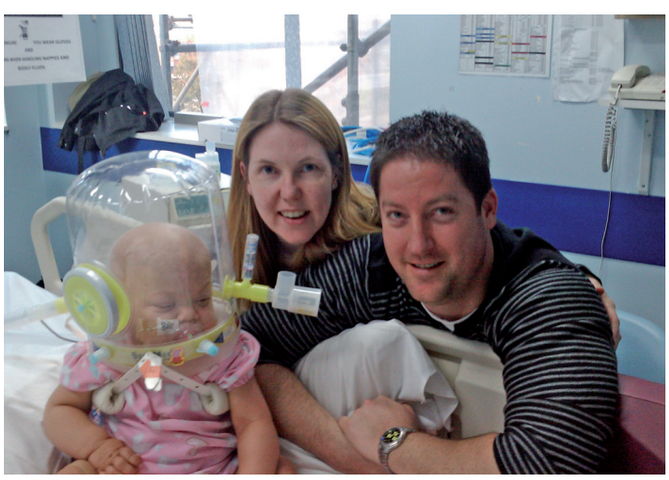
Do you have a story to share?
Please email
newsletter@mpssociety.co.uk
or phone **0845 389 9901**

Emily's story

Emily Jane Bradshaw was born on 25th July 2008 to two very happy and proud parents, Nev and Gayle Bradshaw. When she was born the standard new baby screening tests were used to assess how Emily was. All was fine except that they picked up on a hearing problem with her the day after she was born. She was then referred to the Audiology department at University Hospital North Staffs who, after several hearing tests, determined that Emily had glue ear and also a moderate hearing loss. At 6 months old Emily was admitted to North Staffs to have grommets fitted to drain the excess fluid in her ear canal. After this she was then prescribed hearing aids to wear – lovely pink and glittery ones. It took at least 6 months for her to get used to them as she kept pulling them out and putting them in her mouth!

Emily was 1 by this time and although she wasn't doing everything a 1 year should have been doing we were not overly concerned as we thought due to her hearing problem she was slightly delayed in her progress. By 18 months old Emily was walking around the furniture and with the use of her walker but not of her own accord.

She wasn't saying many words and the doctor at our local GP's referred her to the Paediatrician at North Staffs, Dr Reynolds. We had our appointment on May 20th 2010, Emily was 22 months old. This was the day we will never forget as on this day we were first introduced to MPS and consequently Emily's condition of MPS I Hurlers. This was our darkest day as we were not expecting anything so severe to come out of the appointment. Emily was taken to see Dr Reynolds by her mum and Nan - Susan Williams – as Emily went to her Nanny's to be looked after when her mum was at work. We thought the appointment would advise as to what could be done to improve Emily's communication and mobility and it made sense for Susan to be present. Dr Reynolds examined Emily and did a few puzzles with her before presenting the bombshell – Emily has a condition, mucopolysaccharidosis. At this point we were advised that Emily wouldn't live to adulthood. Our world had now turned upside down. After this we had to wait several weeks for the results of her blood and urine tests to determine what MPS disease she had.



In mid June we had the appointment through to go to Birmingham Children's Hospital where we were to meet with Dr Chaprakani and Catherine Stewart. It was an appointment we were never going to forget as this was our turning point. Although Dr Chaprakani did confirm our worst fears that Emily had MPS I Hurlers he then surprised us by advising that something could be done and all was not lost. He advised us that Emily could have a bone marrow transplant to improve her quality of life and life expectancy itself. He also advised she could receive ERT up until the transplant. We left this appointment with hope in our hearts – our little girl will be with us for much longer than we had first feared. We spoke to all of our family and it was unanimous that we should give Emily the opportunity by having a bone marrow transplant. Once we made the decision everything moved really fast – within a few days we were back in Birmingham so Emily could have tests and also her first ERT. We then went back to Birmingham every week so that she could have ERT up until her transplant – which was scheduled for 14th September. As the weeks went on we noticed how Emily began to improve in her mobility and by August she was finally able to walk on her own (somewhat unsteady).

On 6th September Emily was admitted to BCH where, along with her mum, she was to stay for nearly 4 months. Unfortunately Emily's first transplant didn't take so on 27th October she was given a second one. It had been a rocky road up until that point as she had suffered with the affects of the chemotherapy and had been having breathing problems where she had ended up in PICU with a CPAP hood on.

Just under two weeks after the second transplant Emily's neutrophils started to pick up – we were so excited it was just what we had been waiting for – after all this time being stuck in HDU we were finally seeing the signs that Emily was getting better. Little did we know that this was far from the truth and a week later Emily was back in PICU after a rather horrific Friday night where, after a week of requiring more oxygen, she finally succumbed to the struggle and had a respiratory arrest. The nurses and doctors were fantastic and did so much for Emily.

She was rushed to PICU where she was sedated and on a ventilator for two weeks. In this time the fluid that had built up in her lungs started to reduce and her oxygen requirements via the ventilator started to reduce also. By late November Emily was taken off the ventilator and was awake. It was such a relief that she had made it through – our little girl was so strong and brave.

It took another month before Emily was well enough to go home – even though her neutrophils were picking up and the transplant was starting to work her time in PICU had delayed her recovery. She had been on so much morphine that it took two weeks for her to get it out of her system. It must be said that the other thing that helped Emily through her entire time in hospital was Peppa Pig!

We had 10 DVD's and she watched them constantly. It was her escape, even the nurses got to know what Emily's favourite programme was.

We finally came home on 18th December – it was fantastic – to finally arrive outside our house that we hadn't seen since September, covered in snow, a week before Christmas. Emily even recognised the house as when we pulled up she said "Yay" and had a big smile on her face.

Since getting home Emily's progress has been amazing. During January she was starting to get her strength back. Due to her low immune system she didn't go anywhere except to BCH for her Viagam and appointments with the Oncology clinic. By mid February Emily was back at her Nanny's whilst her mum had to go back to work and since then she has come on in leaps and bounds. She can now walk on her own, she isn't able to run yet but she is definitely trying. She is also saying a few more words – Ooze for shoes, tree, car, doggy, which we think is fantastic. She understands so much, she just struggles to communicate. Her hearing has also improved – whereas before she could only hear 40db and above she can now hear as low as 30db. She still wears her hearing aids as they will help when she is learning to pronounce words.

Our life since last May has been a roller coaster of emotions and events – we have had a few lows but more recently the highs outweigh this. Emily is such a beautiful, happy and affectionate little girl. You would never know what she had been through (except for her appearance – her hair is growing back lovely), she's so animated and still loves Peppa Pig (mum thought we may have seen the last of this as she definitely overdosed on it in hospital).

We know how lucky we are to have Emily and also appreciate how different things would have been if the treatment was not available for her. We know we still have a long way to go with her but every day is a blessing and we would never change any of it. We find it so encouraging to hear of other MPS IH children who are doing so well and are in school, college, etc. This gives us so much hope and we really appreciate all the work that the MPS Society does.

Emily has an appointment in May to go to the Peter Pan Nursery in Newcastle under Lyme which is a special needs nursery. This will hopefully help her to catch up with other children of a similar age in anticipation for her starting school when she is 4.

Emily's dad, Nev, has produced a website which we will update as and when necessary charting Emily's progress. It details Emily's life so far and we hope that others who have been presented with the same condition will find it encouraging that even in your darkest day there is hope. www.emily-bradshaw.co.uk. **Gayle Bradshaw**

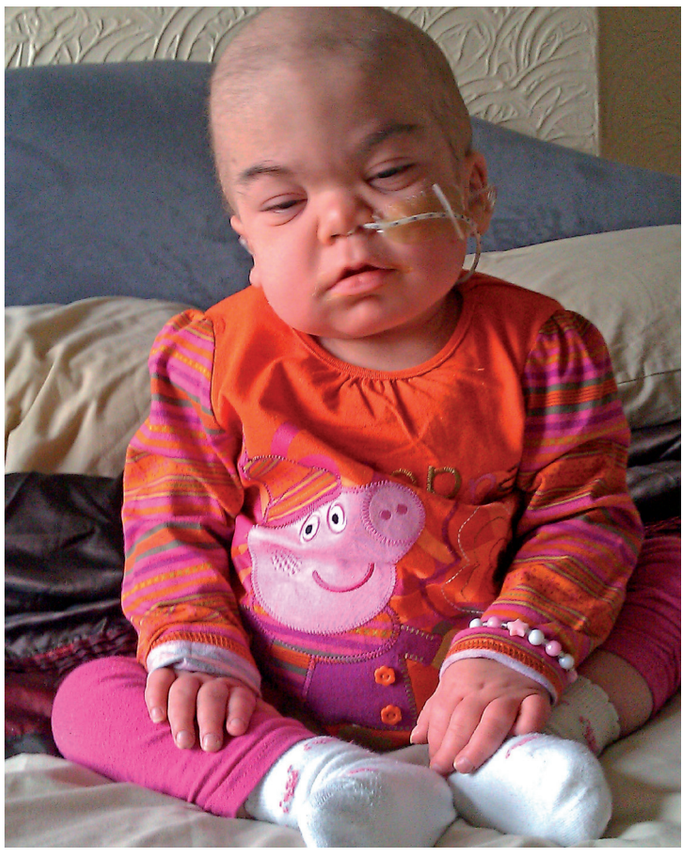


Photo page left: 24 October just before second transplant.
Photo this page top: back home; photo this page bottom: With Daddy, taken 25 April 2011

BMT Clinic for over 6's

Jolanta and I set out early from Buckinghamshire on April Fools day to attend the over 6 years Manchester Bone Marrow Transplant (BMT) clinic. Dawn was breaking and I had a flashback to how dark the morning was on my last visit to Manchester Hospital.

As usual the Team were so very welcoming and warm, the atmosphere vibrated with gentleness, encouragement and merriment.

Our first two visitors were a budding musician and has so much faith in his Daddy he informs everyone that Daddy can fix any musical instrument and a young man with his new baby sister.

The waiting room filled and emptied with gorgeous characters. A young lady who loves being creative and rejoices in glitter; a young lady with an infectious giggle when tickled and who made her Daddy a lovely cup of tea; a young lady looking forward to a holiday the following week; a young lady looking forward to a play visit the following day when she would play with her friend's bunny; a young man who relished tower building with his friend and made us all grin when he fired her in Alan Sugar's style; and a very independent young lady who happily walked off to see the medical team leaving Mummy and Daddy in the waiting room.

I would like to say a big thank you to the Manchester Team, thank you for all your dedication and hard work and for making the clinic such a welcoming place to be.
Lindsey Wingate l.wingate@mpssociety.co.uk

Photos clockwise from top left: Steven O'Reilly with his brother, Melissa McKie, Demi-Leigh Rodden, Holly Campbell with her Mum, Charlie Escalonilla, Rachel Rothwell.

BMT Clinic for under 6's

This time there was a two week gap between the clinics for those over 6 years old, held on 1 April 2011 and the under 6 year old clinic held on 15 April 2011. Although I got up early in the morning to catch the train I couldn't wait to meet the 'little' ones and their parents.

When I arrived I learnt that some families had cancelled for various reasons but my disappointment didn't last long. There were still some children coming and I didn't have to wait long.

First one to appear was Ethan, who came together with his parents and a lovely nana. Ethan was chatting a lot, asking many questions and tried to climb every climbable thing in the waiting area. In the meantime Harvey arrived with his parents. It didn't take long for both boys to start playing together. As Harvey walked into the waiting area, he looked at Ethan and shouted 'FRIENDS!'

I enjoyed greatly playing with Ethan and Harvey, chatting with their families and the Manchester team. I look forward to seeing you all again in the future.
Jolanta Turz j.turz@mpssociety.co.uk

Photos clockwise from top left: Over 6's BMT clinic - Cody Taylor.
Under 6's BMT clinic- Ethan Greening with his Nan, Harvey Houghton

Newcastle Clinic

21 April 2011 saw my first Newcastle clinic and I was to be up with the birds! I arrived at the Great North Children's Hospital as it is now called but everyone still refers to it as the RVI (Royal Victoria Infirmary). The Childrens' Outpatients is in the new part of the hospital and it is still very shiny and new.

Dr Rylance was ready and waiting on the starting blocks. Laura and Jessica arrived with mum, Dr Rylance making both the girls giggle by his teasing. Mum was saying how Laura had been snowed in at her residential college in the bad weather, but was of course continuing to work hard watching DVD's etc… Jessica likes to cook and the family have benefited from her culinary skills.

Next to zoom into the department was Leon with mum, dad and grandma. He was very content to play with the toys and took a shine to the doll's house and chat on the internal phone…. oops!

Daniel arrived with mum; he had been poorly and had to be an in-patient for a week. He was a little tired but was hoping to go home that day, which I think mum and Daniel were both looking forward to.

Luke, his brother and mum and dad arrived. It was short and sweet with the family as they were soon called for their appointment.

Callum, came in, together with Daniel, (Daniel is Callum's bear who has been sworn in as a scout at the same time as Callum) and mum and dad were also in tow.

Callum was not too impressed with the early start but soon cheered up after dad was comparing himself with yogi bear. Callum, you should know that yogi bear is better looking… only joking dad!

Soon it was time to say goodbye and make my way back home. Thank you to all the families and to Dr Rylance, Professor Wraith and the team for making me welcome. I look forward to seeing you all in the future.
Rebecca Brandon r.brandon@mpssociety.co.uk

Photos left to right: Laura and Jessica Fish (both MPS IVB), Daniel Muers (MPS II), Luke Chapman (MPS III) and his brother

Adult Fabry Clinic

The one thing about working for the MPS Society is that you do tend to see a lot of motorways. It was the M40 this time to Birmingham for the Fabry Clinic on Thursday 7 April at Selly Oak Hospital in Birmingham.

Dr Hiwot had very kindly invited the MPS Society to attend the adult Fabry clinic. Fabry is one of the areas of work for which I have a responsibility. Being relatively new to the job I was keen to learn more about Fabry and talking to the patients is a great way to learn about the diversity of the disease.

I arrived at lunchtime and the patients, myself, Dr Hiwot and his team all shared an enjoyable lunch together. I don't think we were too popular with other clinicians as we had taken over their staff room. Kate bribed them with the promise of pens and maybe what was left of the spoils, it's amazing how easily you can get your own way with a bit of bribery, but pens and food…

Birmingham has just launched the LSD (Lysosomal Storage Disorders) Adult Service and the aim is to provide a holistic, inclusive and multidisciplinary service for adults with lysosomal disorders.

Dr Hiwot was very keen to hear how the patients felt about the way the clinics were run and whether there had been any difficulties with the round of tests that they had already had the day before.

There were a couple of new members and I had the opportunity to explain about what the Society does and how the advocacy team can help members.

Before we knew it, it was time to start the clinic and Dr Lipkin and Dr Steele joined the team. Several more patients arrived and everyone chatted in between their tests and talking to the doctors.

The time flew and soon I had to make my way back home. I am looking forward to meeting everyone again at the next clinic.

Thank you to all the patients and Dr Hiwot and his team for making me feel welcome.

Rebecca Brandon r.brandon@mpssociety.co.uk

FIN - FABRY INTERNATIONAL NETWORK

The Mission of FIN is to be a global, independent network of Fabry patient associations whose purpose is to collaborate, communicate and promote best practice to support those affected by Fabry disease. FIN's vision is of a world where every person affected by Fabry disease has the best quality of life possible through early diagnosis, treatment and cure.

With over 250 men, women and children clinically affected by Fabry being members of the MPS Society I was honoured to be interviewed for the FIN Board of Directors and even more honoured to be appointed. I have participated in two Board Meetings to date so am very much the 'new girl on the block'. I hope in time, as I learn the ropes, to be able to support FIN to continue to make a real difference in the lives of those affected by Fabry disease globally. **Christine Lavery**

Help us care for today and give hope for tomorrow, leave a gift in your Will

It is vital that the MPS Society has sufficient funding to be able to look forward to the future with confidence. One way in which you can support the Society achieve its long term objectives is to include the Society when drawing up your Will. For more information please contact us for our Leaving a Legacy leaflet or for more information please visit **www.mpssociety.co.uk**

European MPS Network representatives visit Shire's Lexington Site, Cambridge, Boston, USA

The primary goal of this visit, 6-7 May 2011, was to afford a small group of representatives from the MPS Network an insight into what is involved in bringing new therapies for MPS and related diseases to market and the manufacturing challenges. A number of representatives of MPS patient organisations were invited and Freddie Wiesbauer (Switzerland), Michaela Weigl (Austria), Delphine Genevaz (France) and Hanka Meutgeert and I were able to participate. During the meeting and on behalf of the MPS Network I gave a brief outline of the history of the International MPS Network, how it governs itself and future objects.

We started with a welcome and introductions before going into a round table discussion during which Ken O'Reilly and Sue Bruhn, Senior Vice President Strategic Planning and Program Management for Shire Human Genetic Therapies, outlined the present and future of Shire HGT in relation to the MPS / LSD diseases. Some of the biggest challenges facing any pharma company working in the field of rare diseases are:

- the paucity of knowledge of the disease natural history
- small patient numbers
- the running of clinical studies slowed down by the time it takes to work out appropriate outcome measures
- Requirement for global regulatory strategies

Sue Bruhn highlighted that there are 7,000 plus orphan genetic diseases collectively affecting 1:10 of the population of North America. 25 million Americans are affected by rare diseases and 25-30 million Europeans. More than 50% are paediatric diseases and in the Lysosomal Storage Diseases CNS involvement is present in 54% of all LSDs. Moving onto the future business commitments, Shire HGT confirmed that their core focus is Fabry, Gaucher and Hunter disease. They also confirmed in their new capacity programme they are focusing on an Intrathecal platform involving clinical study programme for Metachromatic Leukodystrophy, Hunter CNS and Sanfilippo A. Future build includes Duchenne Muscular Dystrophy and enabling new therapies. Shire HGT are committed to research into unmet medical need and to staying in the rare disease space and have no plans to go into cancer.

After lunch Arthur Tzianabos gave us an insight into where Shire HGT are with the MPS Intrathecal programmes. The Hunter CNS programme is designed to deliver enzyme directly to the brain via the cerebrospinal fluid and a marketed intrathecal device known as a port-a-cath placed under the skin near the ribs. From the Port-a-cath a catheter is fed into the intrathecal space in the lumbar region. The Phase I/II dosing regime is 10mg/kg, 30mg/kg and 120mg/kg. Arthur told us that in MPS II, animal models being treated intrathecally achieved deep penetration into the brain even into the white matter. This offers a lot of hope that enzyme will get into the brain in the human studies.

The Sanfilippo A Programme dosing regime is slightly different and involves two doses,10mg/kg and 45mg/kg and different times between infusions. We are already seeing good safety data in the children currently in the clinical trial study. Sanfilippo A brain disease is in the grey matter. In animal models good penetration into grey matter of the brain has been seen. What we don't know is whether there will be a positive impact on cognitive development.

The Sanfilippo B Programme appears to be at least 2 – 3 years behind the Sanfilippo A Programme not least because the enzyme is more challenging because it is missing the Mannose-6-Phosphate receptor. This is a significant challenge that Shire HGT is addressing and looking at a whole panel of approaches.

The Metachromatic Leukodystropy (Infantile) is behind the MPS II and MPS IIIA Programmes but about to enter a Phase I Intrathecal trial involving a variable dose regime similar to Hunter CNS.

A major objective of the day was to visit the new Shire HGT Lexington Manufacturing Plant. The Atlas facility supports the manufacture of REPLAGAL® (agalsidase alfa), VPRIV® (velaglucerase alfa), and Shire's robust pipeline of future development programmes. Along with Atlas, Shire owns four buildings in Lexington that serve as the global centre for Shire HGT, the business unit of Shire dedicated to the study of orphan diseases.

The Atlas facility oozes with futuristic technology that is currently being validated for the production of Replagal. As we worked our way through the gowning rooms ending up wearing all-in-one boiler suits with a frock over the top, three pairs of shoe covers, rubber gloves, hair net and protective glasses we could have been mistaken for people working at NASA! But then thinking about it, Shire's approach to manufacturing life-saving new therapies for Lysosomal Storage Diseases is as futuristic and pioneering as those working on the space station.

Christine Lavery CEO, c.lavery@mpssociety.co.uk

Photo: The MPS Network outside the Atlas facility, Lexington, USA

INTERNATIONAL SYMPOSIUM ON MUCOPOLYSACCHARIDE AND RELATED DISEASES
28 June – 1 July 2012, Noordwijkerhout, Netherlands

It is that time of the MPS calendar when we start planning our activities and budgets for next. The big one next year is the International Symposium on Mucopolysaccharide and related diseases hosted by the Dutch equivalent of the MPS Society. After the last two Symposia being held on far continents in Vancouver, Canada and Adelaide, Australia this meeting returns to the mainland of Europe and North Europe at that.

Noordwijkerhout is a seaside town about 30 minutes from Amsterdam and close to Leiden. There is a dedicated conference centre and conference hotel more geared to people attending without children. But wait for it the family accommodation is situated in Wassenaar and inside the Duinrell Theme Park **www.duinrell.com**. As well as attending the International Symposium families can enjoy a real family break by making a week of it. The bungalows can accommodate up to eight people and there is unlimited access to the theme park and most of the swimming pools.

The MPS Society is currently looking at what help it may be able to offer families to participate in the Symposium and thinking laterally whether we can extend the stay to give as many families as possible a few days break following the Symposium. The Dutch MPS Society has negotiated some very attractive rates for Duinrell bungalows.

Why not plan ahead with other MPS families you are friends with and plan to meet up in the Netherlands for the MPS Symposium in June 2012. By creative planning two families might be able to share one of the 8 bedded Duinrell Bungalows and reduce costs even more. There will be volunteer childcare provided by the MPS Society during the Symposium.

We are in the early stages of planning but would love to hear your thoughts: Are you interested in attending the MPS International Symposium in Holland? What are your thoughts to the MPS Society taking bungalows at Duinrell as family accommodation?

Would you be interested in arriving on Thursday 28 June in the afternoon in time for the opening reception, attend the Symposium until close on 1 July and spend the rest of the week on holiday at Duinrell?

If the MPS Society was to offer to pay the MPS Symposium registration including all meals except Friday evening dinner and childcare for up to two adults and two children unaffected by MPS and half the cost of a Duinrell Bungalow for the week, would you be interested? Please note prices are not confirmed but fifty percent of the cost of a Duinrell Bungalow would be around £350 for seven nights. Please also note that registration for children and adults affected by MPS is free of charge.

The Japanese Society of Patients and Families with MPS

Thank you so very much for your thoughts of encouragement and deep sympathies for the people of Japan facing the extraordinary serious disaster. We still can not reach some of the families living in the Tohoku region due to damaged communication lines. We also pray and hope that all of them are safe and in good health.

I am please to send you this abbreviated synopsis of our 25th bulletin of the Japanese MPS Society. I would like to thank the international community for supporting the Japanese MPS Society on behalf of all members.

Greetings come from Mr Toyoshige Mikami, the Present of the Japanese MPS Society, who is a father of a boy with Hunter disease. He says there is fear that the back space in the healthcare plan of the government will become advanced due to the prolonged economy slump and the acceleration of demographic aging in Japan. He emphasises that it is very important for our Society to respond to the changing times and to challenge aiming at new goals.

Prof Tadao Orii, the executive head of the Japanese MPS Society introduced promising research on a new drug for Enzyme Replacement Therapy for Morquio disease presented by Dr Shinji Tomatsu and his team at St. Louis University in the USA. They say it shows dramatic effects on bone lesions in tests using the Morquio mouse model.

Dr Yasuyuki Suzuki at Gifu University evaluates the activities of the Japanese MPS Society, the families and professionals over the past year. He highlights the visit to Taiwan for the natural history study of Morquio disease and the participation in the International Symposium for Mucopolysaccharide Diseases in Adelaide, Australia and the hosting of the Japanese MPS Symposium in Tokyo last year. Dr Torayuki Okuyama of the National Centre for Child Health and Development points out the importance for everyone in the country taking an interest in the problem of rare diseases for their own sake.

Dr Akemi Tanaka, Osaka City University, reports on the many active efforts of the Japanese Community in 2010 including the holding of the 52nd academic conference of the Japanese Society for Inherited Metabolic Diseases she hosted in October; the petition to the Ministry of Health, Labour and Welfare for the acceleration of the development of the therapy and the many aspects of strengthening family support.

Dr Haruo Shintako, also of Osaka City University emphasises the importance of collaboration between doctors, the MPS Society and government in order to solve the various problems associated with MPS.
Nobuhiro Kasa Vice President

Clinical Trial Update

The following clinical trials are open to patients in the United Kingdom:

MPS II Intrathecal Enzyme Replacement Clinical Trial
Shire Human Genetic Therapies is sponsoring a clinical trial at Birmingham Children's Hospital to learn if direct administration of recombinant enzyme into the fluid around the brain and spinal cord is safe and a possible treatment for children with MPS II with developmental delays. The Phase I/II study is a safety and ascending dose ranging study of idursulfase administration via Intrathecal drug delivery device in paediatric patients with MPS II who demonstrate evidence of central nervous system involvement and who are receiving treatment with Elaprase. This study is recruiting and the principal investigator is Dr Chris Hendrikz.

MPS IIIA Intrathecal Enzyme Replacement Clinical Trial
Shire Human Genetic Therapies is developing a sulphamidase enzyme replacement therapy (ERT) for patients with MPS IIIA. rhHNS is being administered into the cerebrospinalfluid via a surgically implanted Intrathecal drug delivery device (IDDD), because when administered intravenously it does not cross the blood brain barrier. This study is a multi-centre, multiple dose, dose escalation study designed to evaluate the safety, tolerability and clinical activity of up to three dose levels (two doses [10 and 45mg] monthly and one dose [45mg] every other week for six months) rhHNS administered via an IDDD in patients with Sanfilippo disease type A, aged 3 years and over.

The phase 1/2 clinical trial is planning to enrol 15 patients and began in June 2010. The study is expected to be completed by March 2012, and the duration of the study for each patient is nine months. The study is being conducted at two sites, the Netherlands by Dr Frits Wijburg and Manchester Children's Hospital under the direction of Dr Simon Jones and Prof. Ed Wraith.

MPSIVA MOR100 Study
BioMarin is developing enzyme replacement therapy (ERT) for patients with MPS IVA. This Phase 3 study will evaluate the efficacy and safety of 2.0 mg/kg/week BMN 110 and 2.0 mg/kg/every other week BMN 110 in patients with mucopolysaccharidosis IVA. This randomised controlled, placebo controlled, double blind safety/efficacy study involves receiving enzyme replacement therapy intravenously either weekly or every other week at one of three paediatric centres or two adult LSD centres in the UK.

Children 5 - 16 years
London – Great Ormond Street Hospital: Dr Ashok Vellodi
Birmingham Children's Hospital: Dr Chris Hendrikz
Manchester Children's Hospital: Dr Simon Jones/Prof Ed Wraith

Adults 17 years and over
London – Royal Free Hospital: Dr Derralynn Hughes
Manchester - Salford Royal Hospital: Dr Stephen Waldek
Birmingham – Queen Elizabeth University Hospital: Dr Tarek Hiwot
Belfast City Hospital: Dr Fiona Stewart (not yet enrolling)

Fabry Disease
Amicus is continuing to develop a chaperone treatment for patients with Fabry disease. This Switch Study (12) is to compare the efficacy and safety of AT1001 and enzyme replacement therapy (ERT) in male and female patients with Fabry disease who are currently receiving ERT and who have AT1001-responsive GLA mutations. This study will take place at the:
London - Royal Free Hospital: Dr Derralynn Hughes
Salford Royal Hospital, Manchester: Dr Stephen Waldek

Fabry Disease
As Amicus continues to develop a chaperone treatment for patients with Fabry disease it has launched and is recruiting to a Phase 3 trial (011). This clinical trial is for enzyme naïve patients, those patients that are not currently on ERT and who have AT1001-responsive GLA mutations and a raised GL3. This study will take place at:
London - Royal Free Hospital: Dr Derralynn Hughes
Salford Royal Hospital, Manchester: Dr Stephen Waldek

Fabry Disease
Shire Human Genetic Therapies are recruiting to the Replagal clinical trial for patients naïve to Enzyme Replacement Therapy. There will be three arms to the trial with patients either receiving 0.2mg/kg of Replagal weekly; 0.2mg/kg of Replagal every other week or 0.4mg/kg of Replagal weekly. This is a one year study. In the first 13 weeks, participants will receive their enzyme in hospital then for the next 43 weeks at home.
Salford Royal Hospital, Manchester: Dr Stephen Waldek

Contact details for those interested in knowing more detail on these Clinical Trials:

Salford Royal Hospital
Marie Meehan /Jo Webb
0161 206 4192/4376, Marie.Meehan@srft.nhs.uk

Royal Free Hospital
Alan Milligan / Linda Richfield
0207 472 6409, alanmilligan@nhs.net

Queen Elizabeth University Hospital
Kate Peers
0121 627 1627 Ex.53407

Birmingham Children's Hospital
Dr Chris Hendriksz
chris.hendriksz@bch.nhs.uk, 0121 333 9907/9908

Belfast City Hospital
Dr Fiona Stewart
fiona.stewart@belfasttrust.hscni.net

Manchester Children's Hospital
Dr Simon Jones/Prof. Ed Wraith
simon.jones@cmft.nhs.uk, ed.wraith@cmft.nhs.uk
0161 701 2137/8

CLINICAL TRIAL UPDATE

EUROPEAN UPDATE

Alpha Mannosidosis

A Phase 1 trial has recently been successfully completed and demonstrated that the enzyme, Zymenex, is safe and well tolerated. A Phase 2 dose-finding clinical trial is now underway. The biotechnologically derived human enzyme product rhLAMAN (LamazymTM), is produced by the Scandinavian biotech company Zymenex and developed for the treatment of patients suffering from the rare disease alpha-Mannosidosis. The clinical trial has now entered a new phase with the 10 patients from the Phase 1 trial, drawn from around Europe, moving forward into a Phase 2a dose-finding clinical trial, with the aim of indentifying the most optimal dose to achieve the desired clinical benefit.
www.zymenex.com/pressreleases

Treatment Therapies

MPS I

Aldurazyme®, administered once weekly, has been approved in the European Union for the long-term enzyme replacement therapy (ERT) in patients with a confirmed diagnosis of MPS I, to treat the non-neurological manifestations of the disease. Aldurazyme was developed by BioMarin and Genzyme under a joint venture agreement that assigns commercial manufacturing responsibilities to BioMarin, and worldwide sales and marketing responsibilities to Genzyme. Additional information can be obtained at **www.aldurazyme.com**

MPS II

Elaprase®, is a a long term ERT for patients with a confirmed diagnosis of MPS II which has been approved for use in the European Union. Elaprase was developed and is produced by Shire Human Genetic Therapies (formerly TKT), and is given as weekly infusions to replace the missing enzyme that Hunter syndrome patients fail to produce in sufficient quantities. Additional information can be obtained at **www.shire.com**

MPS VI

Naglazyme®, is the ERT for individuals with a confirmed diagnosis of MPS VI and has been approved for use in the European Union. It was developed and is produced by BioMarin Pharmaceuticals, Inc. Additional information can be obtained at **www.bmrn.com**

Fabry

Replagal® and Fabrazyme® are Enzyme Replacement Therapies for Fabry disease both approved in the European Union. Replagal was developed and is produced by Shire Human Genetic Therapies. Fabrazyme was developed and is produced by Genzyme. For additional information please visit **www.shire.com** and **www.fabrazyme.com**

Zacharon Pharmaceuticals Announces Research and Development Collaboration with Pfizer to Develop Drugs for Multiple Rare Disorders

Strategic Collaboration to Leverage Zacharon's Platform for Developing Small Molecule Drugs Selectively Targeting Carbohydrate Polymers

SAN DIEGO, April 7, 2011 – Zacharon Pharmaceuticals, Inc. today announced that the company has entered into a strategic research collaboration with Pfizer Inc. to develop drugs for orphan diseases, including lysosomal storage disorders. The potential value of the collaboration to Zacharon is approximately USD $210 million. The collaboration includes the potential development of compounds that may be discovered using Zacharon's innovative platform for developing small molecule drugs targeting specific carbohydrate polymers or glycans.

Zacharon, whose sole venture investor is Avalon Ventures, will receive up-front payments and research and development funding under the collaboration to develop drugs against targets that impact lysosomal storage diseases. Zacharon is also eligible under the collaboration for payments for meeting development milestones, plus royalties and sales milestones upon commercialisation.

"We are very pleased to be working with Pfizer, which has broad compound development expertise, including in the area of small molecules, which should be quite useful to developing drugs for these orphan diseases," said Robin Jackman, Ph.D., president and CEO of Zacharon. "The collaboration provides validation for the potential that lies in Zacharon's broadly applicable technology platform."

Ed Mascioli, M.D., head of Pfizer's Orphan and Genetic Diseases Unit, said: "Part of Pfizer's Orphan and Genetic Diseases Unit's strategic focus relies on collaborations with companies like Zacharon that have promising technologies to help develop treatments for rare diseases. Zacharon is an ideal partner, and we look forward to working with them to develop treatments for lysosomal storage diseases."
For more information, please visit **www.zacharon.com**.

ISMRD (the International Advocates for Glycoprotein Storage Diseases) has announced it 5th Family Meeting, 28 – 29 July 2012 Charleston, South Carolina. Natural History Clinics will be held on Friday 27 and Monday 30 July in Charleston to coincide with the Family Meeting.

Dear MPS Society

We would like to inform you of a new Phase 3 research study of an investigational medicine being developed by Amicus Therapeutics and GlaxoSmithKline. AT1001 (miglastat hydrochloride) is an oral agent under development for the treatment of Fabry disease, a progressive, chronic and often fatal genetic disease with high morbidity. Fabry disease is an X-linked, lysosomal storage disorder which results from a deficiency of the lysosomal enzyme a-galactosidase A (a-GAL A) leading to accumulation of globotriaosylceramide (GL-3) in blood vessels, tissues, and organs, impairing their function and leading to a wide range of symptoms. Should you have any patients with this rare, debilitating condition, you might want to consider if they would be willing to participate in the research program.

The primary objective of the study is to evaluate the effect of AT1001 on kidney GL-3 levels. In addition, the study will assess the effect of AT1001 on urine GL-3 levels, renal and cardiac function, pharmacokinetics, and patient-reported outcomes. Approximately 60 subjects from various countries worldwide will take part in this study, and although enrollment is proceeding well, additional subjects are still being recruited.

The current standard treatment for Fabry disease is enzyme replacement therapy (ERT) that is administered intravenously. AT1001 is an orally-administered pharmacological chaperone designed to selectively bind to the a-Gal A enzyme, stabilise it and restore its biological activity, allowing breakdown of GL-3. AT1001 represents a novel approach to treating Fabry disease that may have advantages relative to ERT, including ease of use (oral vs. infusion) and better tissue distribution throughout the body. Data supporting clinical proof of concept of AT1001 for Fabry disease has been observed in Phase 2 studies.

This latest study consists of 2 stages, plus an optional open-label extension. During Stage 1, participants will be randomly assigned to receive either AT1001 or placebo for 6 months. During Stage 2, all participants will receive AT1001 for an additional 6 months. Upon completion of Stage 1 and Stage 2, participants may be eligible to participate in a 12-month, open-label AT1001 treatment extension phase.

It is important to note that at the conclusion of the trial, patients who participate in the study will return to the full care of the referring physician.

If you or any of your members have any questions about the study, please do not hesitate to contact one of us using the information below. If you or an individual with Fabry disease, would like to learn more about the study, or see the full list of participating clinical sites, please visit the FACETS website at: www.fabrystudy.com or www.clinicaltrials.gov., keyword search AT1001.

Best regards

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Stephen Waldek, MB BCh FRCP
Salford Royal NHS Foundation Trust
Department of Adult Inherited Metabolic Diseases
Manchester, United Kingdom
44 (0)161 206 4365, steve.waldek@srft.nhs.uk

Inclusion and Exclusion Criteria

Inclusion criteria:

- Male or female between the ages of 16 and 74 inclusive, diagnosed with Fabry disease
- Confirmed GLA mutation that has been shown to be responsive to AT1001 in vitro
- Note: Subjects under the age of 18 will be enrolled only at sites with all required regulatory and ethics approvals to do so.
- Naïve to ERT or have not received ERT for at least the 6 months before Screening
- Urine GL-3 ≥ (greater than or equal to) four times the upper limit of normal at Screening
- Subjects taking angiotensin converting enzyme inhibitors (ACEIs) or angiotensin receptor blockers (ARBs) must be on a stable dose for a minimum of 4 weeks before the baseline visit.
- Male and female subjects of childbearing potential agree to use medically accepted methods of contraception during study and for 30 days after study completion
- Subject is willing and able to provide written informed consent, and assent if applicable

Exclusion criteria:

- Subject has undergone or is scheduled to undergo kidney transplantation, or is currently on Dialysis.
- eGFR < 30 mL/min/1.73m2 (chronic kidney disease [CKD] Stage 4 or 5) based on Modification of Diet in Renal Disease [MDRD]) equation at Screening
- Pregnant or breast-feeding
- History of allergy or sensitivity to study medication (including excipients) or other iminosugars (e.g., miglustat, miglitol)
- Subject is treated or has been treated with any investigational drug within 30 days of the screening visit
- Subject is currently treated or has ever been treated with AT1001
- Any intercurrent condition or concomitant medication use considered to be an absolute contraindication to kidney biopsy or that may preclude accurate interpretation of study data
- Otherwise unsuitable for the study, in the opinion of the investigator.

An Update on Work Carried Out on ML II Zebra Fish by Dr Richard Steet

Over the last several years, Dr Richard Steet at the Department of Cellular Biology, University of Georgia, has taken advantage of the speed and utility of the vertebrae model organism, zebra fish (Danio rerio) to study the molecular and cellular pathogenesis of Mucolipidosis Type II (ML II). Defining the disease process of ML II will inform the development of novel therapies that do not rely on the replacement of the defective enzyme. His group's initial findings (published in the American Journal of Pathology in 2009) showed that ML II zebra fish have phenotypes in many of the same organ systems affected in ML II children. Focusing on the craniofacial cartilage defects, they demonstrated striking changes in the composition and homeostasis of the extracellular matrix in ML II zebra fish.

These findings are significant since they point to pathogenic mechanisms outside the lysosomes and even the cell. More recently, the Steet Laboratory has discovered that up-regulation of several classes of protease enzymes also accompany the cartilage defects in ML II zebra fish. Similar findings were observed in cells from a feline model of ML II, demonstrating that this up-regulation is a general feature of the diseases. The research group is now directly testing the contribution of these proteases toward the onset and progression of phenotypes in ML II zebra fish embryos using small molecule inhibitors and rapid gene suppression techniques.

Preliminary results are encouraging and suggest that a reduction in certain protease activities can partially alleviate the disease symptoms. These experiments will be extended to investigate the impact of the proteases and additional protein targets on other phenotypes within the zebra fish model including motility and cardiac malformation. This work highlights the power of the zebra fish system to rapidly address disease pathogenesis and test potential therapies.

This article is reprinted from the ISMRD Pathways Newsletter for Glycoprotein and Related Storage Diseases

PATIENT INFORMATION CARDS

Amie was 18 years and 3 months when she was admitted as an emergency to the Nuffield Hospital, Oxford in May 2009. A few days later she was dead. She suffered with an attenuated form of Mucolipidosis Type II and was a bright, communicative young lady who proved on her 18th birthday that she knew how to party and dance in her wheelchair. This wasn't because Amie was moribund on arrival at hospital or that her condition was terminal at that point in time. Despite the cause of death at Amie's inquest being found to be natural causes it was acknowledged that there were a number of contributory factors that could have been avoided.

- No knowledge of Amie's condition
- No paediatric size monitoring equipment available
- Doctor's failing to heed Amie's parent's constant requests to contact her MPS specialist in Manchester

It is the wish of the MPS Society, and Amie's family that if at all possible no other family suffers in such a traumatic way. Amie's parents had to challenge the doctors to carry out tests and as Amie was dying waiting several hours for a doctor to attend her, her father was removed from the hospital by security guards due to the distress he was showing. We all accept Amie was never going to make old bones, but we do not accept that with years of experience of the condition as a patient, parent or carer that our loved ones should suffer and lose their lives in this way. With National Specialist Commissioned Expert Centres in England and good regional clinical centres in Wales, Scotland and Northern Ireland so much has changed for the better unless it would seem you are admitted to a hospital who do not know you or your MPS loved one, is ill-equipped and does not listen to patients, parents and carers. I am grateful to Amie's mother, Alison, for allowing us to retell Amie's story in this way to highlight why the new MPS Patient Information Card could be a real lifeline.

Using funds raised by the Ollie G Ball we now have the machinery and materials to create a Patient Information Card for each and every one of you. They will be of greatest value if you, your partner or child is seen at or admitted to a hospital you are not familiar with or are involved in an accident.

The card will include relevant patient details and medical information important to the condition you, your partner or your child suffers from. It will carry contact details for you, your partner or your child's MPS specialist and hospital number. At times of emergency and high anxiety it is so easy to forget this important information and experience has shown that having a dedicated patient information card can be the difference between life and death. Enclosed with this MPS Magazine is an application form. If you would like to have a personal patient information card free of charge please complete the form enclosed and return it to the MPS Society.
Christine Lavery c.lavery@mpssociety.co.uk

Challenging BEHAVIOUR foundation

making a difference to the lives of people with severe learning disabilities

Challenging behaviour: a guide for family carers on getting the right support

The Challenging Behaviour Foundation is delighted to announce that three new 'At a glance' guides aimed at family carers supporting people whose behaviour is described as challenging are now available. There are separate guides for adults, teenagers and children.

The guides have been developed in collaboration with the Social Care Institute for Excellence (SCIE).
The guides help family carers to:

- Understand what good support and services look like
- Work in partnership with staff who are involved with the family
- Find information on what to do and who to contact if the family's needs are not being met

David Walden, Director of Adult Services at SCIE, says "These guides look in detail at challenging behaviour and are aimed at family carers. Challenging behaviour can put families under great pressure. So, services should support parents and other family carers in their caring role. Also, problems are often caused as much by the way a young person is supported – or not supported – as by their disabilities. People often behave in a "challenging" way if they have problems understanding what's happening around them or communicating what they want or need."

Helen Marron
The Challenging Behaviour Foundation
Email: info@thecbf.org.uk

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

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

Genetic Alliance UK
Supporting. Campaigning. Uniting.

Genetic Alliance
Get Your Views Heard - Recruiting now!
New medicines: How should we weigh the risks and benefits? And who should do so?

Dear MPS Society
We are very pleased to be launching our recruitment campaign for an innovative and exciting research project about new medicines. Genetic Alliance UK are looking for people who are affected by a severe, rare or genetic condition, to take part in a Citizens' Jury later this year. We are looking for up to 16 individuals, to form a diverse jury that will deliberate a very important issue - the risks and benefits of new medicines. Not only will jurors get their voice heard in an innovative and high profile way, but they will be paid for their time too!

There is further information available about the project and how people can get involved on the www.GeneticAlliance.org/projects/risksandbenefits2 website including a short explanatory video clip and an online questionnaire for people to register their interest. We would be extremely grateful if you could share information about the project and our recruitment campaign, with as many of your patient members and networks as you can.

Our aim is to generate as much awareness and interest in the project as possible! We have a range of material and literature, which can be distributed via emails, patient group websites and newsletters. To get access to these documents (or to simply find out more) please contact Project Officer, Amy Simpson amy@geneticalliance.org.uk or Marketing Officer, Julian Walker julian@geneticalliance.org.uk

Disability Grants

Your Guide to Grants for the Disabled

Do you have a Disability?
Are you a parent or carer of a Disabled child or adult?

If so, use this website to save time finding Disability Grants.

Charities and Trusts provide funding towards the high cost of disability equipment, holidays, housing, days out... in fact anything above and beyond the normal costs of everyday living.

Over the past 15 years my family has used grants to help fund trikes, wheelchairs, adaptations to our home, medical equipment and specialist holidays for my disabled son.

With this support, my son was able to cycle back from school with his friends, join us for a family meal in the kitchen and experience the exhilaration of wheelchair abseiling.

His new wheelchair funded by one of the charities on this site not only increased his independence and mobility but also his street cred - Absolutely essential for a teenager!

Searching for extra funds takes time - in between work, hospital appointments, caring, therapies... and the hundred and one other things we all have to juggle every day.

So lets save time by sharing our Knowledge, Information and Resources... www.disability-grants.org

Carers Week

From 13th to 19th June is Carers week. There is a website dedicated to this week and the aim is to put Carers onto centre stage.

There are a number of organisations involved with Carers Week and you will be able to access who they are, and what they do and what they may be able to do for you and your family.

Find out more about Carers Week, and find out how you could take part by contacting carersweek@carersuk.org or call their hotline 0845 241 2582.

Have your say
The internet site http://carersweek.org/ has a survey of Carers running. Why not take some time to have a look and fill in the survey and contribute towards Carers' needs and views being heard.

Every little helps and every person matters.

MESSAGE BOARD

In the latest magazine under the New Members section, Alison wanted tips and advice about neuropathic pain that her sons who have fabry suffer from.

Louise (Hanson) said that hard massage was the only thing that helped with her youngster's pain.

Daniel's mum, Elaine, wants to know if any members who have children with Hunter, MPS II have leg spasms, and if so, what do they do to help it?

If you would like to share your questions, answers and thoughts, please email ***newsletter@mpssociety.co.uk***

Find us on Facebook

The MPS Society Facebook page is a means of providing information to our MPS Members and Friends quickly and efficiently. In the future we hope to feature some of our events and activities and recognise those that contribute to the Society and the work that we do.

You can find us by entering *MPS Society* into the facebook search engine.

As well as aiming to provide you with news from our fundraising activities and MPS events to coincide with our quarterly MPS Magazine we are also hoping to encourage greater awareness of the MPS Society.

If you have any ideas or suggestions for our facebook page please email **facebook@mpssociety.co.uk**

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

A new DVD wins an award

On 29 March 2011, I made my way over to Belfast to attend the Belfast Health & Social Care Trust, Chairman's award. Both Alison Wilson and I attended as representatives from the MPS Society.

This was in support of the DVD instigated by Dr Fiona Stewart and produced jointly between the Regional Genetics Centre, the Public Health Agency previously the Western Health and Social Services Board, Derry Travellers' Support Group and the MPS Society.

The DVD (Hurler, improving Awareness of Disease) was produced to raise awareness of Hurler Disease in Northern Ireland and to promote early diagnosis and treatment.

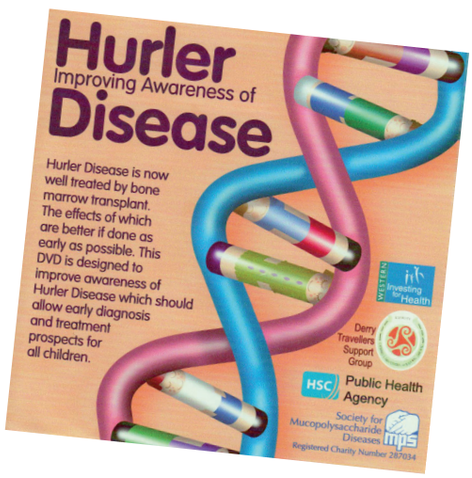
Dr Stewart, entered the DVD to be reviewed by the Belfast Health & Social Care Trust, Chairman's award and the panel shortlisted it to receive an award at the ceremony.

The DVD was up against three other well deserving projects that have been set up to better support the people of Northern Ireland.

Fantastically the DVD came second which brought with it a prize of £3,000 presented to Dr Fiona Stewart, pictured below.

Our congratulations go out to all who were involved in making and producing the DVD.

Sophie Thomas s.thomas@mpssociety.co.uk



Volunteering opportunities at MPS

Are you interested in becoming a childcare volunteer for the MPS Society? Perhaps you're not able to support us through fundraising, but maybe you can spare some time and energy to be one of our childcare volunteers?

Maybe your work place & colleagues could donate some time to support our childcare programme at events?

Can you volunteer your time caring for MPS children and their siblings at events run by the MPS Society? Volunteering is fun and rewarding. It could also help you learn new skills and gain valuable work experience.

We are always looking for new volunteers to help out with events and conferences that we run throughout the year. The MPS Society relies on volunteers to assist in the care needed for children and young adults affected by MPS and Related Diseases. All of our volunteers undertake training in moving and handling and are fully briefed prior to the event.

Becoming a volunteer
To become a childcare volunteer we would need you to complete an application form. The MPS Society accepts volunteers from the age of 16 years on a trainee basis. We will require you to undergo an enhanced Criminal Records Bureau check as the Society supports children and vulnerable adults. If you are a new volunteer we also require two references and ask you to attend a compulsory training day at MPS House in Amersham. Once we have obtained satisfactory references and your CRB check, you will then receive an acknowledgement that your application has been accepted and you will be added to the volunteer mailing list. All new volunteers will be mentored by an experienced volunteer.

The MPS Society organises a programme of events and activities throughout the year. These include sibling weekends for brothers and sisters of children affected by MPS and related diseases, adult weekends for adult individuals affected by the diseases and family weekends, expert meetings and conferences for the whole family to participate in.

Our event programme is exciting and we rely on our childcare volunteers to keep our children and vulnerable adults safe whilst ensuring they have a happy and memorable time.
Please can you help us?
Email: mps@mpssociety.co.uk
or phone 0845 389 9901

How your money helps...

More professional support for more MPS Families

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

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

More MPS advocacy workers

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

More vital information

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

More help to cope with the isolation of a rare disease

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

More noise to force through change

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

More help

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

For more information, to seek support and advice from our advocacy team, or to help raise funds so we can continue our work, contact us now!
0845 389 9901 mps@mpssociety.co.uk

WANTED!

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

for

FREE!

Minimum sponsorship required

Help us raise funds by making an exhilarating 10,000 feet freefall parachute jump. No experience is required and if you raise the minimum amount of sponsorship you will get to jump for FREE!

Phone **0845 389 9901**
or email **fundraising@mpssociety.co.uk**