MPS

Spring 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: Parth (MPS II) from India.
To read his full story please turn to page 32

Society for Mucopolysaccharide Diseases

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Magazine Deadlines

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

IN THIS ISSUE...

Spring 2011

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***

Chief Executive's Report

The MPS Magazine is arriving on your doorstep as Spring breaks and the lighter evenings are on the way. At this time many of you will be concentrating on how the Government Spending Review is going to impact on your healthcare and finances as well as how cuts in local government will affect the services you or your family receive and depend on in respect of disability. The advocacy team are already reporting increased numbers of calls for help as we see growth in the number of Disability Living Allowance (DLA) applications being refused and even more refusals to grant Employment Support Allowance (ESA). If you are affected by cuts related to yours, or your child's MPS or Fabry disease, please do not hesitate to contact the advocacy team for help at the earliest opportunity.

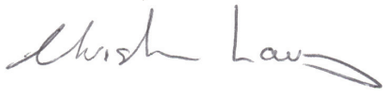
Looking to the summer, we are keen to welcome as many of you as possible to the MPS/Fabry Weekend Conference 24 – 26 June at the Hilton Hotel Northampton where we will be celebrating the start of the Society's 30th Anniversary year. Bookings are coming in thick and fast so don't sit on your booking form too long if you do not want to be disappointed. This weekend is not just a unique opportunity to meet with other families dealing with similar challenges of living with Mucopolysaccharide, related diseases and Fabry but to mingle with the UK's leading experts in the science, clinical management and treatment of these complex diseases. The weekend is not just about you but the children too! The MPS Society is renowned for its amazing childcare programmes designed to meet the needs of all ages and abilities. We can also care for 'adult' children unable to participate in the conference programme and guarantee they will have a stimulating time with our team of experienced childcare volunteers. The Gala Dinner on Saturday will be full of fun and surprises not to mention the draw naming the lucky families to go on the once in a lifetime visit to Lapland in December.

Thank you for your excellent response to the two Fabry workshops to be held in May at the Royal Free Hospital and Sidney Sussex College, Cambridge. Regionally within England we will be hosting transition consultation workshops for young people aged 14 – 25 years and simultaneously for parents. This is a unique opportunity for you to have your say as a young person or parent carer on what you expect from transition to adult health care services if you are in the transition process or to share with us your recent experiences of transitioning to adult care. Your thoughts and experiences are very important as they will help inform the development of a model of transitioning to adult healthcare for those diagnosed with a lysosomal storage disease.

The Society was delighted to learn that the MOR100 Enzyme Replacement Trial is to be extended to include adults with MPS IVA, Morquio disease. The two clinical trial centres will be the Royal Free Hospital in London and the Belfast City Hospital. The Society is writing to MPS IVA adult members individually to advise them of who to contact if they are interested in knowing more about participating but if you are interested in knowing more please do contact us. We are also very pleased to announce recruitment to the MPS II intrathecal ERT clinical trial is open at Birmingham Children's Hospital.

In respect of Fabry disease, recruitment will soon start on a Study 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 Royal Free and National Hospitals in London and Hope Hospital, Manchester. If you are interested in knowing more about clinical trials recruiting or taking place do go to *www.clinicaltrials.gov* and insert the disease you are interested in.

Finally as was announced in the Winter 2010 MPS Magazine, there is to be no more research and support money for the MPS Society from Jeans for Genes. The Society has been preparing for this time to come for nearly a year now but with the worsening economic climate and in these challenging times for the charity sector, we have no choice but to ask for your help in generating new income by asking friends, family, children's schools and employers, anyone you can think of to support MPS to raise funds for our vital support and advocacy services as well as groundbreaking research for projects such as the Clinical Trial for Genistein in Sanfilippo disease. MPS Awareness Day on Sunday 15 May offers a chance to join us for a day of fun and to celebrate at Whipsnade Zoo as well as being an amazing opportunity to not only raise awareness of your MPS or related disease but to raise valuable funds for the Society.



Christine Lavery
Chief Executive

Spring 2011

Management Committee Update

The Society's Board of Trustees meet regularly. Here is a summary of the key issues that were discussed and agreed at the Management Committee Meeting held 4-5 February 2011.

Governance

Trustees reviewed the MPS policies. It was agreed that regular review of MPS policies should be undertaken by staff rather than Trustees and that Trustees should only review the amendments to policies. The CEO is working on a new Equality Policy in line with recent changes in legislation. Trustees requested further information on the benefits of membership to the Fundraising Standards Board before agreeing whether to renew the Society's membership for another year.

Personnel

The CEO presented her personnel report which covered recent staff changes and advised Trustees on updated guidance received from Peninsula with regards to employment contracts and policies which affected MPS staff.

Risk Management

Trustees reviewed the Risk Register and the CEO advised on a small number of points. The Disaster Recovery Plan is still under development.

Accounts

The year end accounts up to 31 October 2010 were agreed, subject to amendments by the Trustees.

Jeans for Genes

The CEO appraised Trustees of the current situation with regards to Jeans for Genes. Trustees asked the CEO to ensure that the Society receives a copy of the Jeans for Genes database as soon as possible and to work closely with other J4G trustees to secure as much income as possible from the 2010 J4G appeal.

Clinical Management

The CEO updated Trustees on the progress of the QIDIS grant 'Building a healthcare transition model for LSDs'. Trustees expressed concern on the part of the Society's members that no ministerial approval had yet been received in respect of decommissioning of the LSD paediatric service at Addenbrooke's Hospital and the proposed move of the LSD paediatric consultant, Dr Uma Ramaswami, to Great Ormond Street Hospital.

Communications

Trustees were advised that the MPS annual national draw is being re-introduced in 2011 to generate income and awareness of MPS.

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 75% is spent in administrative costs, the MPS Society spends 15p in the pound administering the Society for our members. **Please spread the word and think 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. If you are contacted by the Chronic Granulomatous Disease Research Trust (CGDRT) or anyone in the name of Jeans for Genes after 1 April 2011 by email or telephone, please do let MPS know.

All Ireland Advocacy Service

The All Ireland Advocacy Service has now reached its 6 month birthday – so now feels like an excellent time to update members on some of the work that has been done so far and what the plans are for the future.
Alison Wilson *a.wilson@mpssociety.co.uk*

Northern Ireland

Since the launch of the All Ireland Service in September 2010 I have written to all the MPS Society members living in Northern Ireland to introduce the service. The response so far has been fantastic and I was delighted to receive my first phone call from one of our Northern Irish members just a few days after sending out my letters! Without exception, all of the MPS Society members I have had the opportunity to speak to, have sang the MPS Society's praises and let me know how delighted they are to have a local representative – It is very encouraging to be made feel so welcome in the MPS community in Northern Ireland.

Since sending out my letters, I have spent many hours contacting each of the Northern Ireland members by phone (I haven't quite managed to speak to everyone yet – but I'm getting there!); and I have visited many of our members in their homes to discuss their support needs. It has been lovely getting to know the families in Northern Ireland. Your strength and determination, even when things don't go exactly as planned, are inspirational.

So far I have been able to support families through changes in treatment regimes, issues with education and schooling, managing challenging behaviour, applying for home adaptations and accessing expert medical advice. I have also been pleased to be able to offer emotional support to members during difficult times.
I hope to be able to continue to support families as they face challenges associated with the conditions that affect them.

Winter MPS clinic

Before I was employed by the MPS Society I had the opportunity to attend the 2010 summer clinic as a genetic counselling student – I found this clinic absolutely fascinating and was really looking forward to attending the clinic as part of my new role.

Sophie and I attended this clinic and spent the day in the waiting area chatting with families. I also had the opportunity to spend some one-on-one time with a few members who wanted to discuss specific areas of support – MPS specialist clinics are an excellent opportunity to spend some time discussing support issues, particularly for busy families who find it difficult to make time for home visits. We were absolutely delighted with the excellent turn-out at this clinic! Twelve families braved the snowy roads and made it to the Antrim Area Hospital for their appointments. I think this turn-out just goes to show how much our families value these appointments.

Although I had already met a few families during home visits and other clinic appointments, this was my first opportunity to meet lots of families in one place at the same time; and to witness the friendships and camaraderie amongst our members in Northern Ireland. I found this so encouraging and the day flew by as I chatted to families.

I would like to extend a big thank you to all the families who attended, Dr Simon Jones, Dr Fiona Stewart, Dr Joanne Hughes and the nursing team in Antrim Area Hospital for making this clinic such a success!

I look forward to the next MPS clinic in Antrim Area Hospital in May.

Joint Fabry Clinic

In October 2010 I attended a joint cardiac and genetics Fabry clinic held in the Royal Victoria Hospital in Belfast. This is a relatively new clinic and as such it is continually growing and developing. The aim of this clinic is to provide a 'one-stop-shop' for the management of Fabry Disease patients in Northern Ireland. I will be working closely with Joanne McCosker (Inherited Cardiac Nurse Specialist) to help develop this clinic. As with the MPS clinics (that you are all familiar with), the MPS Society will have a presence at these clinics so that we can provide support to our members. These clinics will be held every 3-4 months in the Royal Victoria Hospital.

On arriving at this clinic I was greeted by Dr Pascal McKeown (Consultant Cardiologist), Dr Fiona Stewart (Consultant in Medical Genetics), Joanne McCosker (Inherited Cardiac Nurse Specialist) and Tracy Jardine (Inherited Cardiac Nurse Specialist) and I was given a brief overview of each of the patients that would be attending the clinic – not all the patients attending this clinic were aware of the MPS Society so it provided an excellent opportunity to explain the role of the MPS Society to these patients (and even sign up a few new members!). I received a warm welcome at this clinic and was very much encouraged by this.

This new clinic is an excellent opportunity for the MPS Society to further engage with Fabry patients in Northern Ireland and I look forward to the next clinic in March.

Hurler Awareness DVD

In January 2011, Sophie made a trip over to Northern Ireland to attend the launch of a Hurler Disease Awareness DVD that aims to provide information about genetic testing and bone marrow transplant to members of the travelling community in Northern Ireland.

This DVD was created as part of a joint initiative between the Northern Ireland Regional Genetics Service, Western Improving for Health and the MPS Society. The Derry Travellers Support Group were also involved in ensuring the DVD was appropriate for the intended audience. The DVD featured Christine Lavery, Sophie Thomas and Dr Fiona Stewart - who presented information about Hurler Disease, genetic testing, Bone Marrow Transplant and the role of the MPS Society in supporting families.

In my opinion, however, it was Sandra Hughes and Ellen who stole the show! Sandra spoke about the successes of her son Jamie's bone marrow transplant and Ellen spoke about her daughter, who sadly passed away following a diagnosis of Hurler Disease. I hope that the stories told by these two ladies will be of benefit to all those who have the opportunity to watch this DVD.

This DVD was developed long before I was appointed by the MPS Society, but I am delighted to be getting involved in setting up screenings for members of the travelling community in Northern and Southern Ireland. We can only hope that this DVD will be beneficial to members of the travelling community.

We are delighted that this DVD has been shortlisted for the Belfast Trust Chairman's award. This award recognises ventures within the Belfast Trust that show examples of collaboration between departments and organisations. It also recognises those ventures that have a positive impact on patients. It is an honour to be considered for this award and we would like to thank Dr Fiona Stewart for entering the DVD.

Some quotes from our N.Ireland members

Michelle Worsford *(Co. Down): 'I have found this service extremely supportive and helpful. It takes away a lot of the stress when you know you have this support fighting your corner.'*

Chloe McCauley *(Age 13yrs MPS I Hurler Scheie): 'I like Alison. She is nice to me. She understands what is wrong with me and helps me to deal with it and if I ever need to talk to her all I have to do is ring her.'*

Jacinta and Kevin McCauley *(Co. Londonderry): 'As we all know, the MPS Advocacy Support Team play a vital role in support of our families, however we found that having our support officer based in N. Ireland much more beneficial. Our Support Officer, Alison, has already done so much, not just for Chloe who is 13 with MPS I, but for the family as a whole. Alison was able to do home visits with Chloe and us, able to answer queries we had and was able to attend not just clinic meetings but a school meeting as well. Because this service is locally based we find it much more readily available, and long may it last!'*

Southern Ireland

As part of my role I am also working alongside the Irish MPS Society, the Our Lady's Children's Hospital's Bone Marrow team and The Temple Street Metabolic Unit to provide support to individuals diagnosed with MPS and related disorders in Southern Ireland.

So far I have been delighted to be able to support two families in highlighting the need for access to essential services for their sons (both diagnosed with Hunter Disease); and in the coming months I hope to attend some social events arranged by the Irish MPS Society. As news spreads of our involvement with families in Southern Ireland I hope that we will be able to provide the same level of support to these families that we currently provide for families across the UK.

The service in Southern Ireland is very much in its infancy. I am spending lots of time building up relationships with medical professionals and it is hoped that by building these relationships more families will be pointed in our direction in times when they need support and advice.

The future

As the All Ireland Advocacy Service develops I hope that more families in both the North and South of Ireland will benefit from the support and advice available. Over the next few months my main aim is to begin working towards some social events for families in both Northern and Southern Ireland. By bringing families together it is hoped that the MPS community in Ireland will be strengthened and that families will be able to benefit from meeting other families in similar circumstances.

De-designation of the Paediatric LSD Service at Addenbrooke's Hospital, Cambridge

Following much consideration and a service review the recommendation was made to Ministers in September 2010 that the paediatric LSD service at Addenbrooke's Hospital be de-designated as of 31 March 2011. The plan which had been in place for nearly a year was that the de-designated service move to Great Ormond Street Hospital.

It therefore came as an unwelcome surprise to many of our members to learn at the end of February that Great Ormond Street Hospital (GOSH) management had decided they do not want the whole service, but would be happy to clinically manage the patients. Our 34 children and their families affected by the GOSH decision have had their lives turned upside down. From feedback we have received they are very fond of their LSD paediatrician, Dr Uma Ramaswami, not least for her untiring efforts clinically managing the children's MPS or Fabry disease; advocating on their behalf in respect of social care and education and just being interested in the whole family. As we go to print, all efforts are being made to find a solution possibly with Birmingham Children's Hospital. If successful this would mean that the 34 boys and girls will not lose their much respected paediatrician.

If you are affected by the de-designation of the paediatric LSD service at Addenbrooke's and want to talk or need help or support please do contact us.
Christine Lavery c.lavery@mpssociety.co.uk

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 will not affect the current service already provided by the MPS Advocacy Team to those affected by MPS and Related Diseases.

We are developing a Guide to Understanding MLD fact sheet which will be available shortly 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**

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.

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**

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.

Getting involved with MPS...

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, Fundraising Magazine, Children's Newsletter 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!***

MPS Annual Report and Accounts

The MPS Society annual report and accounts for 2010 will be available to download from our website, www.mpssociety.co.uk, by 1st June 2011. 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.

Thank you to all our supporters!

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

We are so appreciative of your support and for thinking of the MPS Society. We get a number of requests each year to attend cheque presentations or give talks on our work. We always like to do these when possible but to minimise the costs to our charity, try 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
Fundraising fact sheets
Sample press release
Sponsorship forms
Become a Friend of MPS
Promotional Goods Order Form
Publication Order Form
T-shirts, posters, balloons, plus more...

Email us at **fundraising@mpssociety.co.uk**, visit **www.mpssociety.co.uk** or phone 0845 389 9901

WHAT'S ON 2011!

CONFERENCE EVENTS

MPS National Weekend Conference
24 - 26 June *(Please see enclosed booking form)*

SPECIAL EVENTS

Sibling Week
25 - 29 July *(Please see enclosed booking form)*

Fabry Focus Groups
20 May: Royal Free Hospital
21 May: Sidney Sussex College, Cambridge

Lapland Christmas Visit
11 - 14 December

For more information about how you can help us celebrate MPS Awareness Day, please visit **www.mpssociety.co.uk**

AWARENESS EVENTS

MPS Awareness Day
Sunday 15 May
Whipsnade Zoo

MPS REGIONAL CLINICS

Birmingham clinic: 17 June, 18 November
Bone Marrow Transplant clinic (under 6's): 15 April, 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
Bristol clinic: 17 May
Newcastle clinic: 21 April
Northern Ireland clinic: 27 May

MPS SIBLING WEEK

The last MPS Sibling Week was such a huge success that we are holding another event this year!

Windmill Hill, East Sussex
PGL Adventure Holiday
Monday 25 – Friday 29 July 2011

Please look out for the form enclosed with the magazine.

If we do not receive enough bookings then we are unable to hold the event. Siblings must be between the ages 7 and 15 years old. There will be a small cost to families of £40.00 per sibling attending. This is not refundable.

New to PGL in 2009, this impressive Grade 2 listed Mansion House is set in 21 acres of grounds on the Sussex Downs.

The centre has undergone a £7 million investment and now offers brand new lodge accommodation and activity bases as well as a large indoor sports hall.

Aeroball, Abseiling, Archery, Ball Sports, BMX Biking, Bridge Building, Canoeing, Challenge Course, Climbing, Giant Swing, Initiative Exercises, Jacob's Ladder, Orienteering, Raft Building, Rifle Shooting, Sensory Trail, Swimming, Trapeze, Zip Wire

Look out for our new Sibling Factsheet at www.mpssociety.co.uk

ANNOUNCEMENTS

New Members

Alison has recently been in contact with the Society. Her family has a diagnosis of Fabry disease. The family live in the Leeds area. Alison is interested in talking to anyone whose sons have experienced neuropathic pain in their feet for suggestions/tips/advice. Her sons are 9 and 12.

Mr and Mrs Kaliniak have recently been in contact with the Society. Their son named Patryk has a diagnosis of Sanfilippo disease. He is 3 years old. The family live in the South East.

Ms R Clarke has recently been in contact with the Society. Rachel has a diagnosis of Fabry disease. She lives in the Worcestershire area.

Mrs Carter has recently been in contact with the Society. Her daughter Naomi has a diagnosis of Fabry disease. Naomi is 11 years old. The family live in the South West and her brothers are still to be tested for the disease.

Mr and Mrs Hulse have recently been in contact with the Society. Miya has a diagnosis of Hurler Disease. Miya is eight months old. The family live in the South West.

Mr Laycock has recently been in contact with the Society. David has a diagnosis of Fabry disease. The family live in the Bristol area.

Deaths

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

Shinyar Hasan who suffered from Sanfilippo disease and who passed away on 24 July 2010 aged 13 years.

Ansam Hussain who suffered from Sanfilippo disease and who passed away on 15 December 2010 aged 18 years.

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

PEERAGE FOR DAFYDD WIGLEY

Many congratulations to former Plaid Cymru leader Dafydd Wigley who has been granted a peerage by the Queen. Lord Wigley, MP for Caernarfon between 1974 and 2001 will be known to a number of our more established members for his staunch support of the MPS Society particularly in its early days. Lord Wigley and his wife Elinor Bennett are the parents of two sons who lost their lives to Sanfilippo disease in the mid 1980s. Dafydd and I worked together on the Alton Bill and the Human Fertilisation and Embryology Bill over 20 years ago and Dafydd kindly hosted the Society's 10th birthday tea party in the Palace of Westminster (pictured below). **Christine Lavery**

OBE for ALISTAIR KENT

Congratulations to Alistair Kent, Director of the Genetic Alliance who received an OBE in the Queen's New Year's Honours list 2011. I was a member of the Board of Trustees for the then, Genetic Interest Group, who appointed Alistair to his Director post in the 1990's. I am delighted to see Alistair being recognised for his outstanding contribution to healthcare particularly his support of families affected by genetic disease. **Christine Lavery**

MPS Annual General Meeting 2011

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.

Society for Mucopolysaccharide Diseases
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Tel: 0845 389 9901, Fax: 0845 389 9902 www.mpssociety.co.uk, email: mps@mpssociety.co.uk

BMT for Mckayla and Cody

Mckayla (pictured right) and Cody (pictured left) have a diagnosis of MPS I. Kathleen Duncan was pregnant with Cody when Mckayla was diagnosed. Pre natal testing confirmed that Cody was also affected. Both have undergone a Bone Marow Transplant, Mckayla had hers in Scotland and Cody had his at Manchester.

Day out at Polesdon Lacey

On a very, very cold Sunday in January we (Sally, Tim, Will and Sophie) met up with Jan, Bob and Nathan, Donna, Alan and Luke and Liz, Tom and Bobby for a day out. We met up at a National Trust property in Polesden Lacey, Surrey to have a catch up and a walk around the grounds. It was absolutely freezing and everyone had extra layers on! We started off with coffee in the cafe there, which involved a bit of furniture shifting to accommodate the wheelchairs and numbers. Luke, very swiftly and without warning, managed to nearly steal an elderly lady's scone!

After a warm up we then went off for a wander around the grounds. We stopped for a picnic (there was no fight for picnic tables as nobody else was out that day for a picnic!) and then back to the cafe for a warm up cup of tea! The cafe unsurprisingly was now full so we sat outside with our warm drinks and chatted. It was so lovely to meet up and have a catch up. We will be doing it all again soon and will be hoping for a sunny day!

Sally Summerton

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

34km on new straight legs

I am fifteen and, because I have Hunter's, my legs were not growing very straight. When I reached my teens I started to grow faster, helped by enzyme, but I was still knock-kneed. I also started to get more pains on the outside of my knees, below my knee caps. However, a year ago I had a fantastic operation that has meant that my legs have now grown straight again.

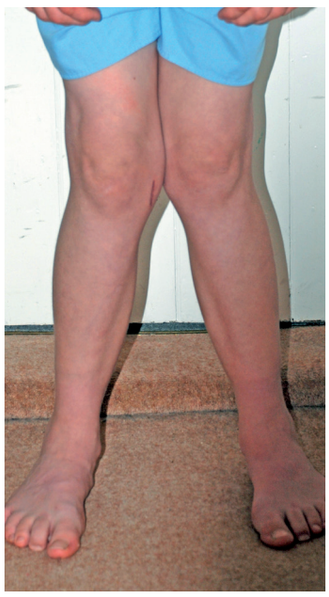
At an MPS Conference my Mum heard about an "eight-plate" operation that Miss Eastwood, an orthopaedic surgeon at Great Ormond Street, had used successfully to straighten legs.

I met with Miss Eastwood and, because I was still growing, she was confident that the operation would work for me. It involves screwing a very small metal plate into the side of the bone in either leg just below the knee on the inside. The metal plates stop one side of each bone (the inside) growing, whilst the other side (the outside) continues to grow – forcing the leg to straighten.

I wasn't completely relaxed as it sounded pretty painful, but was keen to try it as I know how much trouble Tom, my older brother, has with his knees.

Miss Eastwood and the team at Great Ormond Street made the operation seem easy. I was only in hospital for one night and was on crutches for ten days afterwards.

Now a year later I have grown and my legs feel and look completely different as you can see from the photos. I don't have pain anymore, though I do still get a little stiff and sore if I do too much exercise.



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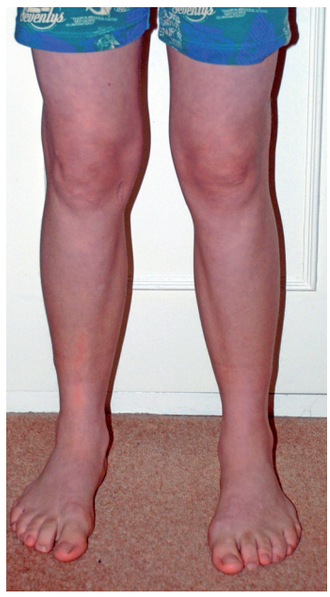


After

Amazingly, last Summer I even completed a 34 km trek over two days for my Duke of Edinburgh Award with a team of school friends. I did have to dose myself up with neurofen and had stiff joints the day afterwards but would never have managed it without the operation.

Whilst I will probably have to have the plates removed in a second operation, the treatment has made a huge difference to me – but it only works on people who are still growing. Having straight legs has made me taller than I would otherwise have been too.

By Louis Garthwaite

Left to right: Tom and Louis

Wedding bells for Mike and Fiona Illingworth

Mike and I got together in 1999 and in 2002 I became pregnant. We were ecstatic when given the news that we were expecting twin boys and as any expectant couple, looked forward to a happy life together with our boys.

We decided to wait to get married until the boys were old enough to enjoy the day with us but in 2005, our world was devastated when we were given the news that our sons had MPS (Sanfilippo Syndrome). We lived day by day and wedding plans were not thought of.

As the years passed by we slowly accepted that our boys would not be with us forever and the subject of marriage came up once again. We desperately wanted the boys to be included in our special day and enjoy the day whilst still mobile. We could not think of the day without them. They are our LIFE.

In September 2010 we decided to set a date for January 2011. This gave us three months to prepare for our BIG day. The boys had no understanding of our plans.

Saturday, 15 January 2011 came along so quickly and I have to say the day went absolutely perfect. The boys sat well while our friends and family awaited my arrival. During the part of the service when the Registrar says "Does anyone present know of any lawful impediment why these two people cannot be joined together..." Ben started to shout out (in a happy way) but thankfully the Registrar just laughed and carried on with the service.

Here we are with our gorgeous boys signing the Register. We are very happy to share this special moment with all the MPS families out there.' **Fiona Illingworth**

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

Ian's column

Ian continues his series of articles written for the MPS Magazine sharing his experiences of living with Fabry

Well, where do I start? So much has happened since I last wrote to you all. As you probably know by now I had quite a serious stroke on 21 September 2010 which has had far more effect on me than the first stroke.

But let me tell you about France. We got back from France on 20 September so I was not home very long before I had my stroke. France is a place I love. This time we stayed in a place called Berni Riveair, it was beautiful and about one hour drive north of Paris. The big thing was that I was still suffering from my first stroke which had left me with difficulties in walking so it meant using my electric scooter. I had recently got my new car which was adapted to carry my scooter and I was excited to see if it was going to make travel easier.

We traveled down to Folkestone to cross the Channel on La Manche (the shuttle). Having used the shuttle many times with trucks and coaches, both Mark (a fellow driver and friend) and I were interested in going across by car. It is not as good as there's no place for refreshments and only one toilet on the train!

Having traveled many times to France I was excited but to be honest never as a wheelchair user so I was also apprehensive to say the least. I can honestly say I had never seen a wheelchair user in all my trips there. So, we started our holiday by looking around the camp (we stayed with Keycamp). After we had settled into the mobile home we had for the week, I used my scooter while Ann and Mark walked around. The first thing we noticed was that on this site they had wheelchair friendly mobile homes with ramps right to the front door. We could not see inside one, we have to wait till May when we go again to try the disabled mobile home.

We had a busy time. While we were there we went into Paris a couple of times. It is an amazing city but parking can be a bit of a nightmare. The blue badge parking scheme is well respected and you don't get row upon row of parking for the disabled but they will have one or two disabled parking places. However, nobody will use them unless they have a badge and they are much harder to get in France than in the UK.

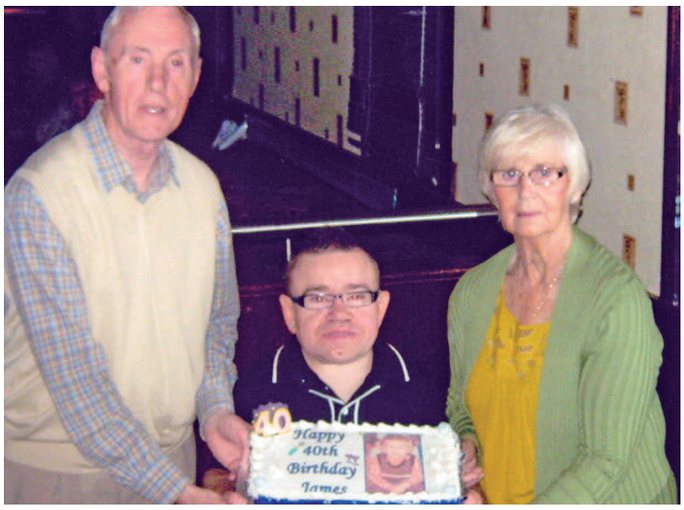
As you may know already, there is a lot of history in France and seeing it was part of my holiday but I was really unsure how I would get around especially places like the Palace of Versailles. But I was very surprised! Firstly the disabled person and the carer gets in for free. Not only that, but there are staff on hand to help you get around and, yes, King Louis fitted lifts - how good is that (hehe!). So it was a nice holiday and I am looking forward to returning in May. We got home Monday evening and straight to bed. The following day I expected to be tired and I was, but there was something else not quite right and I could not put my finger on it. This went on all day then at 8.30pm when returning from the bathroom I collapsed and could not get up. So after some time I was on my way to A&E where I spent an hour or so. Then I was sent home after being told I was overtired from my traveling.

So it was home and to bed. When I went to bed the feelings were terrible. I could not sleep but I could feel my body shutting down. In the end I could feel the fingers on my left hand but I had no control or feeling from the rest of my body other than a feeling of being squashed.

So with blue lights flashing I ended up back in A&E where there was no question about being admitted. After 24 hours I was on the acute stroke ward under my previous consultant. The care was very good and for a time I was under the intensive care unit. Where am I today? Well, as I have said I needed a wheelchair from my first stroke but I was getting better. Now, however, I am limited around the house to using a zimmer frame so a wheelchair is a must, but I am determined to get around without my frame.

My employer asked yesterday if I thought I could ever drive a coach again. If I have anything to do with it I will! I am back in work just 2 - 3 hours a week but I'm back and doing what I can. The staff have been very good and help me get around the very cramped office, even accompanying me to the toilet just in case I fall as you have to go outside to them. I have fallen a few times breaking a finger in the process, but as a sign in the rehab center shows 'recovery does mean falling'!

James celebrates his 40th birthday



Hi everyone, my name is James Stewart (no, not the famous Hollywood actor, just little old me!). I'm writing this article to tell you a little bit about myself and primarily what impact the enzyme replacement drug Elaprase has had on my life.

So, I guess the start is as good as any place to begin! I was born in 1970 in Altnagelvin Hospital, Derry. I was my parents' seventh child and already had five older sisters and an older brother. My mum knew from the first moment that she held me that there was something just that little bit different about me.

I was consequently diagnosed with MPS II Hunter disease when I was three years old. This diagnosis was made at the Royal Victoria Hospital in Belfast by Professor Nevin. He explained some aspects of the disease to my mum and dad but at that time I was the first and only child diagnosed with MPS in Northern Ireland.

Despite suffering from my disease, I had a very full and happy childhood, although I did suffer from some health problems, especially leading into my teenage years. I was always very prone to colds and chest infections and general respiratory problems and continually received physio to help reduce the build up of phlegm. Later I also developed sleep apnea which meant I had to wear a c-pap mask every night whilst sleeping. At this time I was also using oxygen twelve hours a day. This was a dark period in my life when I constantly felt unwell. It was decided at this time that I should start on a course of Elaprase and had been taking the drug for approximately eight months, when I awoke one morning with a severe pain in my arm. I was taken immediately to our local hospital where I was diagnosed with a collapsed lung. It was decided by the medical team that using the c-pap machine every night may have resulted in me suffering from a collapsed lung, therefore it was decided that the best course of action at that time would be for me to stop using c-pap. Consequently I underwent lots of respiratory tests and when the results came back they were so positive that the consultant was extremely happy and I was able to stop using oxygen and c-pap completely.

This was great news to me, as a result of the oxygen being removed I found myself with a renewed sense of freedom as I wasn't tied to my home and I was able to get a good night's sleep (so was mum!).

I continued on my enzyme replacement therapy, Elaprase, and a year later I underwent more respiratory tests again. They like to keep a good eye on me! The results of those tests were fantastic, everyone, especially myself, was ecstatic. I hadn't felt so well for a very long time.

I have now been on my treatment for almost four years and as a result my life has changed completely, for the better of course.

I have not suffered from sleep apnea for almost three years, my spleen and liver have greatly reduced in size so I no longer have a protruding tummy and no longer look like I ate all the pies! My respiratory health has also improved dramatically and as a result I feel stronger. Personally I feel that my hearing has improved also and my hands have lost their claw-like appearance somewhat. Another positive outcome from my treatment is the change in my posture. I definitely now walk taller and straighter, thus my mobility has greatly improved as well. My skin is a lot softer and my hair is less brittle (I've saved a fortune on products!). I've also lost a little weight. I genuinely feel that all the positive changes that occurred in my life during the last few years can be attributed to the introduction of enzyme replacement therapy to my life.

I recently celebrated a huge milestone with the occurrence of my 40th birthday. Needless to say we had a huge party, all my brothers and sisters and nephews came from London, not to mention all my lifelong friends (you've never seen so many good looking people in the one room!). I never would have dreamt that at this stage in my life and at the grand old age of the big '40' that I would be so physically well. I feel blessed and realise the impact that the enzyme replacement therapy has had on my illness. So, I say it's onwards and upwards from now on, there will be no stopping me now, so watch this space guys!

MPS awareness through art

Natasha

As a student of Fine Art I have learnt that the best of my work comes when I express a subject which I am passionate about. Mucopolysaccharide is one of my main concerns throughout my work. I am simply passionate about it and how it affects peoples' lives. Living with a sister who has MPS I I see the effects of how difficult it is for her growing up as a teenager in today's society. I see the emotions she goes through behind closed doors which the outside worlds don't get to see. Anger, frustration and misery, it may seem a bit morbid and of course there's an amazing personality that is bubbly and outgoing beneath these outbursts of emotions but this is the reality on her 'bad days'.

Due to the progression of the disease Chloe now stands on her toes permanently with her heels six inches off the floor. I don't think her health is her biggest concern but more so the effect that the condition has had on her appearance. Any teenager would tell you that the way they look is important to them. There is so much pressure put on teenagers to look a certain way, a way that society accepts as 'normal'. I don't like how society and media has the power to decide what is beautiful. In my eyes the human form is the most wonderful aesthetic, different interests me and the more unique the better. This is why I believe that children with Mucopolysaccharide are stunning.

This exhibition gave me the chance to bring awareness to the audience about the rare disease. Any opportunity to promote awareness I welcome with open arms. I realised that it would be in an unusual context but thought it would be interesting to see how people with a completely fresh mind and with no understanding of the disease interpret the work. I put together recordings I did of Chloe's feet, strapping the camera around her ankles gave the viewer an insight of how Chloe's feet look from her point of view. The video piece is accompanied by sounds of her weekly trip to hospital as well as an outburst of her anger and frustration. My parents had attended the second All Ireland paediatric meeting and seen Chloe's x-rays of her feet. I thought that by including the x-rays of her feet it would be a great way to show people the severity of her feet. I traveled up to Belfast to take photographs of the x-rays. Seeing the x-rays was an amazing experience and I also found it very emotional. It really moved me how beautiful her feet had been formed and I could not wait to share this with others.

Showing my class mates the x-rays I got feedback that I was looking for, amazing, shocking, unbelievable and absolutely beautiful were all the words I was hoping for. I respect Chloe and understand that it is a very personal and intimate issue for her, particularly her feet. I am proud of her for giving me the responsibility to show others how the condition has affected her appearance. This was a brave thing for her to do and I am delighted at the success of the exhibition.

Chloe is my idol, she is my muse and I couldn't think of anything more magnificent to have so I am a lucky artist and a lucky sister. **Natasha McCauley**

Chloe, Eoin and Mum and Dad

The journey continues with Careology by our side

As the short days draw to a close, the lighter evenings are a blessed welcome to what felt like the longest winter in history! After battling every cough, cold, ear infection, upper and lower respiratory infections and sickness bugs, we are finally starting to feel like our GP surgery are happy to see the back of us until October! We are seriously considering a move to the Floridian coast where the sun always shines!

Our year started with a long and drawn out flu for both Caleb and Mark. Christmas 2010 was not the most memorable I have to say. Trying to deal with all the illness that winter brings, as well as trying to still get our heads around Hunter Disease is stressful, add to this Man flu, Mum flu and a poorly little monkey and you have a situation on your hands! We really hope that as time goes on and Caleb's body gets used to the Enzyme Replacement Therapy we will start to see less of the ear infections, virus's and colds.

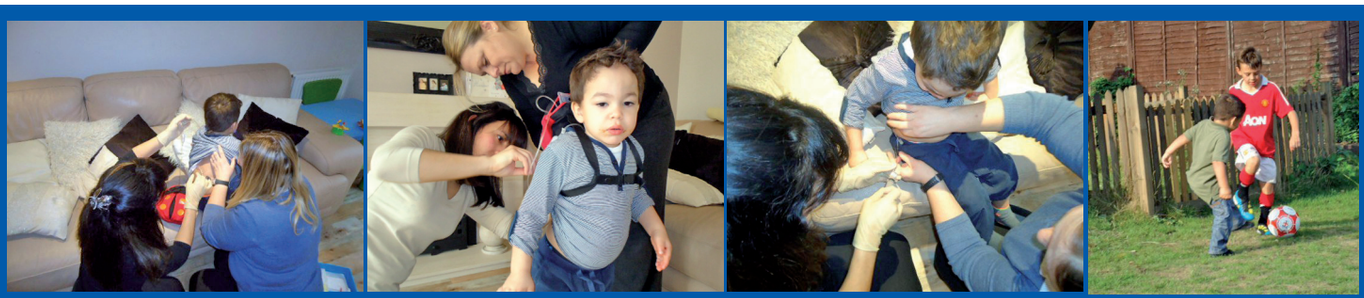
Something that has served as a very good distraction to us all is having such great support from Careology and our wonderful nurse Jitka Pils. After watching a very inspiring video shot by Shire which focused on our friends the Stevens family all promoting the benefits of independent infusions Mark, Caleb and I were truly inspired.
The Stevens spoke of the ability of obtaining some normalcy back into life with ERT without having to adhere to specific dates and times for nurses to come round and administer the Enzyme. Currently Caleb misses one morning of pre school which at the moment is manageable but over a long term period this could prove to be an issue when he starts full time school. Becoming independent would mean Mark could have his whole day at work back, Caleb will not miss out on his education and our life would have a slightly more consistent routine which we are ultimately in charge of.

So, on 2nd February 2011, we started to learn how to prepare Caleb's weekly infusions. Working closely with our nurse Jitka, each week we go through the process of becoming independent step by step. Mark and I start each Thursday by preparing Caleb before Jitka arrives. Caleb takes his pre meds of Calpol and Piraton without any protest, but really doesn't like plasters, so after much coaxing, and bribing with chocolate and Shrek DVD's we manage to put the Emla Cream on to his port which needs about 45 minutes to activate and numb the area. This in itself is a huge achievement as Caleb is faster than a speeding bullet and trying to get him to keep still is very challenging!

Jitka usually arrives at 9am and without wasting any time the process of preparing the access tray begins. It's important to keep our area and our hands as sterile as possible using the 6 step hand washing technique. Once this is done the tray is prepared ready for port access.

Accessing the port is probably the most daunting part of the process for us as the thought of pricking your child with a sharp object is enough to make any parent recoil in terror! Putting the first ever gripper in was terrifying and exciting at the same time. The first time I completely froze and panicked and Mark had his eyes closed the whole time! Jitka had to rescue me and reassure Mark that Caleb was not feeling any pain! I should have realised this as Caleb remained calm and collected during this first time and totally engrossed in watching Shrek! The second week it got easier, I felt a little more confidence and Mark became more comfortable with keeping his eyes open. Each week we found the more relaxed we became and the more we practised, the whole process has now started to feel more and more familiar. Recently, I accessed the port with no panicking and no worries.

Throughout all of this process it helps immensely to see how relaxed Caleb is during all the kerfuffle of me trying to remember the right steps, the draw back, the flushing and the clamping! In fact, his relaxed demeanour is what is spurring me on to master the art of being Caleb's very own personal nurse. Caleb trusts me implicitly and to watch his little face light up when he realises mummy is doing all this is just so wonderful. We still have a long way to go in our training, but we are making progress and we feel like we are doing something to help our little monkey Caleb. **Daniella Vandepeer**



Gloucester parents use the internet in son's diagnosis

Joseph

Rich and Mel Tilling have two children - Erin, 12, and Joseph, 2.

The Gloucester couple have used the internet to help diagnose a rare condition in their two-year-old son.

Rich and Mel Tilling, from Linden, used online research to encourage doctors to reconsider a disease that had already been ruled out after tests on Joseph.

The couple found information and photos that matched their own experience.

Joseph has a rare life-limiting storage disorder called Mucolipidosis II, which means the body can't break down waste products that build up in cells.

Whilst researching online Mel said she "came across a little boy who just looked the spitting image of Joseph".

By this time Richard said "we needed answers because obviously it was a huge worry".

"You've just got to get on with it, make the most of what you've got, make special memories and have as many happy times as possible."

"We were then going to the doctors saying 'is it this?' or 'is it this?' They didn't want to tell us because they didn't want to worry us if it wasn't any of these things," he added.

Now at two years old, the symptoms Joseph has are mainly skeletal.

Mel said: "His hips haven't formed properly, his shoulders are frozen and his knees are starting to fix in place now although he is not going to be able to walk."

"As the disease progresses it can affect his heart, his eyesight, his hearing - the list is endless."

They were both devastated to hear the news, especially that they might outlive their son, but said it's no good being sad about it now.

"You've just got to get on with it, make the most of what you've got, make special memories and have as many happy times as possible," said Mel.

The family said it had been so well supported by The MPS Society - a group which represents those affected by storage diseases - that a number of friends have decided to cycle from John O'Groats to Lands End to help the charity.

"We wanted to do something to raise awareness for the MPS because it's such a rare illness," said Chris Taylor.

"A friend of mine and me, down the pub one night, decided to cycle from John O'Groats to Lands End."

"It then progressed to nine of us, none of whom have any cycling experience, so it should be quite interesting!"

Article appears courtesy of BBC Gloucestershire
http://news.bbc.co.uk/local/gloucestershire

Can you help with our media campaign?

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

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

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

Great Ormond Street MPS III Clinic

17 December 2010
This was my first experience of the Great Ormond Street Hospital MPS III clinic. I attended the clinic with Jolanta and met with the collaborative team of professionals working with our young people and their families.

The ambience of the waiting area was relaxed and cheering which I believe encourages socialising and sharing. As a parent myself I always found that meeting and becoming involved with other parents really relieved parenting anxiety, practical solutions, other people's experiences and a good sharing chat could lift my spirits. As I gained in experience I was able to then help others on their pathway of parenting.

I looked at the little tables with all our children playing and chattering and it warmed my heart. Later on one of the parents crouched by a board on the wall encouraging the children to fill the gaps with the tiles. The Little Tykes Cosy Coupe was whizzing up and down and around and away as it often is, leading to my contacting Little Tykes to find that yes there is a play vehicle made which fits taller children.

While the children played, the parents were able to chat and share in between racing off to bring back escapees and it was lovely to learn how close families become.

We all had had our differing journeys to reach the clinic, one family had travelled up the night before, so the travelling and weather conditions were a topic of conversation amongst the adults. Unfortunately we discovered our camera had broken when we arrived, one Dad emailed us pictures of his son to include in the magazine.

Reflecting back on the clinic I realised and remember it as a smiling day and enjoyed meeting Dr Vellodi, Niamh and Victoria, Michelle and Sonia.
Lindsey Wingate l.wingate@mpssociety.co.uk

Photos above: Tommy Thompson (MPS III)

Manchester BMT Clinic

14 January 2011
A very dark and early start for my first under 6 BMT clinic which I attended with Jolanta.

We arrived in plenty of time and had a chat and catch up with Jean Mercer before the first of five lovely children arrived for their appointments.

We met with Morgan and her family first and what a delightful, engaging little girl she is, full of mischief and fun. A big welcome to Morgan and her family, who are new members.

Next came the whirlwind that is Luke - who, along with his brother Quinton and Dad, built a huge tower from the building blocks in the play area. Before long everyone was engaged in this activity, much to the consternation of some of the staff! Luke is looking so well.

The composed and extremely fashionable Lyla arrived, who looked amazing and obviously has a real 'eye' for fashion – what a lovely young lady.

Mikko then arrived looking very smart in a striped rugby shirt. Congratulations to mum and dad on the arrival of Jani, Mikko's baby brother.

Finally we met Jake, who is also a new member. Jake wasn't too impressed with life on that day and just wanted to be snuggled up with Mum.

Thank you to the team for looking after us and to you, the families for letting us share your amazing children with us for a short while. **Tina Bough**

21 January 2011
I was so pleased on my first ever Manchester Bone Marrow Transplant 6+ years clinic to discover purple-ness in the waiting area. The sun was shining, the staff were warm and welcoming and everything joined to build a cheerful environment.

The cheery environment developed even more once our lovely families arrived. Beautiful smiles and impish light-hearted banter filled the waiting area. I saw parents waiting with their children for other clinics smiling at the animation being shared around.

Rubina came with her parents and had a lovely cheerful chat with the nurses; she showed us her beautiful badge and pretty outfit.

It was lovely to see Thomas's bright smile and how well he was mobilising, Mum has been busy adapting his clothes to accommodate Thomas's halo.

Leighton came with his Mum and Nan and informed us he would have two cakes for his birthday! I'm envious of Leighton, particularly over the chocolate cake.

Matthew along with his Dad was telling us about when he will learn to drive. He also wants to design a new pair of trainers for himself and made us all rock with laughter at his stories about his younger sister.

I must say thank you to everyone for a successful welcoming clinic and I look forward to my next visit to Manchester. **Lindsey Wingate** l.wingate@mpssociety.co.uk

Photos clockwise from top left: Rubina Jalani and Leighton Barker (both MPS IH) from the over 6s BMT clinic.

Bristol MPS Clinic

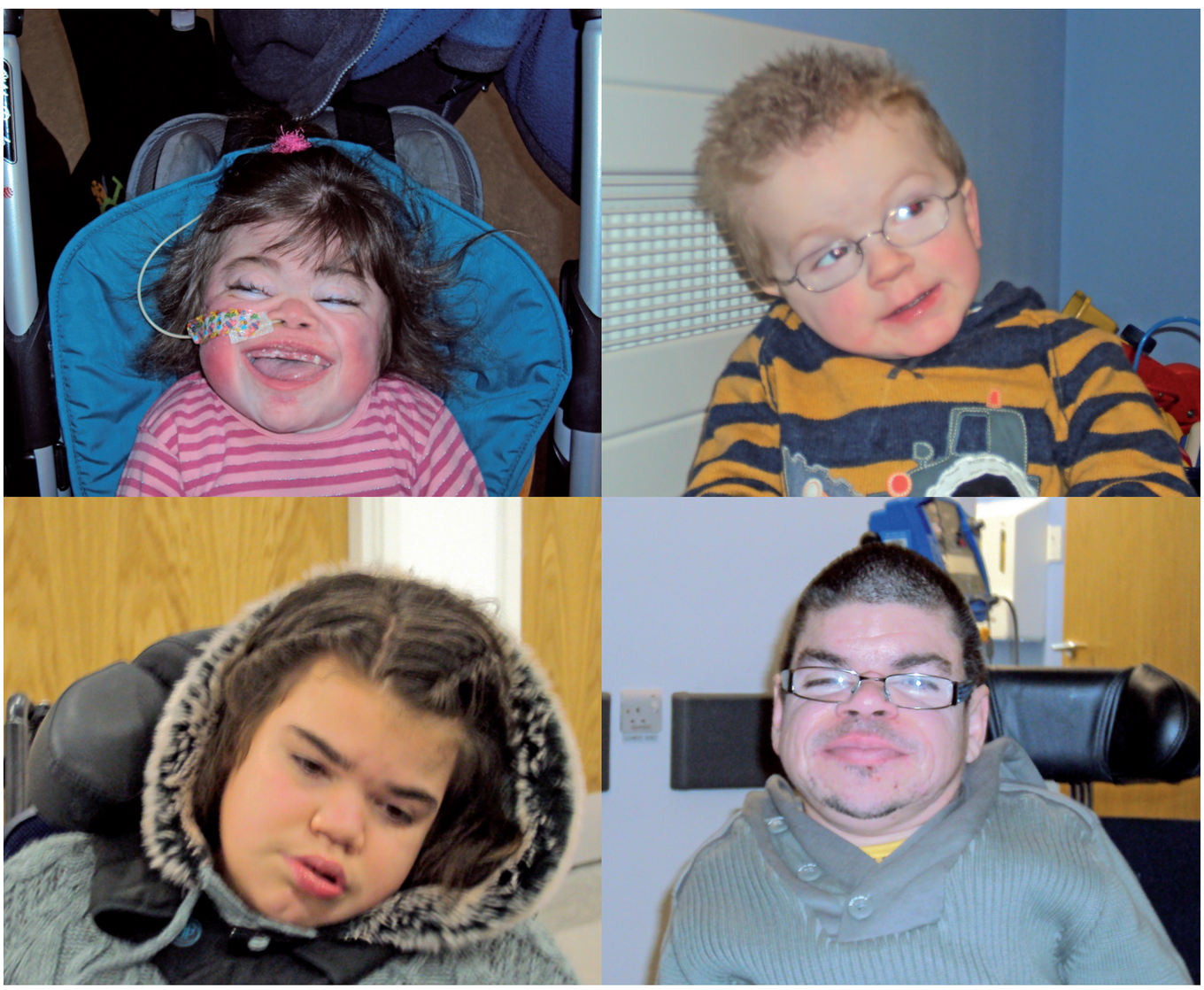
18 January 2011
Although it was just the beginning of the year the Advocacy Team was out and about for the whole month with four MPS clinics and many home visits to attend.

In the early morning on 18 January I set off for Bristol for the second time at a New Year MPS Clinic. I was very excited because all the families were fairly new to me. The whole journey went smoothly but finding a parking space turned out to be a challenge. After four and a half car parks later we eventually found a space, memorised coordinates and bought highly deserved and much needed coffee.

The first to arrive for the clinic was Christian with his parents. In a nonchalant way he said 'fine, OK and cool' to everything, even some teasing about his Manchester United t-shirt. Next one to say 'Hello!' to us was Tara, with whom I had a chat about the hairstyles, clothes and many giggles. The last but not least to meet was Lewis, who didn't find bright orange walls in the waiting area to his liking and desperately wanted to run leaving his parents behind.

It was lovely to meet all the children and their parents. We hope that we'll see all the families which couldn't make it this time during the next clinic.

I would like to thank Dr Pierre, Dr Jardine, Prof Wraith and the team at Bristol Children's Hospital for another successful clinic!
Jolanta Turz j.turz@mpssociety.co.uk



Photos clockwise from top left: **Cardiff clinic** - Abigail Harvey (ML II), Ethan Greening (MPS I), Gavin Hyde (ML III), Georgia Lewis (MPS III)

Cardiff MPS Clinic

20 January 2011
I am going to start the write up on my first Cardiff clinic without talking about the weather as we more than often do!

Sophie and I arrived in good time, and found lots of parking, which I am told is not the case later in the day. Luckily, we had found out that the clinic had moved to the main part of the hospital, however, this was not so for the families who felt as though they were on a route march through the maze of corridors. Ten out of ten to all of you for finding the new location! The new clinic area is bright and airy with chairs and tables, and of course, lots of toys.

Dr Shortland and Professor Wraith were taking the clinic, and first to arrive was Ethan with his mum and dad, he had great fun in the toy corner, investigating everything.

Abigail came with her mum and dad and had a treat in store later on in the week, off to see Father Christmas on the train.

More families and carers arrived; Marshall, played happily, Christopher and Steven had grown into young men since Sophie had last seen them. Georgia came with her mum and looked very trendy with her fur lined cardigan. Sonny just wanted to play with everything… Sarah loved her new I Pad which she had as a Christmas present, she also had a quality street tin of scoobies which she had made, the question is did she eat all the chocolates herself first? Gavin and his sister Sarah arrived after doing the detective work to find the clinic, followed by Megan and her dad.

It was soon time to take the long trip back home and we got engulfed by the fog on the toll bridge, oops, wasn't going to mention the weather…

Thank you to all the team and the families for making my first trip to Cardiff enjoyable.
Rebecca Brandon r.brandon@mpssociety.co.uk

Photos clockwise from top left: **Cardiff clinic** - Marshall Dale (MPS III), Sarah McKnight (MPS I), Sonny Gibbard (MPS I)

Birmingham MPS Clinic

18 February 2011

Well, it's another first for me… I haven't had the pleasure of attending the Birmingham Clinic until now. Jolanta and I arrived in good time and met Viki and Louise all ready on the starting blocks, but no doctors, after tracking them down we were ready to go. The waiting area is in the outpatients department and you can imagine the amount of people and hyper children and young people.

Will was first to arrive with his dad who is so pleased with his progress. Will is even overtaking dad with using technology (dad's words, not mine)! Will was quite happy being wired for sound and looked like a little DJ with his headphones on.

Ali was straight over to the table football and I thought he would be too preoccupied to have his photograph taken, but no problems there…

Luke came with his mum and granddad and met up with his friend Will. Luke is lucky to have a granddad who is a dab hand at growing orchids and he has one named after him, not many people can claim that!

Double trouble arrived in the guise of Nazia and Sophia, who were dressed in lovely bright colours. They were soon joined by Sumaira and with the three of them you didn't get a word in edge ways.

Sumaira then sat down and did a lovely picture for an eye competition being run by the hospital.

Thomas came with his mum and dad but was not happy… he doesn't like hospitals! However, he soon settled down and cheered up.

Caitlin came in like a little whirlwind, singing happily, and kept everyone on their toes as she went to investigate the book corner.

Sultan arrived totally unfazed by the fact that Louise had been on a route march to try and locate him down in another part of the hospital. He looked very smart in his school uniform and was very chilled…

It was soon time to go and tackle the traffic home. It was lovely to meet the team and all the children and families and I look forward to seeing you all again in the future.

Rebecca Brandon & Jolanta Turz

Photos clockwise from top left: Ali Anwar Khan (MPS IV), Will Brodie (MPS II), Luke Edwards (MPS II), Sultan Ali (MPS IV), Sumaira Begum (MPS IV), Sophia and Nazia Khan (MPS IV)

Scottish post-Christmas-blues party

On a dreary Sunday morning in January we made our way to Stansted Airport for what would be a truly fun day out!

We landed at Edinburgh Airport and were taken to the Hilton Hotel. People thought we were mad, when they asked us why we were going to Edinburgh, and we replied "for a Christmas Party of course", or officially "a Post-Christmas Blues Party"!

When we arrived at the hotel we were taken to our "Party Room", which had been transformed, full of streamers, party hats, crackers and the such like.
Who cared that it was January?

Once the families started to arrive the atmosphere was buzzing. With children making use of the space, performing their kart-wheels and forward rolls. Once everyone had said hello to old friends and introduced themselves to new friends, we all sat down for a delicious Christmas Dinner. During the lunch we pulled crackers and watched in awe at the magician (and his band of merry men / followers)! The magician walked round the tables making balloon "everything" for all the children. Anything from a bonnet to an Octopus, (including the odd alien here and there).

After lunch we all retired to the other end of the room to watch a magic show, where the assistants played an excellent part. Audience participation was a must with shouting and clapping and lots of giggles.

This was proving to be a great Christmas Party, you could almost forget that it was January. But there was someone missing! A long awaited "Special Guest". Fresh from his holidays in Barbados, no rest for Father Christmas. With his reindeer safely chilling out in the departure lounge, Father Christmas arrived laden with gifts for all the girls and boys.

We hope you all had a lovely afternoon, and the post Christmas blues are firmly blown away. Here are a selection of photos from the day. **Fiona Hopson**

Rare Disease Day

On 28 February 2011 nearly 200 individuals with rare diseases and representatives of Rare Disease patient organisations came together for tea on the terrace of the Palace of Westminster to celebrate Rare Disease Day.

One of the main objectives of the event is to draw media coverage, as well as to raise awareness of rare diseases and their impact on patients' lives amongst the general public and decision-makers. We heard from Alistair Kent, Executive Director of the Genetic Alliance on the need of Government, health and social care services to recognise those with rare diseases to be treated equitably with those suffering more common conditions and launched the 'Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy'. Earl Howe spoke on the Government's commitment to adults and children with rare diseases and Liz Kendall, Shadow Health Secretary encouraged those present to contribute to the consultation on the Health and Social Care Bill.

This gathering enabled me to network with many old friends from the Rare Disease community as well as make new contacts. It was a pleasure to catch up with Lesley Greene, founder and President of CLIMB as well as Nigel Nicholls from BioMarin (pictured below with Christine).
Christine Lavery c.lavery@mpssociety.co.uk

Health and Social Care Bill

Please do consider reading the Health and Social Care Bill which received its second reading on 31 January 2011. If you should wish to comment or provide evidence on the bill please use this link: http://www.parliament.uk/business/news/2011/january/health-and-social-care-bill-second-reading-/

Help
The Scrutiny Unit can help with any queries about providing written evidence. Contact details are as follows:
Telephone: 020 7219 8387
Email: scrutiny@parliament.uk
Fax: 020 7219 8381
Michelle Edney, Senior Executive Officer, Scrutiny Unit, 7 Millbank, London SW1P 3JA

For written guidance on providing evidence on the Health and Social Care Bill http://www.parliament.uk/business/news/2011/january/health-and-social-care-bill-second-reading-/guidance-on-submitting-evidence-to-a-pbc/

Rare Diseases Day Event

Royal Holloway College (University of London)

Monday 28 February 2011

Royal Holloway College (University of London) held an event on their campus at Egham, Surrey to mark Rare Diseases Day on Monday 28th February. Scientists at the College are playing a leading role in developing safer methods of gene therapy for rare diseases. In particular they are at the forefront of "Genome Surgery", which attempts to repair rather than replace defective genes, thereby avoiding some of the problems of giving a patient an extra copy of a gene.

The Event consisted of talks about the research going on at Royal Holloway, visits to the research laboratories, and a showing of the film, Extraordinary Measures, which is a dramatisation, starring Harrison Ford, of a family's struggle to get enzyme replacement therapy for Pompe disease. There was also an Exhibition in the wonderful Picture Gallery at which Patient Support Groups could present their work.

Bryan Winchester, a Trustee, who has lectured to the students at Royal Holloway on MPS for the last 20 years, manned the MPS stand. The people attending the Event were mainly students already studying biomedical science or very enthusiastic 'A' Level students hoping to study medicine or biology at University. Bryan spent a very enjoyable and exhausting two hours telling the young people about MPS and the work of the Society and encouraging them to become volunteers!

The Event concluded with a Round Table Discussion on Rare Diseases involving the research scientists, patients and their families, the Support Groups and the students, the potential clinical researchers of the future. Royal Holloway College is to be congratulated on organising such an informative and enjoyable event. The Event was largely run by the undergraduates and it would be good for the MPS Society to become more involved with a local university.

MPS Awareness Day

15 May 2011

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

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

Help us celebrate International MPS Awareness Day on Saturday 15 May 2011

In 2011 we're asking all our members, Friends and supporters to do something, big or small, to mark MPS Awareness Day

Visit **www.mpssociety.co.uk** for more information or give us a call on **0845 389 9901** to find out how you can support us...

Helen's volunteer experience

I care passionately about children; this passion stems from the fact that at six months old my mother put me in a care home and I was adopted when I was almost a year old. I was adopted in an attempt to paper over the cracks in their marriage; I was never really the answer to these problems.

I believe that all children are valuable and when I heard about people from my church helping to care for children on an MPS weekend I was keen to get involved. I have four lovely children and as soon as the youngest was old enough to be left with my husband I started my involvement with MPS and over 20 years later I am still involved because I still care passionately about children and want each one of them to know that they are special and valued. This passion means that I am also involved in a volunteer capacity with the 'Toybox Charity' that works with street children in South America.

I have become aware how difficult it is for families with children who have MPS. By acting as a volunteer for the MPS Society I hope that the families know that there are other people who care and value them and their children. I would like each family to know that they and their children are special and that there are other people who care. Hopefully by my continued regular involvement families and children will know this.

The weekends are great fun, well organised and thought out so that as the volunteer you never feel alone or left to sort out problems by yourself. As a volunteer you too feel valued, cared for and supported by staff and other volunteers. You grow to realise that the families value your time and care how important your small offer of time is to them. Their children are valued and loved by them and it is an honour to share for even a small time in the care of their children. Each child is special and when you volunteer you realise that you can be part of their lives for a short time. This enables mum and dad to focus on other things assured that their children are having fun.

I know that each MPS weekend will be a tiring event. I go to give and show love and concern. When an MPS weekend is over I can go back to my family; we have been tremendously blessed, within a few months all my four adult children will have left home and are pursuing their own careers. The tiredness that a weekend brings is a small price to pay for the help that has been given to families challenged by MPS.

By profession I am a teacher; at one time my husband and I looked at alternative careers for me but at every turn my passion for children brought me back to teaching. As I approach retirement and despite owning a property in the Algarve I plan to continue my involvement with children; my focus is on helping children and families who need help. I am therefore hoping to be involved with the MPS for many more years yet and to get involved in other projects that focus on helping children.

My life did not start in an ideal way; along the way a number of people helped me to become the person I am today. I hope that in some way I can bring help and comfort to others because I know how important that help can be in transforming lives. As a long time volunteer I would like to encourage others to give up some of their time to volunteer in the future. It is tiring, fun and above all rewarding to see a smile on the face of a child and hold their sticky hand as they give their trust to you as you share in a small part of their life. So go for it, I hope to see you on our next adventure be it a theme park, a zoo or the bowling alley. **Helen Patterson**

Would you like to volunteer for MPS?

Volunteering is fun and rewarding. It could also help you learn new skills and gain valuable work experience. The MPS Society relies on volunteers for our events and conferences 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. Volunteers should be 16 years or over, will need to provide two references and undergo a Criminal Records Bureau check and attend a training day in Amersham. Those volunteering for our conferences will receive accommodation and all meals throughout the weekend.

Contact us now to register your interest and availability. **mps@mpssociety.co.uk**

A day in the life of a MPS childcare volunteer

Rise early, dress in sensible clothes!
Flat shoes that stay on!
Jewellery, that you don't mind getting wrecked.
Eat a good breakfast, energy needed.
Go to the toilet while still childless.
Find that waterproof top that you told your mum you would never wear again.
Meet your family, smile, gather information.
Make friends with the children.
Go to the coach with children, buggy, nappies, drink and the information buzzing in your head.
Strapped in and away you go.
Remember all those journeys as a child when you asked your mum 'are we there yet'? What were the answers she used to give?
Singing is good, what jokes can you share?
On arrival, study the map so you won't get lost.
Find a volunteer friend to go around with, one who is used to nappies!
Put up the buggy from hell, should have listened to the dad; an engineering degree would help!
Set off into the park to go on rides that you love to hate!
How come children love things that go around in circles and your stomach doesn't!
Lunch time; yummy, sit and rest, you must be joking.
Food everywhere but inside your charge! Down your front, in your hair, how did they get it there, oh they're sharing!
Clean up time, toilet run, help where is the nappy expert!
Into the park again more rides, ones you never liked, smile; hang onto your lunch! How do these children move so fast? At least the buggy keeps up?
Why do they have hills in theme parks?
Why do they have water rides?
How many more hours to go?
Don't children get tired?
Thank goodness for those flat shoes and the waterproof you used to hate!
The gift shop calls, at least it's dry.
Was it this hard to choose when you were a child?
How many things can you buy for two pounds?
Smile at the shop assistant, it's only the tenth thing your charge has tried to eat!
It's time for the coach journey home.
Toilet stop again!
That's it buggies. Buggies should be banned unless you have an engineering degree!
Disbelief your charge has fallen asleep - that's because you can now see the hotel.
Return your charge to their family intact.
Off for a quick swim to unwind.
Dinner with your new friends; chatting over the funny moments of the day.
Baby sitting next, find the room.
Meet your charges who have bounced back into life after their tea.
Mum and dad vanish to the dinner dance.
Read stories, watch television, play games, sing songs and they're still awake!
Then as if by magic they sleep.
You sigh, smile again and sink into a chair.
Mum and dad return happy that all is well.
You stretch and wander down to meet the other volunteers for a drink, recap the day.
Then off to bed, it's the safari park tomorrow!

Hannah's Story

I've been volunteering for the MPS Society for a few years now and I love it.

One of my brothers (Dan) and my sister (Amy) both have Sanfilippo and when I was younger I really enjoyed coming along to the MPS conferences. Our family used to need quite a few volunteers and I have really happy memories of fun volunteers who gave us all such a great time. As a volunteer now, seeing the younger MPS children and their siblings brings back happy memories of when Amy and Dan were little and needed fast runners to chase them!

It feels really good to be able to give something back now. The weekends are always such good fun. What could be more fun than a hotel full of MPS kids running around and causing chaos!

It's great to meet some amazing families and spend the day with fantastic kids in the hotel or taking them out for the day to a theme park or zoo. It's definitely hard work and challenging at times and you need to be prepared to run fast and have hands that can be in 4 places at once!

Sometimes things don't always go to plan either… like when fire alarms go off and you have to evacuate all the children out of the hotel! But it's all great and by the end of the day a good night's sleep is always guaranteed! It's satisfying too; I really enjoy getting to know the kids and their siblings over the weekend. Seeing them enjoy themselves whilst knowing that their parents are able to appreciate a bit of a break is great. It's nice to feel like you've really been a help and it's really rewarding to have a parent say to you "Wow you can come again, she's gone straight to sleep she was so worn out!"

Being trusted to look after someone else's child for the day does feel like a big responsibility, but the weekends are always so well organised. The volunteers are like a big team and all work together and help each other out. It's been good coming back year after year, catching up with all the other volunteers and seeing the families that I've got to know before.

This last year it's been really nice to volunteer on the young adult weekend too. This was in Blackpool and was quite a bit different to the other weekends I'd volunteered on before. It was good to spend time talking and getting to know each of the young people and I was inspired by their positive attitude towards life, especially with the many challenges that they face and overcome every day.

I've really enjoyed volunteering at the MPS weekends, getting to meet some amazing people, experience some really interesting situations and have a great time! MPS weekends are definitely the most exhausting weekends of my year but also some of the most rewarding and fun ones too. **Hannah Donegani**

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 and 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

Clinic dedicated to MPS in India

We have always been aware that the incidence of MPS and related lysosomal diseases in India was likely to be on a par or exceed that of the Western world. With India having a population of 1.2 billion and based on that hypothesis there are likely to be over 25,000 children and adults affected. It is therefore not surprising that over 50 MPS families were seen in one day at the MPS clinic held in Chennai, Tamil Nadu State in South India on 26 January 2011.

This clinic which has been growing in size and expertise over the last five years is organised by the Fetal Care Research Foundation (FCRF) and pioneered by Dr Suresh and Dr Jagadeesh. Dr Ashok Vellodi who many of you will know better as the MPS paediatric consultant at Great Ormond Street Hospital for Children has regularly travelled to Chennai giving his time to support this specialist clinic. More recently Dr Uma Ramaswami the MPS paediatric specialist has also supported this clinic unique to the whole of India.

This clinic also is home to the MPS Support group for Mucopolysaccharidoses and an invitation was extended to me and Tanya Collin-Histed, Chief Executive of the Gaucher Association, to join Dr Vellodi and Dr Ramaswami for the 2011 clinic.

It was an immense privilege to travel to Chennai and to meet so many young people and their families affected by MPS diseases. Some families had flown from Mumbai and the North of India, others travelled on buses and trains over 500 miles to see the experts in their child's disease. It was a most humbling experience sitting in on the clinic and meeting individually with families. I was constantly struck by how uncomplaining, dignified and gracious each and every family I met was. Many tears were shed as Dr Ramaswami confirmed to the Sanfilippo families that palliative care is the only way forward. Sometimes a family would ask if they brought their child to London would they get better and sadly we shared with them that whether they live in India, the USA, England or China there is no cure and approaches to treatment are in the very early stages.

What I had not given much thought to was how Tanya and I were going to communicate with the families. We need not have worried because as there are so many Indian language dialects English is for many a strong second language. Where a helping hand was needed Dr Suresh and his team were on hand to step in.

The UK MPS Society has a long history of involving volunteers. It was a welcome surprise to find that the MPS clinic could not run without the untiring support of a large team of volunteers moving patients and their families from one department to another. In the UK we usually have two advocacy officers to support 10 – 12 families at our MPS regional clinics so you can imagine the volunteer force needed for 60 patients. Another difference was that not only did the patients see the MPS specialist but they were also seen by a number of specialities including cardiology, ENT, neurology and could also undergo ultrasound scans and X-rays before returning to see the MPS specialist for the results.

What about Enzyme Replacement Therapy? Malcolm Johnson from Genzyme was also invited to Chennai for the clinic and I soon found out that many Gaucher patients and a few Pompe and MPS I patients were in receipt of their ERT through Genzyme's compassionate use programme. This does not mean all patients with these diseases in India get treated and indeed I saw a little boy with MPS I Hurler Scheie that was not on any ERT and raised this. We saw three young men with MPS II who in England would without doubt be on ERT and since arriving back in the UK have raised their needs and the possibility of a compassionate use programme for Elaprase with Shire Pharmaceuticals. Whilst I hope we can make a difference in these young peoples' lives we need also to recognise that the Indian States and National Government also need to work towards funding ERT. To this end on Friday 28 January accompanied by Dr Suresh, Dr Jagadeesh and Malcolm Johnson we were received by the Minister of Health for Tamil Nadu State to make the case for government funded Enzyme Replacement Therapy. **Christine Lavery** c.lavery@mpssociety.co.uk

The Story of Parth

An MPS II child from Mumbai, India

I called Parth and he did not even bother to look up from his play activity. I thought that my son, Parth, who was just 22 months old at that time was having a big attitude! I told this to my wife and she said that sometimes Parth did not respond to her either.

Later we spoke to Parth's paediatrician who suggested that we get his hearing tested. The results of hearing tests showed that Parth had a severe hearing disability in both ears. We had a sense of disbelief as since birth, Parth was able to hear clearly and by the time we celebrated his first birthday, he even spoke a few words. A simple hearing aid was provided where the battery operated machine would be tucked in his pocket and a wire running to his one ear. But this was not good enough as every now and then the wire would come out of his ear and he would not be able to hear anything.

We wondered how he lost his hearing without any accident and any apparent illness. A series of visits to different hospitals and doctors followed. A range of medical tests were carried out and finally it was established that Parth was deficient in an enzyme called Iduronate Sulfatase. He was diagnosed as having a regressive metabolic disorder named Mucopolysaccharidosis Type II (also known as MPS II or Hunter Syndrome).

The prognosis was even more discouraging and disheartening:

- Sight and hearing difficulties
- Frequent respiratory infections
- Behavioural problems
- Mental retardation in the severe forms
- Bone deformities and joint stiffness
- Heart disease
- Enlarged organs such as liver and spleen
- Short stature
- And worst of all, life span not beyond teen years.

We were told by the grim looking specialist that there is no known medical cure. For two months, we tried different doctors, hospitals and medical tests, hoping against hope.

We went through a range of negative emotions. Our first reaction was of disbelief ("No, it cannot happen to Parth and us"). Next reaction was anger ("Why us and why my child?" "What has he done to deserve this?"). This was followed by self pity and despair. Our small world went upside down. All our future plans and dreams seemed to go up in smoke.

In those two months, from running from one doctor to another, we were emotionally drained, physically exhausted and financially almost bankrupt. We avoided social outings as we were tired of explaining to others what had happened to our beloved Parth and why he cannot hear.

On Parth's second birthday, we fitted Parth with a pair of advanced Behind-the-Ear hearing aids. My wife and I both cried a lot on that day having to give such a birthday gift to Parth. We still had hope. We knew that Parth is a born fighter. Parth was born four weeks premature with a birth weight of 1.9 kg. His mother had toxemia of pregnancy and the baby Parth had to be delivered in an emergency operation. The baby suffered from Pneumonia, jaundice and lowered platelet counts in the first 10 days and was very critical in that period. However, Parth survived all these and by the age of 10 months, his weight and other developments were normal. With his fair skin, blond hair and a sweet smile, he was the darling of everyone who came into contact with him.

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***

With his new hearing aids, Parth could hear and learnt to speak. When it was time to admit him to a school, we debated whether to send him to a school for children with special needs or to a school for normal children. There were special schools for hearing disabled children or mentally disabled children or for children with learning disabilities. Parth was not fitting into any category. His mental abilities seemed good and with hearing aids he could hear and even speak. Finally, we decided to send him to a school where normal children go. He did very well in the first two years. Thereafter, as the effects of the disorder progressed, he found it difficult to keep up. Even then, in June 2010, he passed our Grade X or the Secondary School Examination conducted by the State Level Board in the first attempt. He can no longer cope with the mental and physical strain of higher studies. Currently, he is getting trained in drawing which is his area of interest. While we do not expect him to be financially independent ever, we hope that in the future, his drawings and paintings are sold by NGO's (Non Government Organisations) either directly or by way of greeting cards and Parth earns something for his efforts. One of his paintings is shown here on the right.

Currently, Parth is 16 years old and has the following symptoms:

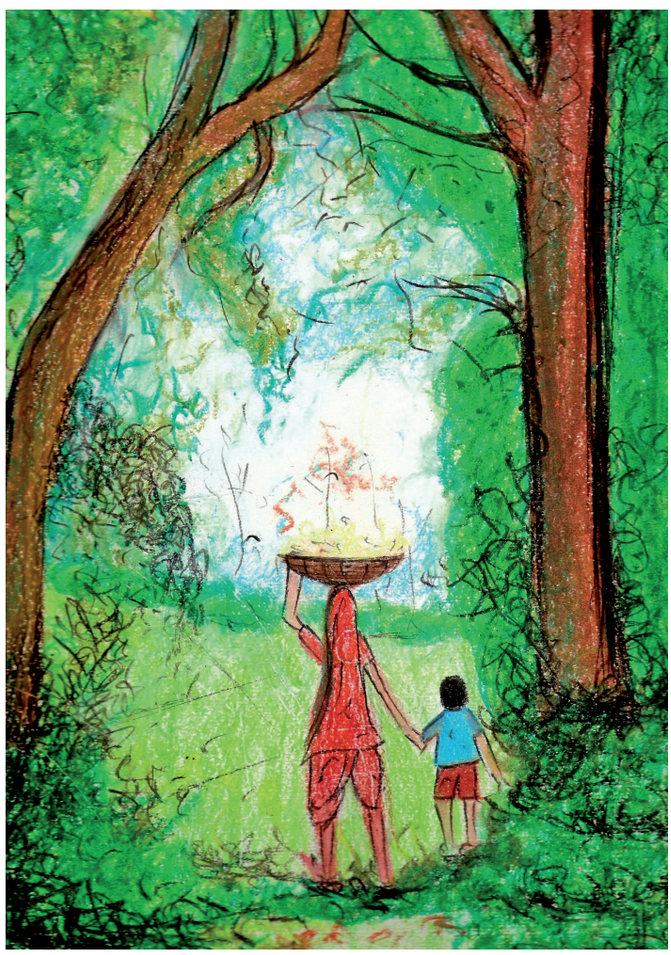
- Hearing disability in both ears to the extent of 85-90 db. (He is able to hear after fitting Behind the Ear hearing aids).
- Enlargement of liver and spleen
- Frequent bouts of cold, cough and throat infections
- Stiff joints including:
- He cannot make a fist
- He finds it difficult to hold anything including a pen or a painting brush
- He cannot extend his arms or legs fully up to 180 degrees Presently he can extend only up to 150 degrees and it is worsening
- He cannot sit cross legged
- His posture while standing, walking or sitting is awkward because of joint stiffness
- Generally, the mental development is less than his age by about 5 years
- Physically his height is just 110 cm (about the same as a 5 year old normal child) and his weight is 28 kg.

A polyp has developed in his left nostril and is blocking it almost completely. He also has umbilical hernia. We are thinking of a surgery to address both these matters but are concerned about possible risks to his life due to problems that may arise while administering anaesthesia. In spite of his multiple problems, Parth is able to get ready himself, eats on his own and is generally able to look after himself. After wearing hearing aids he is able to communicate with others although his pronunciations are not clear. He has never thrown tantrums. He is mentally alert and his observation is very sharp. However, in the past couple of years we noticed a deterioration in his alertness - both mental as well as physical. He became slow in his actions, in grasping and occasionally said he had headaches or joint pains. He becomes breathless on a little walking.

But he never complains. He is always smiling. While he cannot do many of the activities, he encourages his younger brother to take part in sports or adventurous activities like rock climbing, and horse riding and is happy as if he is doing that himself.

In the end, let me share with you one incident. When my wife was pregnant with our second child, one day, Parth, who was less than 6 years old at that time, told her that this new baby should be "normal" and we will not make this child wear a pair of hearing aids! How touching!

Rajesh Motee



7th WORLD Symposium

16 – 18 February 2011, Nevada, USA

Basic Research; Translational Studies; Clinical Application

For nearly twenty years the UK MPS Society has had a presence at the American Society of Human Genetics (ASHG) Meeting that attracts over 10,000 health care professionals and always attracted significant interest from the Lysosomal Disease community. The WORLD Symposium came to life 7 years ago out of an initiative between the Lysosomal Disease Network in the United States and the National Institute of Health in Bethesda, Maryland. From modest beginnings this Lysosomal Disease Network Platform has grown into the leading meeting for lysosomal disorders not just in the United States but globally and well and truly taken over the voice of LSDs from ASHG.

This year's WORLD Meeting was a major educational and unifying activity with the first day focused on basic research, the second day on translational research and the third on clinical research. There was a significant UK presence and we were delighted that Dr Brian Bigger who leads the MPS Stem Cell Group at the University of Manchester present 'Lentiviral Mediated Stem Cell Gene Therapy Corrects a Mouse Model of Mucopolysaccharidosis Type IIIA'.

Apart from the Conference sessions I was involved in many breakfast, coffee, lunch and dinner meetings with other LSD patient groups as well as scientists and the pharma industry. This allowed time to be brought up to date on ongoing and prospective clinical trials. This is all very important information that is brought back to the UK for the benefit of the MPS Society members.
Christine Lavery c.lavery@mpssociety.co.uk

BRAINS FOR BRAIN 5th European Workshop

Frankfurt, Germany, 4 - 6 March 2011

The aim of this important scientific meeting was to:

- Discuss research achievements in the field of neurodegenerative disorders at a clinical, basic science and therapeutic levels;
- Explore how Brains for Brain (B4B) might collaborate with the European Union to stimulate interest in the research on Lysosomal Storage Diseases (LSDs) and the Blood Brain Barrier;
- Review collaborations with international patient associations and corporations to increase knowledge about LSDs and research projects

Over the two days of presentations we heard from most eminent scientists on their current research programmes but what was crucial to the learning was the in depth discussions and sharing of ideas by scientists from the different research groups following each presentation.

As well as Brian Bigger from the MPS Stem Cell Group speaking on 'The Role of Genistein in the Treatment of MPS III we heard Perry Calias from Shire HGT speak on 'Intrathecal (IT) Delivery of Recombinant Lysosomal Enzymes; and Rheinhard Gabathuler from biOasis Technologies, Vancouver, Canada talk on his work with p97, a vector for drug delivery into the brain. biOasis Technologies are able to show that therapeutic drugs 'piggybacked' as conjugates of p97 can be shuttled across the blood brain barrier (BBB) for treatment of brain diseases. Furthermore, they have been able to show that Aldurazyme (IDU) modified by incorporation of the p97 vector can be transported across the BBB in MPS I mice. **Christine Lavery** c.lavery@mpssociety.co.uk

Spanish MPS Society present to S.A.R. the Princess Letizia Ortiz

Asociación MPS España

www.mpsesp.org

On 31st January 2011,the Spanish MPS Society was delighted to attend a hearing by S.A.R. the Princess Letizia Ortiz at Zarzuela Palace. The representation was formed by five families affected by Hurler, Hunter, Sanfilippo, Morquio and Maroteaux-Lamy disease accompanied by the Dra. Mercè Pineda, the researcher Dra. Fàtima Bosch and Dr. Josep Torrent (EMEA representative), an important group of experts and collaborators involved with the Spanish MPS Society.

Each year we celebrate the National MPS Spanish Congress and I have always invited the Princess to assist. These invitations have been sent during the last six years and six congresses. Last year we received a personal letter from the Princess saying that we are invited to visit the Zarzuela Palace to show and explain to her the work, objectives and the affected patients of our Society. She wanted to know us!

During the hearing the Princess Letizia Ortiz proved to be very nice with the families, and especially attentive with the affected children. Besides talking with the families about living with the diseases on a daily basis she also revealed her interest to know more about the medical aspects and the development of the research of these diseases.

In this respect, thank you to the support and help of the scientific and medical experts who were able to explain the severe effects of the MPS diseases.

S.A.R the Princess Letizia showed immense sensitivity to the families and affected individuals and she offered us her support to continue with the work that we have been developing for years.

From all at the MPS Society we would like to thank her for receiving us at the Zarzuela Palace, for her kindness and attention to the Spanish MPS Society.

Thank you very much to the UK MPS Society for giving us the opportunity to write this article for the MPS Magazine.

Mercedes López
Secretary of
Spanish MPS and Fabry Society

Protecting patients

The European Union is to upgrade medicine safety monitoring to enable patients to be better informed on how to use medicines and enable patients themselves to report their adverse effects directly to national authorities. This has come about thanks to updates of EU law agreed with the Council and endorsed by the EU Parliament. The EU member states will be required to set up pharmacovigilance websites, and medicines that need special monitoring after being placed on the market will be marked with a black symbol.

To read the European Commission press release please go to www.europarl.europa.eu
Christine Lavery c.lavery@mpssociety.co.uk

Book Now!

MPS Society Conference Weekend

24 - 26 June 2011

Northampton Hilton

Places are going fast!

Earlybird deadline extended to 18 April 2011.

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. 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 Frtis Wrijberg 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
Belfast City Hospital: Dr Fiona Stewart

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
Hope 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 the:
London - Royal Free Hospital: Dr Derralynn Hughes
Hope 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.
Hope Hospital, Manchester: Dr Stephen Waldek

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

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

BIOMARIN®

A Phase 3, Randomized, Double-Blind, Placebo-Controlled, Multinational Clinical Study to Evaluate the Efficacy and Safety of 2.0 mg/kg/week and 2.0 mg/kg/every other week BMN 110 in Patients with Mucopolysaccharidosis IVA (Morquio A Syndrome)

Protocol Number: MOR-004
Investigational Project: BMN 110 (recombinant human N-acetylgalactosamine-6-sulfatase)

Study Objective: To evaluate the safety and efficacy of 2.0 mg/kg/week BMN 110 (GALNS) and 2.0 mg/kg/qow (every other week) BMN 110 compared with placebo to change from baseline endurance in patients with MPS IVA, as measured by an increase in the number of meters walked in the 6-minute walk (6MW) test from baseline to Week 24

Duration of Subject Participation: Up to 27 weeks

Selected Inclusion Criteria: Individuals eligible to participate in this study must meet all of the following criteria:

- At least 5 years of age
- Documented clinical diagnosis of MPS IVA based on clinical signs and symptoms of MPS IVA and documented reduced fibroblast or leukocyte GALNS enzyme activity or genetic testing confirming diagnosis of MPS IVA
- Must have an average screening 6MW test distance ≥ 30 and ≤ 325 meters

Individuals who meet any of the following exclusion criteria will not be eligible to participate in the study:

- Previous hematopoietic stem cell transplant (HSCT)
- Previous treatment with BMN 110
- Major surgery within 3 months prior to study entry or planned major surgery during the 24-week treatment period

For more information, please visit *www.morquioBMRN.com* & register for future program updates.

BioMarin Initiates Pivotal Phase 3 Trial for GALNS for the Treatment of MPS IVA

Novato, Calif., February 1, 2011 – BioMarin Pharmaceutical Inc. (Nasdaq: BMRN) announced today that it has initiated a pivotal Phase 3 trial for N-acetylgalactosamine 6-sulfatase (GALNS or BMN 110), intended for the treatment of the lysosomal storage disorder Mucopolysaccharidosis Type IVA (MPS IVA), Morquio A.

"In under two years, we have progressed the GALNS program from Clinical Trial Application to initiation of the Phase 3 trial. We have received FDA feedback and have finalised the design of the Phase 3 pivotal trial," said Jean-Jacques Bienaimé, Chief Executive Officer of BioMarin. "The study will be conducted at approximately 40 centres worldwide including Brazil, Japan, Taiwan, most Western European countries, Canada and the U.S. The trial is expected to enroll approximately 160 subjects and will be the largest enzyme replacement therapy trial conducted. There are no therapeutic options for MPS IVA patients who have a high unmet medical need. Initiation of this well-designed pivotal study is an important milestone for both the company and the MPS IVA community."

The Phase 3 trial is a randomized, double-blind, placebo-controlled study to evaluate the efficacy and safety of GALNS in patients with MPS IVA. The study will explore doses of two mg/kg/week and two mg/kg/every other week for a treatment period of 24 weeks. The primary endpoint is the six-minute walk test, and the secondary endpoints are the three-minute stair climb test and urine keratan sulfate concentration.

SANOFI-AVENTIS ACQUISTION NEWS

Earlier this week, we shared the news that Genzyme and sanofi-aventis have entered into an agreement under which sanofi-aventis will acquire Genzyme.

Sanofi-aventis is a global diversified healthcare company, headquartered in Paris, France and a leader in diabetes, oncology, innovative medicines, vaccines, consumer healthcare products and animal health. With this transaction, Genzyme will move into a new phase of our development, but continuing our patient-focused mission and developing treatments that change the lives of people with serious diseases.

Sanofi-aventis and Genzyme have a shared vision for our future together and believe we will emerge even better prepared to serve you. We will keep the Genzyme name, and become a division of sanofi-aventis when the transaction closes. Our mission and our deep commitment to you remains the same.

We expect to close on this transaction early in the second quarter of this year, and we have created a team involving members of each company dedicated to ensuring a seamless integration.

As we work through our integration, we will share details with you as we are able. Our future is dependent upon the support and involvement of our customers, and we are eager to hear your thoughts and feedback as well.

I thank you for your continued support of Genzyme and will update you soon on our progress and path forward. **Genzyme Therapeutics**

Stroke in Fabry disease:
Natural history data from the Fabry Registry

Emma James DPhil
Senior Project Manager
Global Registry programme, Genzyme

With an established family history, Fabry disease may be diagnosed at birth or even prenatally. However, for the majority of people, the symptoms emerge so slowly that a diagnosis is not made until later in life. About a quarter of people with Fabry disease are diagnosed after the age of 40 years, and unfortunately, some people only learn they have Fabry disease after they have suffered serious clinical events, such as a stroke or a heart attack [1]. Stroke is an important cause of disability and loss of life in young adults, and often the cause of the stroke is unknown. Young adults with Fabry disease, however, are known to experience strokes and other neurological complications [2]. For example, investigators in Germany screened 721 people (aged 18–55 years) who had suffered a stroke for Fabry disease. They found that 21 patients had a significant mutation within the α-galactosidase gene, meaning that 4.9% of men and 2.4% of women who had experienced a stroke, actually had Fabry disease [3]. Based on these results, it is possible that up to 1–2% of strokes are associated with Fabry disease [3].

The Fabry Registry was established in April 2001 to collect clinical and laboratory data on people with Fabry disease. All individuals with this disorder are eligible for enrollment, regardless of age, gender, symptoms, or whether they are receiving enzyme replacement therapy (ERT) [1]. This article describes a study by Professor Sims and her colleagues about the occurrence of stroke in people with Fabry disease. The study was published in early 2009, and was based on information from more than 2400 people enrolled in the Fabry Registry [4].

This analysis comprised data from only people who had not received enzyme replacement therapy:

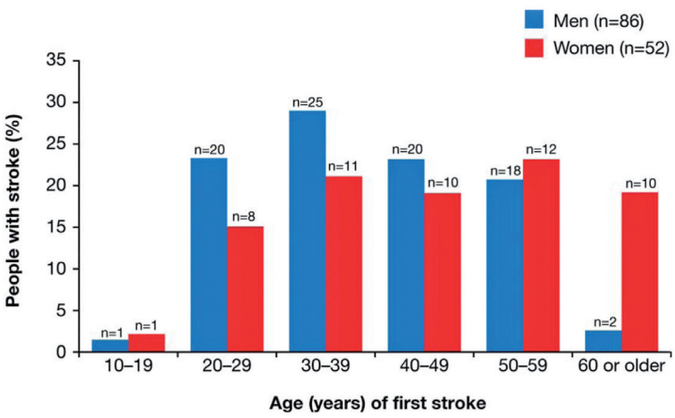
- 1243 men, of which 86 (7%) had suffered a stroke;
- 1203 women, of which 52 (4%) had suffered a stroke.

The results showed that men tended to experience their first stroke at a younger age than women, and the majority of people in the study had their first stroke between the ages of 20 and 50 years (Figure 1) [4]. Thirty people (22%) were aged less than 30 years when they experienced their first stroke, and two people had a stroke during their teenage years. In both men and women who had a stroke, their Fabry disease symptoms appeared later in life, compared with those with no stroke [4].

- In men who had a stroke, the first symptoms of Fabry disease occurred at a median age of 11.1 years, compared with 9.6 years in men who had not had a stroke;
- The median age of Fabry disease symptom onset among women who had a stroke was 24.0 years, compared with 14.6 years among women with no stroke.

Importantly, 46% of the people in this cohort had a stroke before they were diagnosed with Fabry disease, thus demonstrating the importance of early diagnosis to help prevent progression of the disorder [4].

Figure 1. The age of first stroke among men and women in the Fabry Registry [4].



The occurrence of stroke in people with Fabry disease was compared with the occurrence of stroke among the general population in the United States.

- Strokes were significantly more common in people with Fabry disease than in the US general population;
- People with Fabry disease experienced strokes at a much younger age than in the US general population.
  - o The average age at which men in the Fabry Registry had their first stroke was 40 years, compared with 76 years for men in the US general population;
  - o The average age at which women in the Fabry Registry had their first stroke was 46 years, compared with 81 years in women in the US general population.

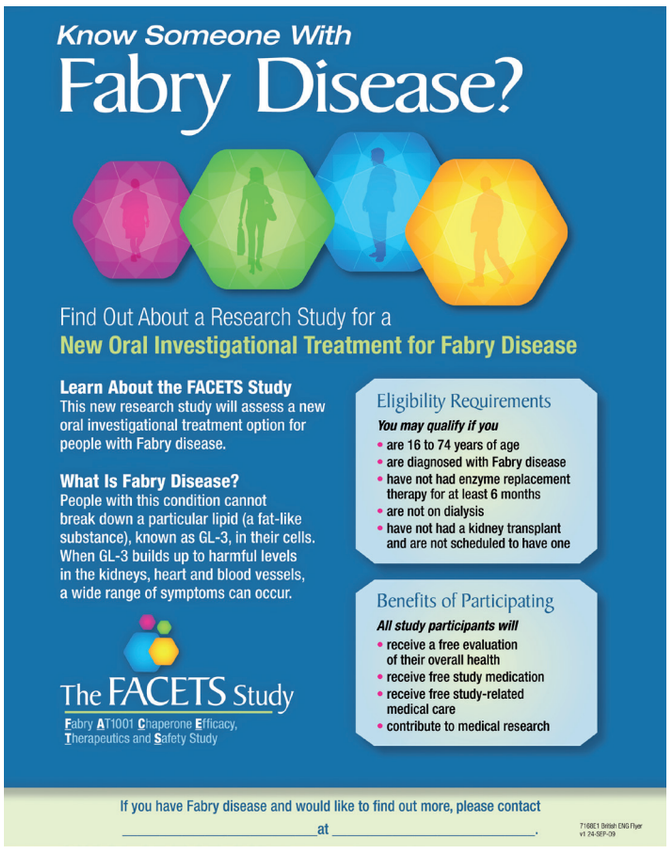
In addition to stroke, people in the Fabry Registry also frequently suffered from heart and kidney problems [4].

- Of the 138 people who had strokes, 67% of people had also experienced problems with their heart and kidneys;
- Men were much more likely than women to have problems with the heart and kidney as well as a stroke;
- Of the women who had a stroke, 50% had no other clinical events, compared with only 22% of men;
- Some other factors that were associated with stroke were history of a ‘mini stroke’ (transient ischaemic attack), abnormal heartbeat rhythms (arrhythmias), and high blood pressure (hypertension).

The findings from this analysis of Fabry Registry data by Sims and colleagues, offer new insights into Fabry disease [4].

- The finding that 4% of women in the Fabry Registry had had a stroke supports the hypothesis that women with Fabry disease are not, as previously thought, carriers of the faulty gene with a low likelihood of developing symptoms, but rather they are at risk of experiencing serious clinical events.
- The finding that nearly half of the patients in this cohort who had a stroke, had not at that point received a diagnosis of Fabry disease, contrasts with the 'classical' view of the disease, where symptoms appear in childhood, and progress to more serious clinical events, such as stroke, in middle age.
- People who had a stroke typically developed symptoms of Fabry disease later in life and most had not experienced major renal or cardiovascular events prior to their first stroke. Thus, it is imperative that individuals with Fabry disease are diagnosed as early as possible, particularly given the possibility of this relatively 'silent progression' to stroke.

Given the potential for progression to stroke, all patients with Fabry disease, regardless of age or gender, should be monitored for other risk factors for stroke. Further research is warranted before the effects of ERT on the occurrence of stroke in people with Fabry disease can be established, which will be supported by the ongoing participation of individuals enrolled in the Fabry Registry.



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Declaration of interest: The author is employed by Genzyme Corporation.

Protocol and Methodology of the stroke in young Fabry patients (sifap1) study: a prospective multicentre European study of 5,024 young stroke patients aged 18 years to 55 years. Cerebrovascular diseases (Basel, Switzerland). 2010 December 21;31(3):253-62

Background: Stroke in the young has not been thoroughly investigated with most previous studies based on a small number of patients from single centres. Furthermore, recent reports indicate that Fabry disease may be a significant cause for young stroke. The primary aim of the study was to determine the prevalence of Fabry disease in young stroke patients, while the secondary aim was to describe patterns of stroke in young patients.

Methods: The researchers initiated the Stroke in Young Fabry Patients (sifap1) study as a multinational prospective European study of stroke patients aged 18-55 years and collected a broad range of clinical, laboratory and radiological data using stringent standardised methods. All patients were tested for Fabry disease and blood was stored for future genetic testing.

Results: 5,024 eligible young stroke patients in 15 countries and 47 centres across Europe were enrolled between April 1997 and January 2010. The median number of patients included per centre was 98 with a range between 8 and 315. The average duration of patient recruitment per centre was 22 months, ranging between 5 and 33 months. The database was closed in July 2010. This paper describes protocol and methodology of the sifap1 study

Conclusion: The sifap1 study included the largest series of young stroke patients so far and will allow going forward for analyses on a large number of aspects of stroke in the young with Fabry disease.

Genetic Alliance UK
Supporting. Campaigning. Uniting.

Can you help the Genetic Alliance's campaign for equal access to PGD?

The Genetic Alliance are planning a Spring campaign to highlight inequity in access to preimplantation genetic diagnosis in the NHS. Could you help?

Preimplantation genetic diagnosis (PGD) is a method of artificial conception that allows a couple to ensure they do not pass a genetic condition on to their child. It is essentially in vitro fertilisation (IVF) where embryos are tested for a genetic condition before they are implanted in their mother.

Though we live in a time of exciting developments in the field of biomedical technology, the fact remains that the vast majority of life-limiting genetic conditions have no cure or treatment, and any treatments that do exist do not reverse the full effects of the condition they treat. The opportunity, therefore, for parents to avoid the birth of an affected child, without practicing ante-natal diagnosis and possible termination of pregnancy, is clearly valuable, and in most cases by far the most powerful tool we have to fight genetic disease.

The same technique as PGD can be used by parents to ensure their child is a tissue match for another child with a condition that would benefit from a bone marrow transplant, a "saviour sibling", this is called preimplantation tissue-typing (PTT).

At the moment very few genetic conditions can be cured (rather than treated), and probably (it is difficult to be categorical in this fast moving world) the only current cure is bone marrow transplant. For suitable conditions, a group of anaemias and blood disorders, PTT represents the best means by which a suitable match can be found for bone marrow transplant.

Taken together, PGD and PTT are extremely powerful tools that have the potential to rid families of the effects of genetic conditions.

Access to PGD is variable between and within the devolved nations of the UK. Northern Ireland has the best record for funding PGD; Wales is currently changing arrangements, but couples are subject to delays; and England and Scotland both have variations between different parts of the countries. In England variations are at the Primary Care Trust (PCT) level, so there could be up to 152 different policies on PCT funding within England.

The Genetic Alliance have had experience of PCTs deciding their funding criteria for PGD based upon their criteria for IVF, which is to ignore the many differences in situation between couples wishing to use IVF because of infertility, and couples wishing to use PGD to avoid the birth of a child affected by a genetic condition. These are entirely different situations, for which the intervention is similar, but not identical.

Similar issues are most likely occurring with PTT, the Genetic Alliance wish to investigate this further.

The aim of the Genetic Alliance's campaign is to present an account of the benefits of PGD and PTT to the Departments of Health in England, Scotland, Northern Ireland and Wales. To achieve this, the Genetic Alliance will gather information from a number of sources. They hope to have statistics of use, success, and access across the UK, and will make the many arguments in favour of PGD and PTT.

The Genetic Alliance believe that the voice of families who have benefitted from PGD or PTT, or are struggling to access PGD or PTT will be extremely valuable for this account. Real life stories of the effects that these funding decision have on families will be more powerful than statistics and arguments.

So please, if you have any experience of PGD or PTT or have had problems getting PGD or PTT funded on the NHS, then please get in touch with Nick Meade at the Genetic Alliance either by phone on 020 7704 3141, or by email at nick@geneticalliance.org.uk.

Please be assured that any conversations will be held in the strictest confidence and will only be used in this campaign if patients and families are happy to do so. Please do contact the Genetic Alliance by the end of April 2011.

More information on PGD and PTT can be found on the HFEA's website at: www.hfea.gov.uk.
Nick Meade Policy Analyst

Naidex National
- The First Naidex of 2011

Naidex National, (re-branded from Naidex Birmingham) will be taking place on 5-7 April 2011 at Birmingham's NEC. Building on its 36 years of success, Naidex National will be the largest Naidex ever, demonstrating once again how much it has to offer those wishing to achieve greater independence, their carers and healthcare professionals.

Over 390 exhibitors will be showcasing the best the industry has to offer those living with a disability, or working within the disability and healthcare sector. From wheelchairs to scooters, adaptive vehicles to pressure relieving furniture and shower commodes to stair lifts, Naidex National is the UK's largest one stop shop for all disability and healthcare products, services, features and advice.

A keen Naidex visitor, Mark Edworthy commented, "I am a passionate believer in Naidex, because as such a big exhibition it has all the products in one place which makes life easier. For example, when I was choosing a through floor lift, not an everyday purchase, I was able to see lifts from three different manufacturers all in the same day. If you are spending a lot of money, it is important to compare products. I have recently designed a new house and many of the disability features which helped make that house were found at previous Naidex exhibitions".

In addition to viewing and testing thousands of new products for independent living, a huge variety of features and seminar sessions will be on offer for all visitors. Healthcare professionals will have the chance to attend a comprehensive free CPD seminar programme (details will be available in the New Year), whilst members of the public can book one-to-one sessions with an expert OT, enabling them to discuss any issues they may have regarding caring and the help which is available.

Naidex National 2011's new and exciting features include the Wellbeing Walk which is designed to revitalise visitors whilst doubling as a hub of information and showcasing products benefiting everyday life. Free massages, alternative therapy and nutrition advice will be available with a team of experts on hand to answer questions.

For visitors looking for the latest new products to aid independent living there is the Innovation Zone. As part of the zone, there will be an interactive voting system where visitors will have the opportunity to vote for their favourite products in the New Product Award Showcase.

Within the Home Exercise and Rehab Zone experts will be offering practical demonstrations, help and advice in home rehabilitation. The Inspiration Theatre, previously known as the Lifestyle Theatre, is an interactive forum where visitors can listen to inspirational case studies. Organisations including Canine Partners will provide highly informative talks touching on topics such as the benefits and work of assistance dogs. The Special Yoga Centre seminar will examining body positioning and illustrate the beneficial results of yoga for many people.

Naidex favourites such as The Car Zone, Communications Village and KideQuip will once again be at the show offering everything you need to know about the latest advances in wheelchair accessible vehicles, communication aids and paediatric equipment and services.

Event Director Liz Virgo commented, "We are very excited about the new features at Naidex National. Combined with the new products and services on show we believe a trip to Naidex National will be extremely beneficial for members of the public, healthcare professionals and anyone who is caring for a friend or family member. Confirmation of the seminar programmes and a continually updated exhibitor list is available on the website".

www.naidex.co.uk

PATIENT FEEDBACK WANTED ON HOMECARE COMPANIES INVOLVED IN THE DELIVERY AND ADMINISTRATION OF ENZYME REPLACEMENT THERAPY

Currently there are two companies dominant in the delivery and administration of Enzyme Replacement Therapy for LSD patients receiving Homecare. In recent weeks the Society has received feedback from a number of patients or their caregivers / parents suggesting all may not be well with the service. So that the Society can assess the depth of the problems we would appreciate receiving feedback from all our members receiving ERT from a homecare company and /or having their enzyme delivered to the home. If you are content with the service please do tell us what is working well for you. If you encountering problems please do let us know so that we can address these with the Advisory Group for National Specialised Services (AGNSS), and the Homecare services themselves. Those of you who were with Careology will be aware that Careology has been acquired by Medco and once we have received your feedback we will request a meeting with this new company. The Society regularly meets with Healthcare at Home who are also keen to receive feedback. We assure you that you will not be identified by name, disease or geography in any matters we take up with the Medco or Health Care at Home.
Christine Lavery c.lavery@mpssociety.co.uk

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.

Careology

Announcement from Careology

Careology Ltd which provides specialist nursing support for people receiving medication at home for rare or complex conditions has just announced that it is combining with another specialist medicines homecare provider Medco Health Solutions [UK] and that this should mean even better services for members of the MPS Society in the future.

Careology, which prides itself on providing the highest quality of services to patients, has been providing enzyme replacement therapy support since 2005 for people affected by mucopolysaccharide disease and their team are very experienced in the needs of our members around the provision of their treatment. All Careology's highly specialised nursing team are joining the new company, so we should see no change to personnel on a day-to-day basis.

The skills of their new partner Medco Health Solutions [UK] should complement Careology's nursing expertise as they have extensive experience in handling and using specialist medications and its team provides expert advice to support people receiving these treatments.

Commenting on the news Mandy Wakefield of Careology Ltd told the MPS Society 'We are very excited about this new company. Client satisfaction with our services and helping people get the most benefit from their treatment is what drives both partners. The people we serve will continue to receive at least the same high quality service they've become accustomed to and our aim is to get even better.'

She added 'We know relationships between the Careology nursing team and the patients we serve are very important and we have no intention of disrupting these. Therefore I am delighted to be able to advise that current contacts at Careology Ltd will remain the same. People receiving our services will continue to be served by the same Careology Ltd team members.'

Careology and Medco Health Solutions [UK] will, of course, continue to work in partnership and under the instruction of the hospital treating MPS Society members, as they do currently.

DISABLED CHILDREN AT THE HEART OF NEWLIFE

Newlife Foundation for Disabled Children is a national charity, founded in 1991, that specialises in making life better and brighter for disabled children and their families Newlife achieves this through its services and funding of four key areas of action.

Nurse Services
The National Helpline is staffed by a qualified team of Nurses who listen, advise and offer help and support to all who call or contact them. The Nurses use their clinical skills and understanding to provide an invaluable service to families who have a disabled child. Whether you choose to contact them through a call or via the instant-messenger Live Nurse Chat service, Newlife Nurses are a confidential and trusted source of support and information to thousands of families every year. With 123 years of combined experience between them, Newlife Nurses offer immeasurable support and have recently expanded to respond to the growing number of calls to the service. All calls are free on 0800 902 0095.

Equipment Grant Services
Newlife Nurses are the access point for families applying for a grant for equipment relevant to their child's disability. This could be anything from wheelchairs to beds, hoists to seating systems and much more. One free phone call can result in completion of a simple application form for one of Newlife's Equipment Grants or signposting to another funder. Newlife Equipment Grants are non-means tested and often a decision on equipment can be made in days. A new equipment loan scheme 'Just Can't Wait' started in Jan 2011, and it is targeted to specifically help children with terminal/life-limiting conditions, where the need for equipment at home is urgent. Newlife has spent £5.6 million on Equipment Grants in the last five years.

Medical Research
Newlife believes that medical research holds the key to finding the causes of disabled and life-threatening conditions. By funding this research Newlife aims to improve understanding, diagnosis, treatment, prevention and improve children's health in the UK. Newlife has spent £11 million on research in the last 19 years.

Campaigns and Awareness
Many parents feel alone in the daily struggles and battles they face when looking after their disabled child so Newlife Foundation's campaign and awareness activities help to give them a voice on important issues.
All this help starts with a phone call. Newlife Nurse Helpline (0800 902 0095) is available Monday - Friday 9.30am-5pm and Wednesdays until 7pm.
Live Nurse Chat is available during office hours at www.newlifecharity.co.uk

Newlife ask for no subscriptions or memberships and all services are free. Newlife's Lead Nurse Karen Dobson says: "Our Nurses have great real-life experience and so this service does not operate like a Call Centre. We are real nurses helping real families, facing real difficulties, providing information so do give us a call and we will be pleased to help."

'Newlife Foundation for Disabled Children
- Registered Charity Number : 1001817'

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**

MPS Awareness Day

15 May 2011

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

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

Help us celebrate International MPS Awareness Day on Sunday 15 May 2011

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

Visit our website **www.mpssociety.co.uk** or give us a call on **0845 389 9901** to find out more.

How your money helps...

More professional support for more MPS Families

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

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

More MPS advocacy workers

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

More vital information

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

More help to cope with the isolation of a rare disease

The chances are you have never heard of 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**