

MPS magazine

Society for Mucopolysaccharide Diseases

Autumn 2017 • www.mpsociety.org.uk

YOUR BACK
TO SCHOOL
PHOTOS INSIDE

Walkabouts

GET OUTSIDE AND GET INVOLVED
WITH OUR AUTUMN CAMPAIGN

UNDERSTANDING TRANSITION

Feedback from the young person's workshop

NATIONAL DRAW

Our annual raffle has an artistic revamp

VIMIZIM QUALITY OF LIFE

Poster presentation with the first year of results

MPS and 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 neurological deterioration, and can result in death in childhood.

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

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 affected children and adults, their families, carers and professionals. We aim to:

- act as a support network for those affected by MPS and related diseases
- promote and support research into MPS and Related Diseases
- bring about more public awareness of MPS and related diseases.

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The articles in this magazine do not necessarily reflect the opinions of the MPS Society or its Board of Trustees.

The MPS Society reserves the right to edit content as necessary. Products advertised in this magazine are not necessarily endorsed by the Society.

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INFORMATION & RESOURCES

Our advocacy team have put together articles explaining carrier testing, mental capacity and transition



INTERNATIONAL
MPS Consensus
Programme meeting



NATIONAL DRAW
All about our donations
for this year's prizes

What's your story?

Are you a teenager or young adult with a lot to say? Have you had to overcome a challenge in your life or had a big achievement recently? Share your story with us and inspire others who are facing life's challenges



WELCOME

Welcome autumn! We are all preparing for chilly weather now and hope you enjoy our new photos for the walkabout campaign in this issue which will get you digging out the wellies and walking for MPS. See the back cover for more information.

It is also nearly time to book for the MPS I or MPS II expert meeting in Northampton next year, so we urge you to save the dates of 28–29 April 2018 and look out for the speakers, programmes and booking forms which will be available from the end of October.

With this issue of the magazine you'll find tickets for the National Draw – please sell as many as you can and let us know if you need more as we can send extras. Also, as we start the countdown to Christmas, we have included a Christmas card leaflet featuring the new designs ordered this year. Not only do we have a great selection but you will be helping the MPS Society to support individuals and families living with MPS and related diseases for another year.

From the Group Chief Executive



*Christine Lavery
Group Chief Executive*

In 2017 it is very encouraging that an increasing number of our children and adults diagnosed with MPS and Fabry have access to therapies. However we recognise there is still a long way to go to achieve reimbursed treatment across all the 25 diseases represented by the MPS Society. For some diseases we are still dependent on scientific developments, for others the outcome of clinical trials and of course when hope is looking you in the face frustratingly NHS England and the devolved nations payers.

Even in 1974 when our son, Simon, was diagnosed with Hunter disease Robin and I lived in hope that a new experimental treatment would save Simon from the inevitable. In 1981 we and a handful of other parents were given 'hope' when Dr Adinolfi at Guy's Hospital, London decided to transplant 6 young children with MPSI and MPSII with amniotic implants using the hypothesis that this embryonic membrane might produce and supply the missing enzyme. As parents we were keen to see the slightest signs of improvement in Simon and happily delivered weekly urine samples to the team at Guy's. In May 1982 Simon choked to death at school and gradually his 'amniotic implant' friends succumbed to MPS. For decades the use of amniotic membranes was put to one side. The silver lining to this is that in recent years use of the amniotic membrane has been re-introduced and expanded to different areas of medicine including treatment of burns, skin wounds, chronic leg ulcers and ophthalmology.

Jumping forward 35 years and the need for 'hope' following a devastating ultra-rare disease diagnosis has not diminished.

Since the early 2000's the MPS Society has encouraged and advocated for access to Food and Drug Administration (FDA)

“ the need for 'hope' following a devastating ultra-rare disease diagnosis has not diminished

and European Medicines Agency (EMA) regulated clinical trials. Over the last 15 years over 200 children and adults in the UK with MPSI, MPSII, MPSIIIA, MPSIIIB, MPSIVA, MPSVI, Fabry and Alpha Mannosidosis have been on a clinical trial or are on a clinical trial.

Perhaps the positivity of seeing so many of these clinical trials show an excellent safety profile, and prove efficacy through meeting their primary and secondary endpoints we have had a tendency to put to one side the reason for clinical trials.

In the past eighteen months regrettably we have seen two clinical trials for MPSIII terminated abruptly. The first to stop was the intrathecal enzyme replacement therapy clinical trial for MPSIIIA sponsored by Shire Pharmaceuticals and only in the last few weeks the intravenous enzyme replacement therapy clinical trial for MPSIIIB sponsored by Alexion.

Both of these clinical trials required the drug to be administered to the children every 2–4 weeks. All the children on the Shire clinical trial and many on the Alexion clinical trial have required a surgically implanted port. In both these two clinical trial terminations there was no time provision for the families to come to terms with the situation. A frequently asked question has been 'given the safety profile of both clinical trials was good why stop

WHAT'S ON?

Regional Clinics

Great Ormond Street Hospital

MPSIVA – 10 October 2017

MPSI – 12 September 2017, 28 November 2017

MPSIII – 26 September 2017, 12 December 2017

Birmingham Children's Hospital

MPSIVA – 15 September 2017, 19 January 2018, 17 August 2018

MPSIII – 20 July 2018 (am), 21 December 2018

Transition clinic – 29 September 2017 (pm)

Fabry – 20 October 2017 (pm), 16 February 2018 (am), 20 July 2018 (am), 21 September 2018 (am)

LAL - 16 February 2018 (am), 21 September 2018 (am)

MPSII – 17 November 2017, 16 March 2018, 19 October 2018

MPSVI – 29 September 2017 (am), 18 May 2018, 21 December 2018

Queen Elizabeth Hospital Birmingham

Adult Fabry – 12 September, 10 October, 14 November, 12 December

Manchester Children's Hospital

Post HSCT clinic (over 6 years) – 13 October 2017

Post HSCT clinic (under 6 years) – 6 October 2017

Regional events

Weekend for bereaved families • Warner Leisure Hotel • Thoresby Hall, Nottinghamshire 13–16 October 2017

Childhood Wood Planting

15 October 2017

Pantomime of Aladdin • Cophthorne Hotel, Manchester

26 November 2017

Conferences and expert meetings

MPS I Expert and Patient Meeting • Hilton, Northampton 28–29 April 2018

MPS II Expert and Patient Meeting • Hilton, Northampton 28–29 April 2018

15th Annual International Symposium on MPS and Related Diseases • San Diego, California 1–4 Aug 2018

immediately'? In both clinical trials there was no safety profile issues and therefore a managed approach to the termination of the clinical trial that was patient sensitive would have considerably reduced the immeasurable distress to the family and extraordinary time required by the principle investigator accounting for or even having to second guess to parents individually and collectively the reasoning behind in these cases Shire's and Alexion's thinking. So many times over the past months I have been asked forlornly 'Where is the evidence?'

Recently in the corridor of a busy Children's Hospital I had to remind a well-meaning individual from a pharma company that whilst the publishing of data from a failed clinical trial is important it is not going to happen overnight and back in

 we implore pharmaceutical companies to work with the patient organisations to integrate into their clinical trial planning a worst case scenario strategy that puts the patients and their families at the forefront in the regrettable event that a...clinical trial has to be terminated

the hospital café are parents mourning 'lost hope' for their children whilst having a brain full of questions that just won't go away because they don't believe what they have been told third or more hand.

In my 35 years' experience with the MPS Society and speaking personally as a mother of four children however unpalatable the truth ultimately it is the truth you want and you want it quickly. Going forward we implore pharmaceutical companies to work with the patient organisations to integrate into their clinical trial planning a worst case scenario strategy that puts the patients and their families at the forefront in the regrettable event that for safety, efficacy or business reasons a clinical trial has to be terminated.

Learning from these two experiences and the understandable position of parents to go to the end of the world to get 'treatment' via a clinical trial we would strongly suggest as parents you very carefully read the patient information sheet and consent form before entering your child onto a clinical trial. Do not hesitate to ask questions of the clinical trial principle investigator (PI). You need to become as knowledgeable as possible on the clinical trial especially where other clinical trials for the same condition are close to opening and not hesitating to ask to talk to the PI of any other forthcoming clinical trials and the MPS Society before reaching a decision.

Christine
c.lavery@mpssociety.org.uk

Announcements



We were sent this lovely photo of Ella and Jacob recently. Their two families met and became close during the MPSIIIB clinical trial, which has now ended. The photo was taken on holiday in Devon where they met up for a few days.

Judy Holroyd

Judy and her husband Chris joined the MPS Society in March 1986 following the diagnosis of their son, Will, with MPSIII. Will remained very strong and active well into his teens and one of my endearing memories of Judy is doing a run in the South West coaxing Will round in aid of the MPS Society.

Judy is qualified as a Genetic Counsellor and has been a highly valued member of the Board of Trustees since 2001. Judy has consistently used her expertise and MPS experience to progress the MPS Society and not more so than when it has come to evaluating and monitoring MPS research grants.

Very sadly Judy has needed to resign from the Board of Trustees for personal reasons. We will miss Judy's valuable contribution and the Board of Trustees and employees who know Judy are grateful for all she has done for the MPS Society over the years and wish her well for the future.

Christine Lavery



Judy Holroyd, MPS Society
Trustee for 16 years

NEW MEMBER

Christine Coghlan has recently been in contact with the Society. Her son Luke has a diagnosis of MPSIII disease. Luke is five years old. The family live in the North West of England.

Circus Starr

the circus with a purpose

FREE tickets to Circus Starr on 9 January at North Cambridge Academy and 10 January at Harrow High School are available now.

Email fundraising@mpssociety.org.uk
if you're interested. Find out more at
www.circus-starr.org.uk

Sweet sixteen

Rubina celebrated her 16th birthday this year and spent it with friends and family. Congratulations from the MPS Society.



Advocacy



Sophie

Manages the MPS Advocacy Team



Debbie

MPS IVA Morquio, MPS I Hurler HSCT, Hurler Scheie, Scheie, MPS VI Maroteaux Lamy, MSD, ML II (shared with Sally)



Rebecca

Fabry, MPS II Hunter, ML III/ML IV, alpha mannosidosis, beta mannosidosis, Fucosidosis



Louise

MPS III Sanfilippo (shared with Steve), LAL D, Gangliosidosis



Alison

Supports members living in Northern Ireland and the Republic of Ireland



Steve

MPS III Sanfilippo (shared with Louise), MLD, AGU, Winchester, Geleo Physic Dysplasia, MPS VII Sly, Sialic Acid Disease



Sally

MPS IVA Morquio, MPS I Hurler HSCT, Hurler Scheie, Scheie, MPS VI Maroteaux Lamy, MSD, ML II (shared with Debbie)

Update from the Advocacy Team

Wow! What a busy few months it has been since the last magazine. We hope you all had a fantastic summer and managed to get out and about, having fun and making great memories. We would love to hear what you got up to over the summer, any great holiday destinations or places you went to that would be worth sharing with other individuals and families.

We also wish all the children starting school for the first time this September. Please do share your photos and write any articles, we would love to see how everyone got on.

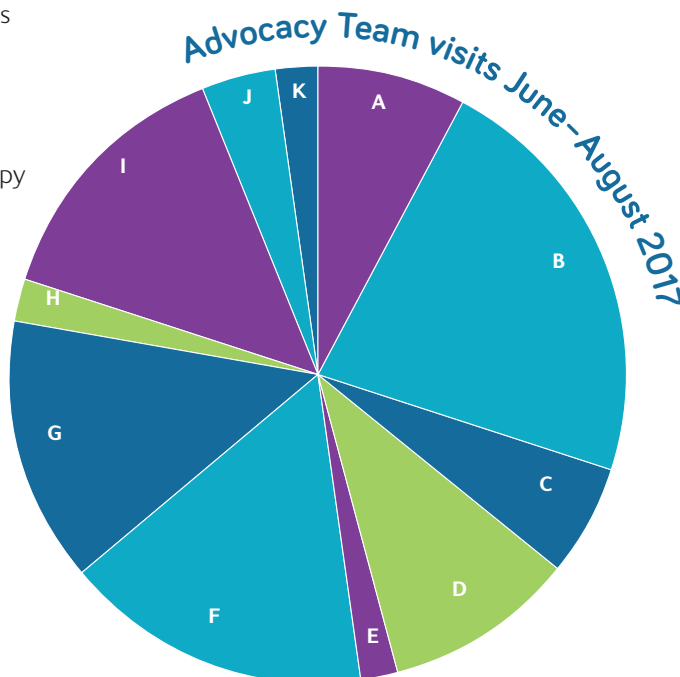
So...what have we been up too!

We continue to support a large number of you with various support needs and this is reflected in the various articles in the magazine from the team but we have scope for more. Please, if there is anything we can support you with, no matter how big or small, whether it is just a question or an idea please get in touch. We have been trying hard to reach out to all our membership so if you have a missed call, email or letter do get in touch and let us know how you are getting on.

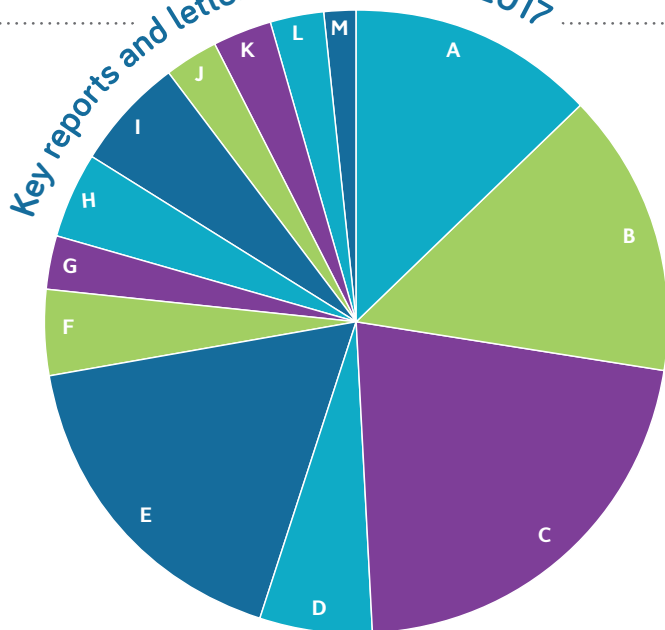
If you're not ready to access the support team but would like to support the Charity by fundraising, do get in touch with us. We can support and guide you through the process and are currently busy developing resources to help.

“ Thank you to all the advocacy team for everything that you all do, it makes a massive difference to have your support.

- A New home visits
- B General home visits
- C School/Nursery talks
- D Medical
- E Fundraising
- F Clinics
- G Education
- H Occupational Therapy
- I Social care
- J Benefits
- K LSD collaborative



Key reports and letters June–August 2017



- A Housing
- B Education
- C Benefits
- D Social Care
- E Complaints
- F Grants / Funds
- G CCG
- H General needs
- I Clinical management
- J Befriending
- K Mediation referrals
- L Mental Capacity
- M VAT exemption

“ I can’t thank you enough for your help. It was stressful but without you I couldn’t have done it.

News from the newbie

Sally, the latest addition to the advocacy team, gives an insight into her job role, meeting MPS families and all that she’s learned since joining the MPS Society

Joining the Advocacy Team was both really exciting and daunting all rolled into one! Exciting because this was a new fresh role with new learning experiences and lots of interaction with people, be it our members, other professionals and of course a new team. But daunting because there was so much to learn! With 25 diseases under the MPS umbrella where does a newbie even start?!

My concerns about the latter were soon reduced when my new Manager, Sophie, told me that I would be given a few of the diseases to learn about, and then begin supporting members with those conditions alongside my colleague Debbie. So that was it. MPS I, MPS IVA, MPS VI were to be my main areas that I needed to focus on. So 4 months in, that is where I am, still learning and hopefully making my own positive contribution to supporting our members.

Attending the various clinics around the country has been a great way to meet our families and the health care teams that look after them – and a good way to remind them that they are not alone, that there is help and support available should they want it.

The range of support needed is vast and varied, I wouldn’t even know where to start, so a few examples of current issues that I am helping members with are writing a report for inclusion in an application for funding for 1:1 support for a child in primary school, sourcing a care company to support a gentleman live independently in his own home. Attending a meeting at a member’s home to support an application for an extension/adaptations for their child. Not to mention the support requests for help with DLA and PIP applications that are coming through thick and fast! An area that is totally

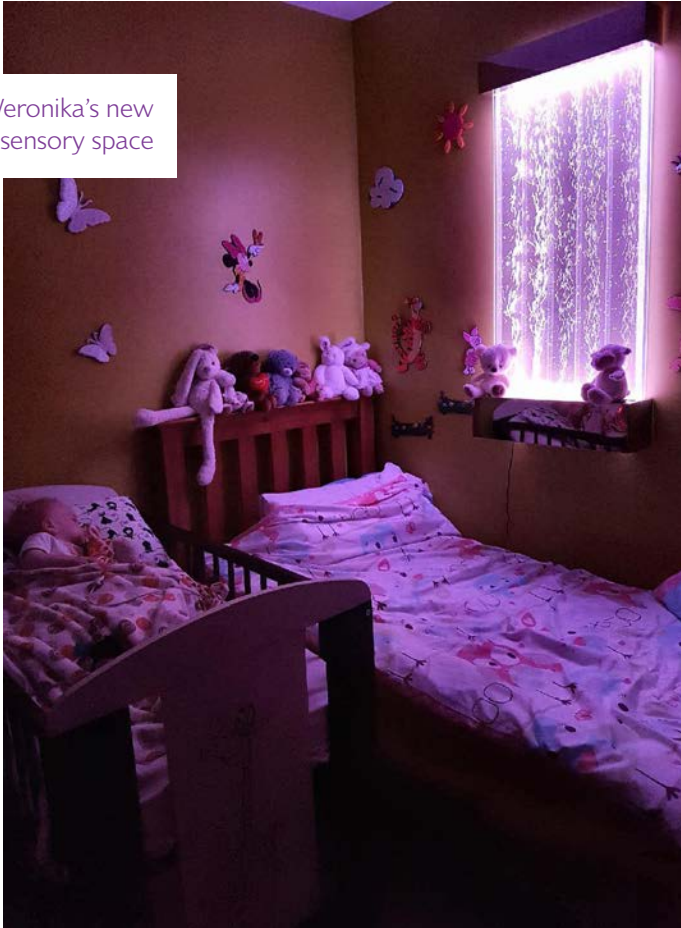
new to me but I suspect I will be able to call myself an expert by the end of the year!

I hope that the coming months will see me be able to build on my knowledge of MPS Disease, meet more of our families, and support all of those that would like it. Life is tough enough without dealing with these conditions and I have learnt enough to know that for our members, there are a multitude of obstacles to get over and around just for the most basic of needs. Often, needs that most people take for granted. So I am very glad to have joined my new colleagues in the Advocacy Support Team for the purpose of just that! Providing advocacy and support and I look forward to meeting and working with our members.

All Ireland Advocacy update

News from Alison, our very own all Ireland advocacy officer, on recent successes and the continued Vimizim campaign in Southern Ireland.

Weronika's new sensory space



The lovely Weronika (GM1 Gangliosidosis) is fascinated by lights and I was delighted to support the family to apply for funding to transform her room into a lovely sensory space. (see photograph).

It has also been encouraging to see families re-housed into accommodation that meets their access requirements following the provision of MPS Society Housing Reports. If you require support with housing please don't hesitate to contact your Advocacy Support Worker who will be able to assist you.

I have also been invited into lots of schools to support staff teams in meeting the needs of students affected by MPS. Having a member of the advocacy support team speak with nursery/school/respite staff is a great way to ensure that everyone supporting your child is well informed and has the opportunity to ask any questions they might have about MPS.

“So often families wait until things get overwhelming before they get in touch, when often a quick chat can help to put things right before life starts to feel out of control.

Where has the time gone! It seems like no time since I was last sitting at my desk preparing to update you on the Advocacy and Support work going on in Ireland. As always, the life of an Advocacy Support Officer is never dull and it is always a privilege to know that families feel they rely on us when times are tough. If you have a support need that you've been keeping to yourself, or that you feel is too small to mention, please do get in touch. So often families wait until things get overwhelming before they get in touch, when often a quick chat can help to put things right before life starts to feel out of control.

In the last few months we've had lots of little (and not so little) successes in our work with families in Ireland.

Some of our Northern Irish families have been supported to access charity funding to enhance the lives of their children.

Southern Ireland

In the South of Ireland things have been very active for the past few months.

We have been supporting the Irish MPS Society in their campaign to secure access to Vimizim for MPS IVA. You will all remember the uphill struggle we faced in the UK before Vimizim became available, so you will know just how difficult it is when you are a family in the midst of a battle for access to treatment.

Ireland is a small country with only a handful of MPS IVA families. Please support us in helping the MPS IVA voice in Ireland to travel further by supporting their social media campaign. Sometimes when you are few in number you need an army of friends behind you to make your voice louder and stronger!

Press release from the Irish MPS Society in response to the HSE decision to refuse funding for Vimizim

Statement Date: August 2017

The HSE have confirmed that they WILL NOT FUND the life changing drug VIMIZIM for those suffering with the ultra-rare disease, MORQUIO. This announcement comes 3 years after Vimizim gained licensing approval from the European Medicines Agency (EMA) on 28th April 2014.

The Irish Society for Mucopolysaccharide Diseases is very disappointed with this decision on behalf of all patients with Morquio in Ireland who have waited so long for a treatment to be developed and are now being denied access to it.

The HSE has stated that it has “decided to refuse to reimburse Vimizim in Ireland due to lack of clinical data”; in spite of the fact that it is currently funded in more than 10 European countries, including Northern Ireland. We find it very surprising that the HSE has taken such a stance based on data that has been well documented and understood in other countries and has not been the cause for refusal elsewhere.

It is proven that Vimizim (Elosulfase alfa) brings multiple benefits: avoiding/delaying the requirement for invasive ventilation; improving cardiac function and muscle strength; improving respiratory function, resulting in improved sleep which reduces sleep apnoea; lessening incidence of chest infections and breathing difficulties; and increasing energy and stamina. Vimizim also shows improved growth and promotes stronger posture, which benefits mobility and organ function. Vision is preserved by preventing corneal clouding. Pain levels become more manageable.

Collectively, these changes enable those with Morquio to complete normal day-to-day activities independently and offer the hope of very significant long-term benefits through extended use. One of these very significant benefits is to prolong life expectancy.

Vimizim has been provided compassionately by BioMarin for the past three years to those who participated in the clinical trials. BioMarin has agreed to continue to provide Vimizim compassionately until 5th December 2017. After this date, vulnerable children with this degenerative condition, Morquio, will no longer have access to this life changing drug.

Vimizim is manufactured by BioMarin in the Republic of Ireland. BioMarin is considered a major employer. This drug is now being reimbursed in more than 10 European countries, including Northern Ireland. Thus it is difficult to accept that some patients living on the Island of Ireland have access to this vital drug while others will not.

We are hugely grateful to BioMarin for their solid and continued support in providing free access to this drug to those who were involved in the clinical trials and working hard to enable access to Vimizim for everyone with Morquio. It is also important to note that there is already one child receiving treatment funded by the HSE. This is decidedly unequitable and discriminatory.

We are hopeful that the HSE will see sense, consider the clear positive benefits and make the right decision to ensure continuity of care for everyone and we will work hard for a positive decision.

About the Irish MPS Society

The Irish Society for Mucopolysaccharide Diseases (The Irish MPS Society) is the only registered ROI charity providing support to families affected by MPS and related disease. The Irish MPS Society provides an advocacy service to individual families as well as supporting funding towards innovative and life changing clinical and academic research.

As a registered charity, the Society is entirely supported by voluntary donations and fundraising

Email: irishmpssociety@gmail.com

A week in the life of an advocacy support officer

No week is ever the same in the life of an MPS Advocacy Support Officer. Whilst there are some tasks we do regularly such as giving a talk about specific diseases or attending children's clinics at specialist centres, we often find ourselves doing a variety of activities to support our members.

For example recently I visited a family in the South of the country and then a few days later visited families in the North West of the country. The needs of every individual family can vary greatly, from needing support with housing, to finding respite, charitable funds and befriending links, suitable toys and equipment. Some families need advice about how to complete Disability Living Allowance forms, or specific advice on how to manage education, housing, social care or continuing healthcare meetings.

As a small service that covers the whole of the UK, we often try to see as many families as possible in one area as we understand that for many families meeting us face to face and having a listening ear is all part of being supported. Whilst it is not always possible to see everyone in one area at a time, we always try to make ourselves available for those that need our support to attend specific meetings.

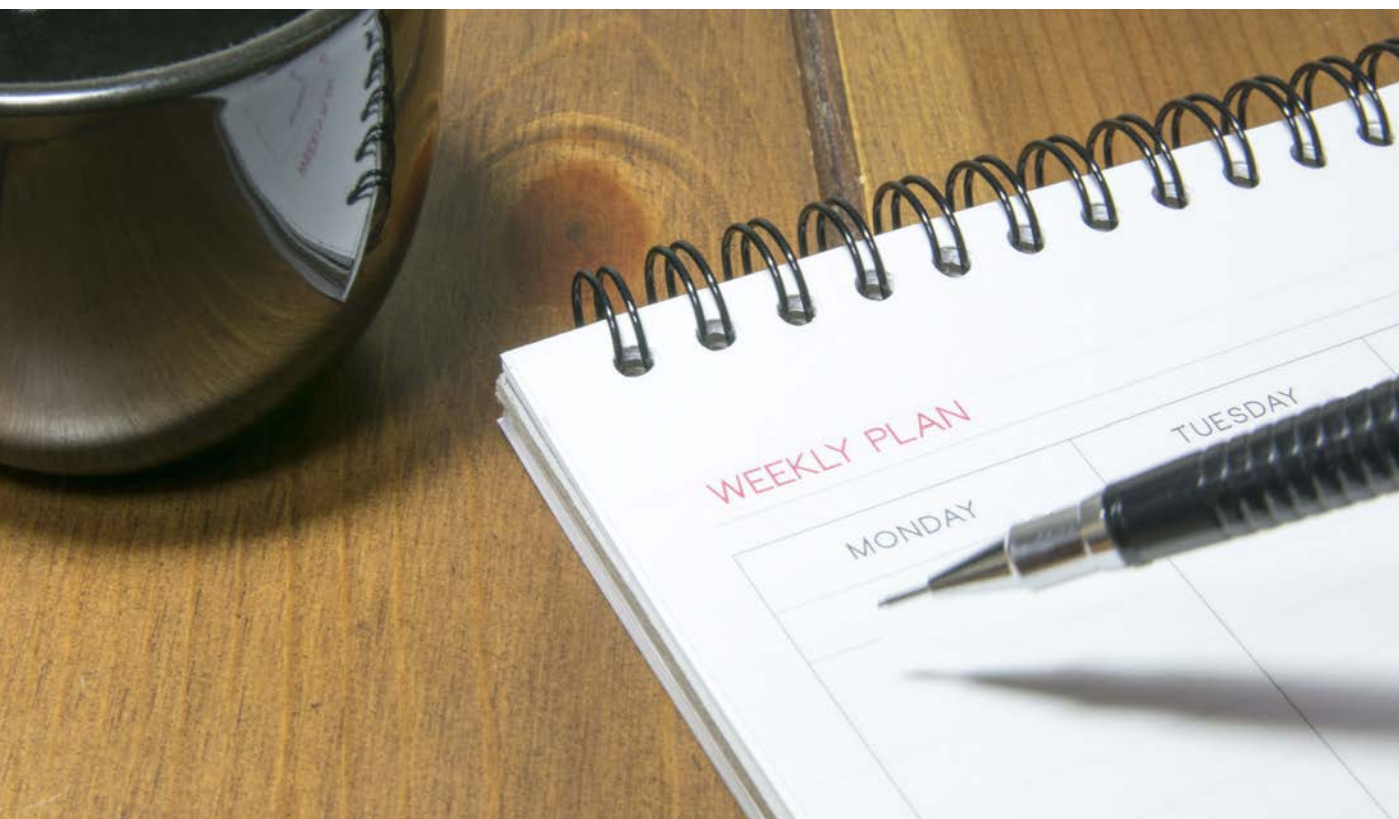
Then there are times were we are in the office, we support families from a distance. We take many calls and emails from members whose needs vary greatly from needing active support, specific advice or a listening ear. We get a range of calls from those who need advice prior to diagnosis all the way through to those who may be in the final stages of their disease. From time to time we also make contact with some of the members we have worked with, as we know it can be hard for some families to ask for help.

The weeks where we are undertaking a number of visits, time in the office can be precious. It provides us with a chance to respond to written communications, carry out research on local policies and legislation and gather information from families that may help us to best support them. Every piece of support work we do is personalised to the individual and their family and therefore varies in the time it takes for us to complete.

All of this is in addition to carrying out tasks such as basic administration, gathering further information to support fundraising and the development of the MPS magazine, and at times seeing if members are interested in participating in research.

*Louise Cleary
Advocacy Support Officer.*

“ We take many calls and emails from members whose needs vary greatly from needing active support, specific advice or a listening ear.



Remembrance

In memory of
our wonderful grandson
Luke John Bown
16 August 2004–26 April 2017



You were very special
And that's why we mourn
A wonderful grandson
Who from the day that you were born
Were such a shining light
A gift so precious too
Amazing in many ways
You brightened our world it's true
And gave us so many wonderful
memories
Which we will always cherish
You would have grown, become a
man
And it seems it is unfair
You had your life to live
And we were blessed to know you
And now that you have gone
We hold onto your memory
And our love for you goes on...

We will love you forever Luke
Lots of love
Nan and Grandpop



Donations have been made in memory of Bob Silcock, grandfather to Bobby Gill (MPSIIIA). Bob passed away on the 4th June 2017 after a short battle with Motor Neurone Disease. Bob was a wonderful painter, with his work exhibited at various galleries including the Saatchi gallery. Every painting Bob sold he donated all the money to the MPS Society which is why the family asked for donations to the Society at his funeral raising a total of £3040 so far. Bob is greatly missed and forever loved by his family and friends.



Our sincerest thanks go to friends and family of Lisa Nurse who have donated €1028.75 in her memory. Lisa, who passed away on 28th April 2017, was and always will be deeply loved by those that knew her.

Bereavements

We also wish to extend our deepest sympathies to the family and friends of Kathleen Ghai who had Fabry and passed away on 29 July 2017.



Senior Advocacy Support Officer, Debbie, gives advice on supporting your child at school and how the MPS Society can help you

It's that time of year again when the summer holidays are over and thoughts turn to the new school year. For some of our members this may mean a new start at nursery or school.

How can the MPS Society support our younger members at school?

- The advocacy support team can provide information, booklets and advice to schools and nurseries. The team can give verbal support by telephone or visit schools and give a staff training session or information to students.
- The advocacy support team can assist schools in preparing care plans and risk assessments and can support schools in making plans for school trips.
- The advocacy support team can also guide a family through the Education, Health and Care Plan process.

How can parents support their children at school?

- Communication between home and school is key. It may be a good idea to have a communication book to jot down upcoming medical appointments or how your child is feeling. For example, letting the school know if your child had a bad night's sleep.
- Give the school lots of information about your child's needs and MPS disease before they start at a new school so that the school can plan and prepare a good start.
- Inform the school of any equipment the child may need at school such as wheelchair or specialist seating. Schools often have links with occupational therapists who can inform and supply equipment for school.

- Keep in touch with the school calendar. It helps to know when school trips and events are coming up so that you can plan ahead.
- Suggest regular meetings with your child's teacher so that you can monitor progress and any difficulties can be dealt with early on. Does your child have behavioural needs? Are they too tired after school to manage homework? Discuss these with the teaching staff so they are aware of your child's needs.
- Discuss routines and boundaries with teaching staff so that behavioural strategies can be shared to provide consistency and support children with behavioural needs.
- Invite the MPS Society advocacy support team to offer support and give information to the school.
- Encourage your child to have friends visit for play dates.
- Talk to your child about their school day and celebrate their achievements!

Back to

Our autumn issue would not be complete without a back to school special feature and your lovely new term photos. Thanks for sharing!



Sam and Alex



Lily



Maddison

school



Emily

Clinics



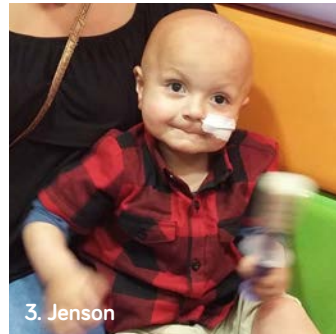
1. Gurkiran and little brother

BIRMINGHAM CHILDREN'S HOSPITAL BMT CLINIC 16 June 2017

(1-5)



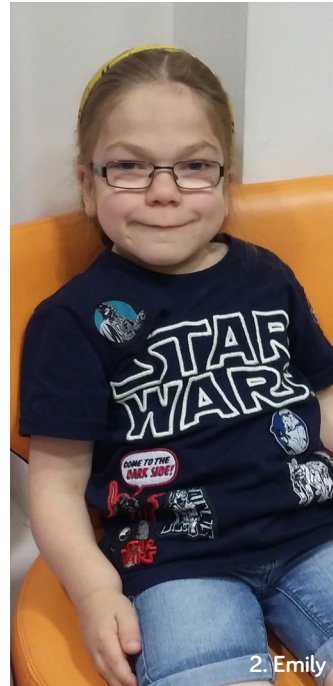
4. John



3. Jenson



5. Meleeha



2. Emily

As the newest member of the Advocacy Support Team, I attended my first ever clinic and introduced myself to several families there. It was great to meet them and see how they were getting on, if they needed any support with anything and just generally saying hello! I got some big smiles when taking pictures which was lovely.

Sally

GREAT ORMOND STREET HOSPITAL MPS IVA MORQUIO CLINIC 25 July 2017

(6)

A visit to Great Ormond Street Hospital to attend the MPS IVA clinic was very noisy! The doctors and nurses were seeing lots of children with all sorts of conditions, so it was extremely busy. However, the children with MPS seemed to congregate together in one play area of the clinic with their parents so that made it slightly easier to meet them all and see how everyone was doing. All of the children were at the start of the 6 week school holidays, and they all seemed to be happy about that! It was great to meet (if only briefly because she was running around like a headless chicken) Michelle Wood the resident Specialist Physiotherapist who looks after our MPS children. In fact I have enjoyed meeting many of the dedicated nurses and specialist staff over the course of the three clinics and it's been a good way to meet so many of our children and families. I look forward to attending more clinics and hopefully offering support and advice to those that would like it.

Sally



6. Hayyan



7. Demi Leigh



8. Harvey and Elmo

ROYAL MANCHESTER CHILDREN'S HOSPITAL HSCT CLINIC

14 July 2017

(7-13)



9. Rubina



10 Morgan



11. Jessica

I attended a very busy clinic at the Royal Manchester Children's Hospital. Most of the children were running lots of tests and some had travelled a long way with their families to attend clinic. Despite this, they were all cheerful, talkative and very importantly...

co-operative! It was great to meet the dedicated staff that are so expert in understanding their health issues, but also know the children so well. It was as much of a catch up for everyone as well as being an important health appointment.

Sally



12. Mikko



13. Mason

BIRMINGHAM CHILDREN'S HOSPITAL MIXED CLINIC

18 August 2017

(14-19)



14. Jacob with brothers, Samuel and Max, and dad, Chris



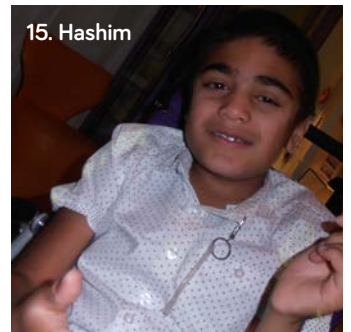
18. Dylan



17. Jake



16. Beau



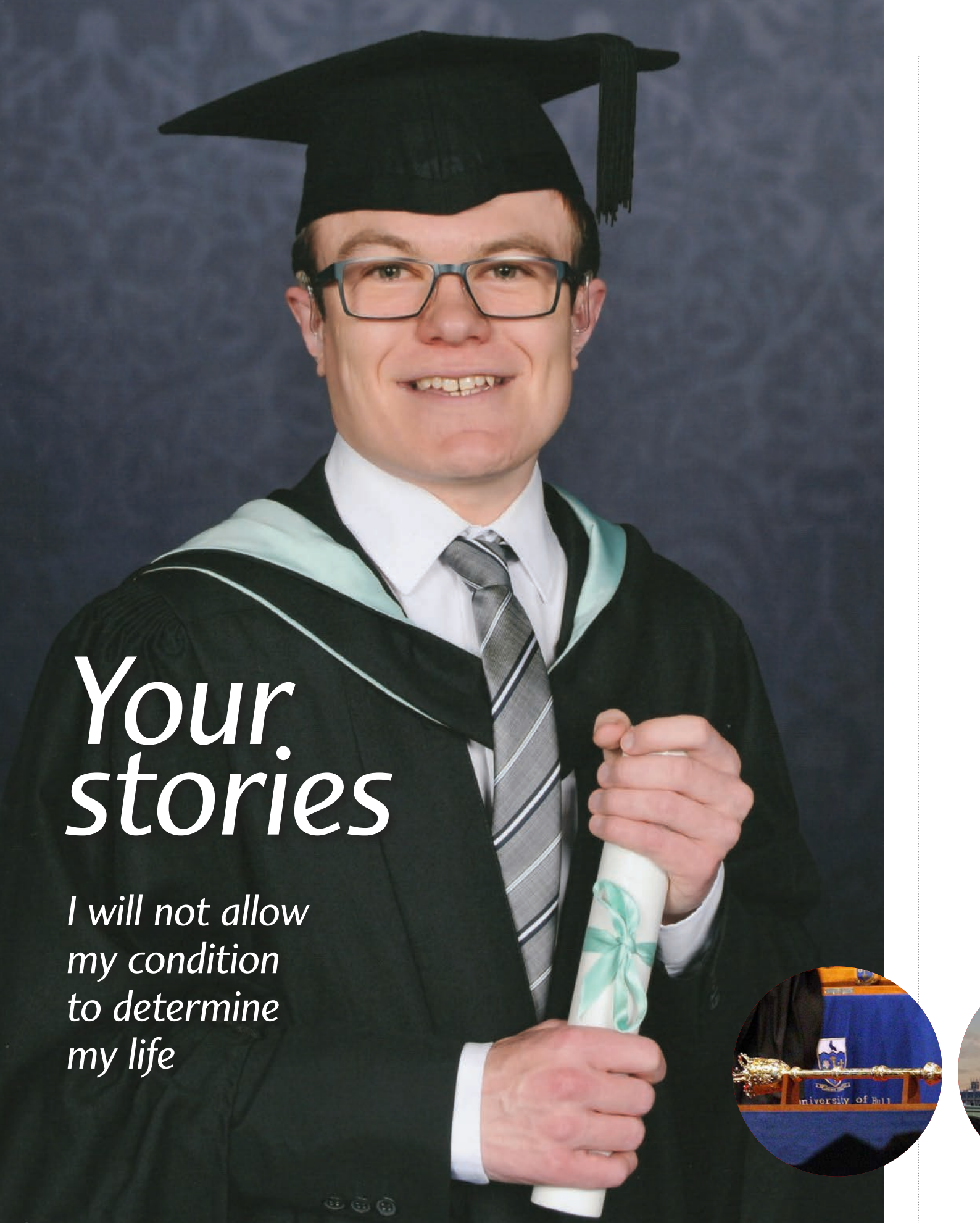
15. Hashim

Hello! I travelled to Birmingham Children's Hospital recently and attended one of their clinics, where roughly seven families attended. As usual it was lovely to see the children and families and meet those that I have only had phone or email contact with. I always find the face to face contact improves my understanding of how families can be supported. As you will see I managed to get some lovely photos. I hope you enjoy them, bye for now.

Louise



19. Anya and sister, Aliza



Your stories

*I will not allow
my condition
to determine
my life*



Ben Cooper has not let his diagnosis of MPS II Hunter hold him back or affect his life decisions. He explains why it's important to him to keep his condition separate from the rest of his life.

"I did it", I thought immediately coming off the stage at my Graduation. I hadn't tripped over my gown and embarrassed myself in front of hundreds of people. That sense of relief was greater than the happiness I felt after discovering I was going to graduate with a First from the University of Hull.

I was probably a little too ambitious the day I applied to study British Politics and Legislative Studies (BPLS) at Hull University. A core part of my degree was an 11 month internship working for a Member of Parliament in London. I didn't really think about how my condition would impact that work or any work at university. My choice of Hull allowed me to live at home and avoid being independent, while also setting this huge challenge of living and working in London in just two years. I didn't really think about that. But applying for something and thinking about the consequences only when it's too late to alter course is my way of doing things.

I have constantly striven to keep my condition separate and distinct from the rest of my life. Increasingly that barrier between medical life and the rest of my life is crumbling, but it is vital to attempt to keep them apart for as long as possible. I will not allow my condition to determine my life. It is a constant fight, but a necessary one, to not allow anyone else to reveal the details about my condition to others.

Over 11 months in London I found that an increasing challenge. Treatments were difficult, hospital appointments exasperating, and educating a new group of people about my condition at

times infuriating. But working for a Member of Parliament in the majestic Palace of Westminster was a privilege few get to experience. And for all its negative reputation, living in London attracts millions for a reason. Absolutely more needs to be done to tackle the discrimination faced by disabled people, especially those with invisible illnesses, but London remains one of the most accessible cities in Britain. I loved every moment of it.

When my parents were first told about my condition, they believed it to be all bad things. It's hard not to adopt that mentality when faced with the basic facts of this condition. It is also easy to let the condition define you and your family, and I could not let that happen. Making jokes, telling funny stories and cherishing unique moments are my means of striking blows against my condition.

I will continue to make my life decisions without concern for my condition, much to the consternation of parents. And I'll continue to laugh at it. My condition won't extinguish my ambition.

The next challenge? The University of Cambridge and a Masters. Let's see what they make of me.



Understanding transition: the views of young people aged 13–18 years

Transition is a natural part of growing up. It is the opportunity where possible, for young people to take responsibility for their own lives including choices related to healthcare, social care, further education and employment. With this in mind, the Society has been working with members aged 13–18 years to better understand their knowledge of transition, expectations, helpful resources, who should talk to them about transition and any concerns or worries they have as they approach this area in their lives.

Here is a summary of the questions asked and the feedback so far.

What do you know and understand about transition?

- When you change from being a child to teenager and then a teenager to an adult.
- When you start making more arrangements for yourself.
- Where you move from one place to another for example from a children's to adult's hospital.
- A change that happens, like moving schools.
- My healthcare will change at 18yrs.

- You move to an adults hospital at 18yrs
- Your healthcare at home changes at 18yrs
- You move to college at 16yrs
- You move to university from the age of 18yrs
- Your entitlement to benefits changes at 16yrs
- You can enter employment from the age of 16yrs

What services do we transition from and at what age?

Save the date!

MPSI and MPSII Expert Meetings

A warm welcome awaits all those affected by MPSI and MPSII, their families and healthcare professionals attending the forthcoming MPSI and MPSII expert meetings on 28–29 April 2018 at the Hilton Hotel, Northampton.

There will be a separate Expert Meeting for each in respect of MPSI and MPSII with all delegates and speakers coming together for dinner on Saturday evening. In true MPS Society tradition there will be a dedicated child, vulnerable adult and sibling programme on both days.

The final Expert Meeting programmes for MPSI and MPSII with booking form will be available at the end of October 2017. The speaker programmes will include UK and international delegates and as a taster we are delighted that the following speakers have confirmed:

MPSI Expert Meeting

[Andrea Jester \(Birmingham\)](#)

Speaking about improving poor hand function

[Elaine Robinson \(Belfast\)](#)

On the surgical interventions for managing the MPSI hips and legs

MPS II Expert Meeting

[Alex Broomfield \(Manchester\)](#)

Speaking on the clinical aspects of the attenuated MPSII

[Lynda Polgreen \(USA\)](#)

The effect of arthritis treatment on physical function

You can expect a new addition to the Expert Meeting programme by way of a Dinner and Learn session on the Saturday evening.

Information & resources

Specific questions young people had about transition

- What happens when we move services?
- How quickly will the move happen?
- Who can I ask for support?
- What changes can I expect?
- Is there someone that can give me tips on how to become employed?
- Is there someone that can explain what services are there to support me?
- Is there someone to guide me through the process?
- What information do we need to know?

- Will I know anyone?
- Can we talk to someone that has experienced transition?
- Will I know where to go?
- Any tips for organising my own appointments?

Specific worries that were raised

What resources and communication aids did young people want?

All young people were wanting resources and communication to be via online resources suggestions included

- You tube videos showing medical procedures or young people sharing their experiences
- Apps that give information and advice on things such as benefits, opportunities in further education, driving, employment, how to complete forms or make job applications
- Opportunities to ask questions to medical professionals about their condition and surgeries
- Online opportunities to speak with other young people

Free MOOC about research opens

A Massive Open Online Course (MOOC) developed by the National Institute for Health Research (NIHR) Clinical Research Network (CRN), University of Leeds, and FutureLearn is due to run again in November and is open for registration.

The MOOC is a four week online course and is free of charge. The course works alongside online materials and an online community of learners, and aims to answer some pertinent questions, including: 'How are treatments and cures discovered?' 'What is the impact of clinical research?' 'How do we undertake clinical research?' 'Why do we do clinical research?'

The fifth MOOC is entitled 'Improving Healthcare through Clinical Research' and will run from 6 November 2017.

Over four weeks the course includes:

- Learning about the roles of different members of a research team
- Hearing from people who have taken part in clinical research studies
- The ethical and scientific principles that underpin the research process
- Getting a better understanding of the challenges of conducting clinical research and the benefits to modern healthcare

You can sign up now to register at:

www.futurelearn.com/courses/clinical-research

We would love to hear from more of you. Do you share these views? Is there anything not covered? Is there any information not identified that you feel would be helpful?

Also it would be great to hear from parents in respect of what your expectations are, what your fears and hopes are for the future and how you feel the Society and other professionals could support you and your child through this process.

Please feel free to contact [Sophie Thomas](mailto:s.thomas@mpssociety.org.uk), Advocacy Support Team Manager, on 0345 389 9901 or email s.thomas@mpssociety.org.uk

The Mental Capacity Act 2005

In recent times the advocacy team have taken a number of enquiries regarding The Mental Capacity Act and the implications in terms of decision making, the caring role and supporting vulnerable adults. We know and understand that this is a complicated and daunting area of law and as such we thought it would be helpful to publish a series of brief guides to the legislation within this and future editions of the magazine. To begin **Steve Cotterell** gives a brief over-view of the Mental Capacity Act 2005.

The Mental Capacity Act 2005 is relevant to England and Wales and applies to everyone involved in the care, treatment and support of people aged 16 years and over who are unable to make decisions. In Northern Ireland there is the Mental Capacity Act (N.I.) 2016 and in Scotland the Adult with Incapacity Act 2000, each of which follow similar themes and principles.

The Mental Capacity Act Code of Practice is a useful document that relates the provisions within The Mental Capacity Act (2005) to practice and defines the key principles within it.

There are two questions to answer when deciding capacity:

- Is there an impairment or disturbance in the functioning of a person's mind? If so,
- Is the impairment sufficient that the person lacks capacity?

Another question to ask is "can the decision wait?" Capacity is required to be considered for each decision to be made. Loss of capacity may be

considered temporary for example due to seizure activity or substance misuse, and so if a person would have the capacity otherwise and the decision is not considered urgent then it is possible for a decision to be delayed.

The MCA says that a person lacks capacity if they cannot do one or more of the following:

- **Understand** information
- **Retain** information to inform their decision
- **Weigh up** the information given
- **Communicate** their decision

People can lose capacity to make decisions due to a number of reasons for example:

- A stroke or brain injury
- A mental health problem
- Dementia
- Learning disability
- Substance misuse
- Illness or side effects of treatment for illness

The Act sets out five key principles:

1. Every adult has the right to make his or her own decisions and must be **assumed to have capacity** to make them unless it is proved otherwise.
2. A person must be given **all practicable help** before anyone treats them as not being able to make their own decisions.
3. Just because an individual makes what might be seen as **an unwise decision**, they should not be treated as lacking capacity to make that decision.
4. Anything done or any decision made on behalf of a person who lacks capacity must be in their **best interests**.
5. Anything done for or on behalf of a person who lacks capacity should be the **least restrictive** of their basic rights and freedoms.



is conflict amongst decision makers; when no agreement can be reached via the “best interest approach”.

Deprivation of Liberty Safeguards (Dols)

This process is in place to protect vulnerable people from inappropriate restraint and restrictions. Permission is required from the Court of Protection to deprive a person of their liberty; examples of types of care that may be deemed restrictive and requiring this permission include; constant supervision and where a person is not able to leave a premises unaccompanied, high sided bedrails, stairgates and any physical restraints. The Court will only approve a Dols application where it is evidenced that such care is in a person’s ‘best interests’. Dols procedures apply to care received within a hospital or care setting including care in the community and within a person’s own home.

Independent Mental Capacity Advocates (IMCA)

Independent Mental Capacity Advocates are specially trained and appointed advocates, independent of the local authority and the health authority, and who can support individuals to challenge applications for deprivation of liberty. They can also be used to support individuals who lack capacity through a best interest’s process and to contribute to the decision making process for vulnerable people. Their role is to ensure that the principles of the Mental Capacity Act are followed.

Each of the provisions will be explained in further detail in future editions. Should you have any questions or if you need to talk about any aspect of this article please contact the advocacy service and we will be happy to help.

Provisions within the Act

Best interests

Act states that where a person is deemed to lack capacity then any actions or decisions taken must be considered in the person’s best interests. The Code of Practice provides a checklist that decision makers must work through in deciding what is in a person’s best interests. This must be a collaborative approach with those people involved in caring for the person concerned.

Court of protection

This court is designed to protect people who lack capacity and to advise those who make decisions. The court, upon receipt of an application for a decision or a request to resolve a dispute, has powers to appoint a deputy to make decisions for someone who lacks capacity, but also has the power to make decisions where there

Useful contacts and information

Office of the Public Guardian

PO Box 16185, Birmingham, B2 2WH
Email: customerservices@publicguardian.gsi.gov.uk
Tel: 0300 456 0300
Fax: 0870 739 5780
Open: Monday, Tuesday, Thursday, Friday 9am to 5pm
Wednesday 10am to 5pm

Court of Protection

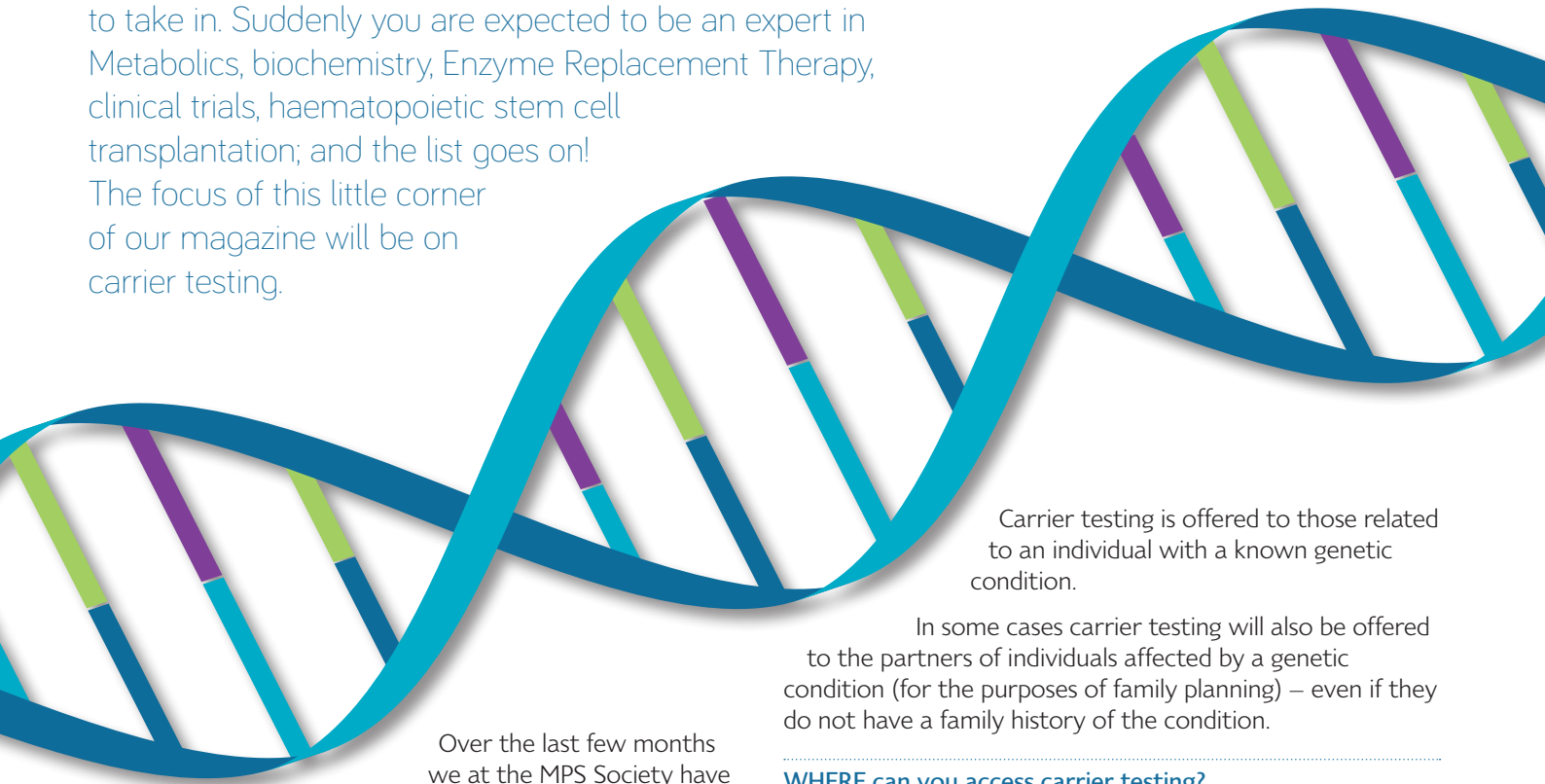
PO Box 70185, First Avenue House, 42-49 High Holborn
London, WC1A 9JA
Email: courtofprotectionenquiries@hmcts.gsi.gov.uk
Tel: 0300 456 4600

Further information

www.gov.uk/make-decisions-for-someone
www.gov.uk/courts-tribunals/court-of-protection
www.gov.uk/government/uploads/system/uploads/attachment_data/file/224660/Mental_Capacity_Act_code_of_practice.pdf

Carrier testing explained

When a family receives a diagnosis of an MPS or related condition it can often feel like there is too much information to take in. Suddenly you are expected to be an expert in Metabolics, biochemistry, Enzyme Replacement Therapy, clinical trials, haematopoietic stem cell transplantation; and the list goes on! The focus of this little corner of our magazine will be on carrier testing.



Over the last few months we at the MPS Society have had a few queries about genetic testing (another one of those areas that MPS families need to become overnight experts in!) and we thought it might be helpful to include a little crash course in this issue of our magazine.

When an individual receives a diagnosis of an MPS or related disease this usually comes after a series of tests. The first of these tests will be analysis of the waste product that is building up in the affected individual's cells as well as analysis to determine which enzyme is missing/deficient. The combination of these two results point towards a diagnosis. The next and final step is to look directly at the genes to identify the route cause – the genetic alterations that are causing the condition.

Once the genetic alterations are identified this information can then be used to test other family members.

WHAT is carrier testing?

Carrier testing is genetic testing of unaffected individuals to determine if they 'carry' a genetic alteration in a disease causing gene. This type of testing involves having a sample of blood taken for genetic analysis. It is important that carrier testing is only carried out once the individual seeking testing has been given information about the testing and the potential results. Being identified as a carrier does not indicate any risk of health problems but it does provide information that might affect reproductive decision making.

Carrier testing is offered to those related to an individual with a known genetic condition.

In some cases carrier testing will also be offered to the partners of individuals affected by a genetic condition (for the purposes of family planning) – even if they do not have a family history of the condition.

WHERE can you access carrier testing?

If you wish to have carrier testing, the best route is to contact your GP who will refer you to a regional genetics centre. It can be helpful to have a copy of your affected relative's genetic results with you so that this can be forwarded to the genetics department. If you do not have a copy of their results it would be helpful for your GP to include the name and date of birth of the affected individual in your family and which Specialist Centre your relative is/was looked after by, in your referral letter.

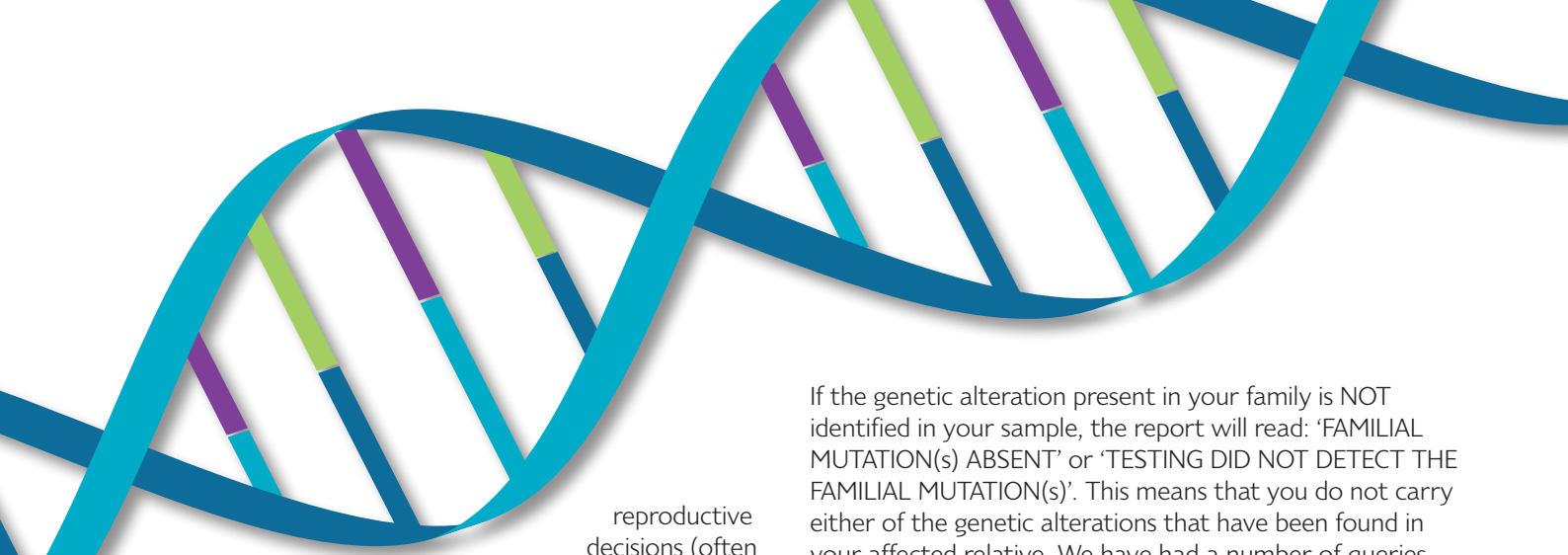
It is not recommended to have genetic testing directly at your GP surgery in most circumstances. A large part of the process of carrier testing is having genetic counselling to support you in understanding the potential outcomes of testing and your options.

If you have any trouble in accessing referral for carrier testing please contact the MPS Society Advocacy Support Team (0345 389 9901) who will be able to advise you.

WHEN can you seek carrier testing?

The parents of a child affected by an MPS or related condition will often be offered carrier testing at the time of diagnosis. Although this does not always happen.

Carrier testing is not an essential test and the results will have no impact on an individual's future health so there is no urgency for an individual to have carrier testing. Those seeking carrier testing usually do so in order to make



reproductive decisions (often to determine the risk of them having a baby affected by the condition). It is perfectly acceptable for an individual related to someone with MPS to decide not to have carrier testing at any point.

The only time when carrier testing is urgent is when the individual seeking testing is already pregnant. If this is the case this should be noted in your referral letter.

Carrier testing is most commonly carried out in individuals with a known family history of a particular condition. For example, the parents and siblings of an individual affected by one of the MPS or related conditions.

An affected individual will have two altered copies of the gene associated with their condition. When family members are being tested the laboratory will look directly for the specific genetic alterations that have been identified in their relative.

It is expected that both parents of an affected individual will be carriers of the condition. Except in those conditions that are X linked (MPS II Hunter & Fabry)

Siblings of an affected individual will have a 50% chance of being carriers of the condition. It is usually recommended that individuals wait until adulthood before seeking carrier testing. However, if you are a teenage sibling of an individual affected by an MPS or related condition your local genetics centre will often be happy to see you to discuss your family history and your options for testing.

Before you have carrier testing the genetic counsellor will often calculate the risk of you having an affected baby based on the information already available.

The RESULTS

Genetic carrier testing results are quite different from other blood tests.

If you are found to be a carrier of the same genetic alteration as your affected family member your report will read: 'FAMILIAL MUTATION PRESENT' or 'X IS HETEROZYGOUS FOR THE FAMILIAL MUTATION' (the wording will vary depending on which lab is undertaking the testing). The results will also detail the specific genetic alteration identified. This information can then be used to recalculate your risk of having an affected baby based on your partner's carrier risk. In some cases, carrier testing may be offered to your partner.

If the genetic alteration present in your family is NOT identified in your sample, the report will read: 'FAMILIAL MUTATION(S) ABSENT' or 'TESTING DID NOT DETECT THE FAMILIAL MUTATION(S)'. This means that you do not carry either of the genetic alterations that have been found in your affected relative. We have had a number of queries about the wording of these reports – instead of stating that you are NOT a carrier, genetic test results will sometimes state that you have a 'VERY LOW RISK' of being a carrier of the condition in question. They may also state that you 'DO NOT CARRY THE MOST COMMON MUTATIONS'. The reason for this is that the lab cannot completely rule out that you have a different genetic alteration that has not been picked up by their test. The chances of this happening are very low but it is important that you are aware of this residual risk.

Should you require any further advice or support in relation to genetic testing please do not hesitate to get in touch and we will do our best to point you in the right direction.

This article focusses on the recessively inherited MPS and related conditions. We will include a focus on X-Linked conditions (Hunter and Fabry Disease) in our next magazine. If any families require advice about X-linked inheritance or genetic testing before the next magazine they can contact the advocacy support team.

“ Carrier testing is genetic testing of unaffected individuals to determine if they 'carry' a genetic alteration in a disease causing gene.



This meeting was a continuum of an important initiative run by Lucid to develop recommendations on best practice clinical interventions in MPSIVA and MPSVI.

The medical and surgical experts included Paul Harmatz (US); Ken Berger (US); Hernan Amartino (Argentina); Tord Alden (US); Andrea Borgo (Italy); John Mitchell (Canada); Elizabeth Braunlin (US); Jim McGill (Australia); Jo Muenzer (US); Jeffrey Gold (US); William Mackenzie (US); Maurizio Scarpa (Italy); Robert Guigliani (Brazil); Chris Hendriksz (UK).

I was joined by Andrew McFadyen from the Isaac Foundation in Canada in presenting 'Leaving a Legacy' a talk giving insight into the MPSIVA and MPSVI patient experiences of medical and surgical interventions. The experiences shared in Box 1 were drawn from five patient organisations: the German MPS Society; Hunter Casa, Brazil; National MPS Society, USA as well as the Isaac Foundation and the UK MPS Society.

As patient representatives we explored six surgical and medical interventions, ERT; Cervical Spine decompression; Hip & knee surgery; Cardiac surgery; Corneal transplant and ENT surgeries that dramatically improved MPSIVA and MPSVI patients quality of life.

Box 1: Key messages in Unmet Needs in MPSIVA and MPSVI

Lack of treatment expertise

- Lack of physician expertise and understanding of MPS diseases
- Misguided or delayed care leading to serious complications

Delayed treatment timing

- Treatment and/or surgeries need to be performed at young age – critical to quality of life and survival
- Causes strain on families and care givers

Access to treatment

- No treatment or delayed access to treatment in many countries
- Stressful situation for the patient and their family

Need for infrastructure

- Necessity to adapt homes and vehicle
- Cost implications for the family and /or individual with MPS
- Society is not set up to support height issues

Achieving Independence

- Difficult to gain control over life
- Managing wheelchair dependency
- Unknown life expectancy

Prejudice

- Lack of public awareness of MPS diseases
- Disadvantaged in securing employment despite high level of intelligence

International



MPS Consensus Programme Second Steering Committee Meeting 2–3 September 2017 Rio de Janeiro, Brazil

Key observations were:

- Reduced pain
- Enhanced mobility and energy
- Increased independence
- Increased social opportunities and interactions with peers and colleagues
- Improved ability to go to work

Factors that benefitted patient prognosis and quality of life included:

- Early intervention
- Performing of prophylactic and proactive assessments
- Decision making for surgery being based on multiple factors including imaging, patient reported outcomes; balance of risks versus improved quality of life
- Need for experienced physicians for challenging surgeries and high-risk patients
- Highly experienced anaesthetists in MPS patients
- First class patient management

Andrew and I having given the expert clinicians and surgeons a graphic insight into the life of patients with MPSIVA and MPSVI they went on to spend nearly two days discussing best practice across a number of medical and surgical interventions. The recommendations will now be subjected to the Delphi process and the outcome will be published in a few months' time.

Christine Lavery

Our ambition as a charity is to support all those affected by Mucopolysaccharide and related diseases and reach all families of children, and to be there as soon as they need us.



Gene Therapy for MPSIIIB at University of Manchester

The Stem Cell & Neurotherapies Group at the University of Manchester led by Dr Brian Bigger have just finished a long study on a haematopoietic stem cell gene therapy (HSC GT) approach for MPSIIIB compared to normal haematopoietic Stem Cell Transplant (HSCT) and also to the anti-inflammatory drug prednisolone. It is important to note that although prednisolone does mediate some positive effects and is also useful in combination with Haematopoietic Stem Cell Gene Therapy (HSC-GT), it is not a drug that is safe to deliver in patients. This was more of a proof of principle that inflammation is important in disease progression. Mice do not suffer the same nasty side effects as patients of prolonged steroid use.

Anyhow, HSC-GT appears to completely correct the brain in MPSIIIB mice with normalisation of lysosomal storage in neurons, normalisation of inflammation, correction of hyperactive behaviour and 13% of normal brain enzyme levels. It works even better in the peripheral circulation, with full normalisation of somatic organs and enzyme levels well above normal. It improves but does not normalise survival, but this is similar to what we found in MPSIIIA. This suggests that HSC-GT is a good candidate for treatment in patients. The Group have submitted for publication and presented at the UK MPS Society Conference in July 2017, WORLD, BSGCT and ASGCT 2017.

The Group have also sought funding for completion of a longer term (1 year study) proof that they can transduce human CD34 cells and are negotiating the commercialisation of the treatment.

The remaining steps to go to clinical trial will be the manufacture of the vector under Good Manufacturing Practices (GMP), full scale transductions in normal peripheral blood mononuclear cells (PMBCs) and a bio-distribution study. For obvious reasons any trial design would look a lot like a trial for MPSIIIA

In terms of timeline to trial Dr Bigger says 'this is always a difficult one. In theory we could be in trial in under a year with the right funding and vector availability'.

PROTALIX Biotherapeutics

Protalix Biotherapeutic's new clinical trial will be investigating the treatment of Fabry disease with once-monthly doses of their enzyme replacement therapy, PRX-102.

The FDA has approved Protalix Biotherapeutic's application for a study into the use of a once-monthly dose of PRX-102 in Fabry patients.

What is pegunigalsidase alfa (PRX-102)?

PRX-102 is an enzyme replacement therapy (ERT) in development for the treatment of Fabry disease. The ERT aims to supplement the decreased or absent α -Gal enzyme in people with Fabry. This treatment is injected into the blood via intravenous infusions, in the same way as currently available ERTs Fabrazyme® and Replagal®.

The α -gal A used in the PRX-102 ERT has been modified so that it is more stable and lasts longer in the body than current treatments.

What have previous clinical trials shown?

Protalix's Phase I/II clinical trial in 16 Fabry patients found PRX-102 to be safe and well tolerated when given every

two weeks by intravenous infusion. All patients on the trial showed stable heart and kidney function, and significantly reduced pain at the end of the study. It also found that a 2 mg/kg dose of the drug lasted approximately 40 times longer in the body than other ERT therapies which require infusion every two weeks.

What does this mean for people living with Fabry?

These results suggest that PRX-102 could potentially be given as a Fabry disease treatment in once-monthly doses, instead of every two weeks. The clinical trial that the FDA has approved will aim to confirm this theory. Monthly infusions would minimise the number of visits Fabry patients have to make to their treatment centers and would go a long way to help normalize their lives.

What's next for PRX-102?

Protalix plan to enrol 30 Fabry patients who will switch from a currently approved ERT to monthly infusions of PRX-102. They plan to begin this study in the second half of 2017.

Research & treatment

Abnormal polyamine metabolism is unique to the neuropathic forms of MPS

Potential for biomarker development and insight into pathogenesis

A newly discovered biomarker associated with a rare metabolic disorder may facilitate better diagnosis and identification of new drugs for clinical trials for the disease, according to researchers in the Perelman School of Medicine at the University of Pennsylvania. Their findings are described in *Human Molecular Genetics*. Development of treatments for the neurological symptoms of mucopolysaccharidoses (MPS), a family of rare genetic disorders, have been hindered by the lack of objective measures of the extent of central nervous system (CNS) damage in patients.

“This new biomarker for CNS symptoms in MPS patients may help families better understand their child’s diagnosis and prognosis and should help clinicians and regulatory agencies to evaluate the efficacy of new therapies”

said senior author James Wilson, MD, PhD, a professor of Medicine and director of the Orphan Disease Center (ODC) at Penn.

The ODC team screened metabolites from cerebrospinal fluid (CSF) in a canine model of MPS I. This assay revealed a marked elevation of a compound called spermine in affected animals. Gene therapy to reduce CSF spermine corrected brain lesions in these dogs.

Additional studies in cultured neurons from MPS I mice showed that elevated spermine was responsible for the abnormal overgrowth observed in the mouse cells.

In humans, spermine is elevated in the CSF of four MPS subtypes in which cognitive declines are seen, but not in two subtypes in which cognitive function is preserved. In MPS I patients, elevated CSF spermine was restricted to patients with genotypes associated with CNS disease. CSF spermine in these patients was reduced following hematopoietic stem cell transplantation – the only therapy currently capable of improving cognitive outcomes.

“Our findings offer new insights into CNS symptoms in MPS patients,” said first author Christian Hinderer, MD, PhD, ODC Research Director. “These studies suggest CSF spermine could be used as a biomarker to evaluate the outcome of novel therapeutics designed to treat the CNS manifestations of MPS diseases, which will greatly simplify clinical trials.”



A prospective natural history study of Mucopolysaccharidosis Type IIIB (MPSIIIB)

This is an observational natural history study for children up to 18 years of age who have been diagnosed with MPSIIIB. The information gathered from this trial may help inform the design and interpretation of subsequent interventional studies. In this study sponsored by BioMarin Pharmaceuticals no clinical intervention or study drug will be involved.

The Primary Outcome Measures will be based on:

Neurocognitive function assessments based on testing every 24 weeks from Baseline to 192 weeks

Behavioural Function assessed using an MPSIIIB specific behaviour rating scale based on testing every 24 weeks from Baseline to 192 weeks

Quality of Life Tests will be used to capture physical, mental and social well-being of the patient as well as to examine the impact of the patient's disease on the parent / guardian and family based on testing every 24 weeks from Baseline to 192 weeks

Sleep habits will be assessed using the Children's Sleep Habits questionnaire based on testing every 24 weeks from Baseline to 192 weeks

Blood and urine samples will be used to evaluate biochemical, molecular, cellular and genetic markers of burden of disease at Baseline

Estimated enrolment: 60

Anticipated start date: Quarter 3 of 2017

Estimated completion date: November 2022

Eligibility: ages up to 18 years

Study population: males and females with a documented diagnosis of MPSIIIB

Exclusion criteria

- Has another neurological illness that may have caused cognitive decline
- Has received stem cell, gene therapy or enzyme replacement therapy for MPSIIIB
- Has received any investigational medication within 30 days prior to the Baseline visit or is scheduled to receive any investigational drug during the course of the study
- Has a medical condition or extenuating circumstance that, in the opinion of the investigator might compromise the patient's ability to comply with the protocol requirements
- Is currently participating in another natural history study

Currently there is not a clinical trial site in the United Kingdom however if you are seriously interested in your child participating in this study please contact Christine at the MPS Society at c.lavery@mpssociety.org.uk

Lucerastat in development for Fabry disease

Lucerastat is a small molecule iminosugar that inhibits glucosylceramide synthase and has the potential to provide substrate reduction therapy for the oral treatment of certain lysosomal storage disorders. It is being evaluated for the treatment of Fabry disease, a X-linked genetic disorder that is estimated to impact approximately 5,600 patients in the US and 5 major EU countries.

In an exploratory clinical study in patients suffering from Fabry disease receiving enzyme replacement therapy, treatment with oral Lucerastat demonstrated a marked decrease in the plasma levels of metabolic substrates thought to be related to the development of the disease. Lucerastat is an oral monotherapy that has potential for patients with Fabry disease regardless of their mutation. The design of a pivotal Phase 3 study, expected to start in 2018, is currently under discussions with health authorities.

Lucerastat for Fabry disease has received Orphan Drug designation in the US and in the EU.

Fabry members
your help is
NEEDED

National study on involvement of the heart in Fabry Disease

University Hospital Birmingham are conducting a national study supported by the MPS Society, which is focussing on involvement of the heart in Fabry disease.

As part of this study we are trying to get as much information from all patients who have had any cardiac device implanted. This would include a pacemaker, defibrillator or a Reveal device.

If this includes you and you would be happy to help in this study, we would be extremely grateful if you could get in touch with Dr Ravi Vijapurapu the Clinical Research Fellow conducting this study on the following secure email address: fabry@uhb.nhs.uk.

The type of information requested from you is:

- What type of cardiac device do you have (pacemaker, defibrillator or Reveal).
- Which hospital do your pacemaker checks take place?
- If you have a defibrillator, have you ever had a shock from the device?
- Who is your consultant Cardiologist and where do they see you?

Thank you for your help in this very important study.

Christine Lavery

European Medicines Agency orphan designation for Fabry disease

On 20 March 2017, orphan designation (EU/3/17/1849) was granted by the European Commission to Freeline Therapeutics Ltd, United Kingdom, for adeno-associated viral vector serotype 8 containing the human alpha-galactosidase A gene for treatment of Fabry disease.

How is this medicine expected to work?

This medicine is made up of a virus that contains the gene for alpha-galactosidase A, the enzyme the patient lacks. When given by injection,

it is expected that the virus will carry the gene into the patient's liver cells, allowing the patient to start producing the missing enzyme and thereby relieve symptoms of the disease. The virus used in this medicine ('adeno-associated virus') does not cause disease in humans.

What is the stage of development of this medicine?

The effects of the medicine have been evaluated in experimental models.

At the time of submission of the application for orphan designation,

no clinical trials with the medicine in patients with Fabry disease had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for Fabry disease or designated as an orphan medicinal product elsewhere for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 16 February 2017 recommending the granting of this designation.

Inventiva's iMProveS (improve MPS treatment) Phase IIa Clinical Study to start patient recruitment before year end

The biopharmaceutical company Inventiva aims to develop and provide patients with new therapies and is currently developing odiparcil as a new approach to treat several forms of mucopolysaccharidosis (MPS).

The orally-administrated small molecule odiparcil (formerly IVA336) was initially developed for the prevention of

Odiparcil

the first oral treatment for MPS VI patients

post-operative thrombosis and has so far been studied in over 700 healthy volunteers and 1100 patients in this indication. Odiparcil can increase the production of

two circulating glycosaminoglycans (GAGs), dermatan and chondroitin sulfate, of which dermatan sulfate inhibits thrombus formation without causing bleeding.

After analysis of its mechanism of action, Inventiva discovered and demonstrated its potential in the treatment of several forms of MPS, in particular MPS I (Hurler/Scheie syndromes), MPS II (Hunters syndrome) and MPS VI (Maroteaux-Lamy syndrome).

Unlike enzyme replacement therapy (ERT), odiparcil is well distributed to organs and tissues, which may improve the treatment of bone, joint and corneal lesions. It is able to reduce lysosomal accumulation in patients' cells by producing soluble glycosaminoglycans (GAGs) which can be then secreted outside the cells.

As a result of its unique mechanism, odiparcil by resolving the symptoms occurring in the eye, joints, cartilages and cardiac valves may address some of the so far unmet medical needs in MPS VI and become the first substrate replacement therapy in this indication.

Inventiva is engaged in a clinical program to validate the potential of odiparcil in MPS patients. The clinical program includes:

- a biomarker study in MPS VI patients;
- a phase IIa clinical study named iMProveS to investigate the safety and efficacy of odiparcil in MPS VI patients;
- a phase Ib study in children with MPS VI to investigate safety and pharmacokinetics; and
- pivotal phase III clinical studies to obtain marketing approval for MPS I, II and VI.

iMProveS (improve MPS treatment) clinical study aims to investigate the safety, tolerability and efficacy of odiparcil in MPS VI patients. It is a 26-week phase IIa study, with 24 patients diagnosed with MPS VI, male or female of at least 16 years of age, with the exception of persons with coagulation deficiency and pregnant women. Patients receiving ERT on a regular basis and for more than 6 months will receive two doses of odiparcil (250 mg and 500 mg, two times a day) with ERT therapy versus a placebo. The study will also include an additional arm where six patients untreated by ERT will receive a 500 mg dose of odiparcil two times a day. The study is currently planned to run in two clinical centres in the UK and Germany from the fourth quarter of 2017.

In parallel to the iMProveS study, a short phase Ib study in children will be conducted mainly to determine the dose to be administered during the phase III.

If positive, the iMProveS study will allow Inventiva to initiate pivotal phase III trials in MPS I, II and VI

For information about the iMProveS trial please contact Mireille Tallandier at Inventiva (Mireille.tallandier@inventivapharma.com) or visit Inventiva's iMProveS web site www.improves-mpsvi-trial.com


IMPROVE MPS TREATMENT



MPS Commercial

Managed Access Agreements

- Our dedicated Managed Access team are able to meet the unique demands of the new and evolving Managed Access route for the treatment of rare diseases
- Our close association with the MPS Society and other patient organisations gives us unparalleled access to patient groups and disease expertise
- We have well established contacts with expert clinicians and clinical centres, NHS England and NICE

Research and communications

- Our highly qualified and experienced team can create a bespoke solution to answer your research question
- We will design and conduct a custom survey to meet your needs
- We provide full publication support for presentation at International congresses and manuscript development

Clinical trial logistics

- We are experts in the ultra-rare diseases field, providing fully managed logistics to ensure patients can access clinical trials
- Our service includes out of hours support, which can be accessed by non-English speaking families
- We provide a patient-led service and are experienced in finding solutions to unique challenges

Supporting the MPS Society

As the non-profit subsidiary of the MPS Society, we are proud of the contribution we are able to make to the work of the charity.



Vimizim Managed Access Agreement

We would like to thank all those on the Vimizim Managed Access Agreement for taking the time to complete the 4 monthly telephone calls with us to collect your quality of life information. We look forward to speaking to you again soon. If you have any questions regarding these calls please contact Samantha Wiseman at s.wiseman@mpspact.com

We were pleased that the latest poster (see page 35) we developed to support the Vimizim Managed Access Agreement was accepted for presentation at the 13th International Congress of Inborn Errors of Metabolism (ICIEM 2017) held in Rio de Janeiro, Brazil between 5-8th September.

Meet the team

Christine Lavery
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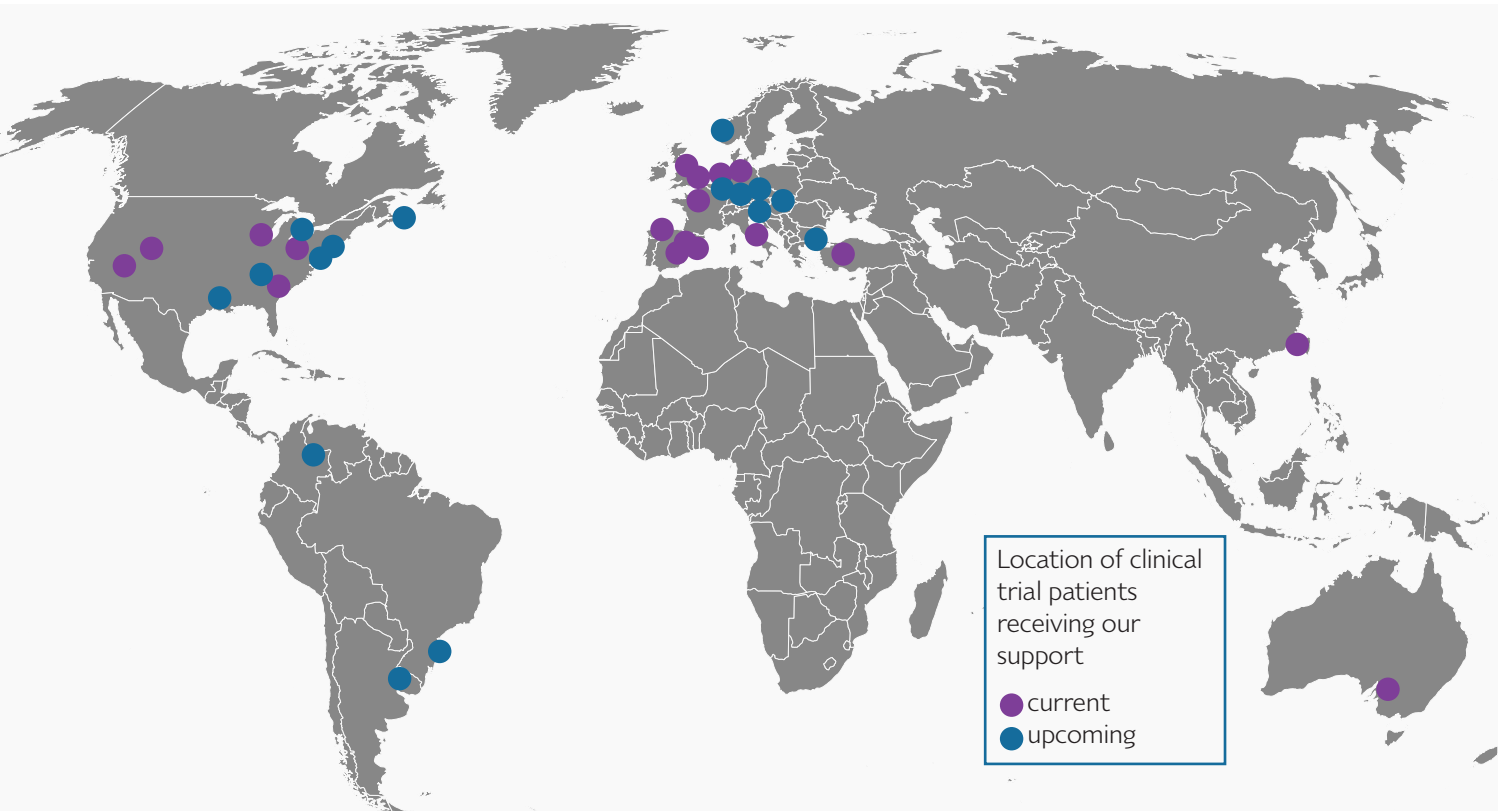
Sam Wiseman
Clinical Project Administrator
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Sophie Henry
Clinical Trial and Finance Administrator
s.henry@mpspact.com



Clinical trial logistics

We are currently supporting patients in clinical trials in 15 countries across the globe



Research and communications

Here is an overview of projects MPS Commercial have been busy working on over the past three months:

A study to review the needs of individuals with MPS VI Marateaux Lamy

Thank you to everyone who has taken part so far. Ideally we need one more person to complete this. If you would like to take part please contact Jackie at j.adam@mpspect.com

Fabry focus group

We recently held a successful focus group for one of our pharmaceutical clients. Thank you to everyone who took part on the day.

Understanding Fabry in families

This survey aims to gain insight into pedigree testing (looking at how Fabry may be passed down through a family) and the level of genetic support across individual Fabry International member countries.

We received over 700 responses so thanks to all of you who took part. We look forward to sharing the results with you later in the year.

International Collaborative MPS III Survey

This project is taking place across Europe in Germany, Switzerland, Austria, Spain, Greece, Serbia and the UK to understand the pathway to diagnosis in each country in order to help aid earlier diagnosis of those with MPS III in the future. The survey also examines the natural history and burden of MPS III. We are hoping 200 people will take part which will make it one of the

largest studies of its kind in MPS III undertaken.

If you would like to take part please contact Alex Morrison at a.morrison@mpspect.com

Alpha-Mannosidosis Disease Progression Survey

We are currently conducting some research on Alpha-Mannosidosis. If you would like to take part please contact Jackie at j.adam@mpspect.com

Supporting the MPS Society

In 2016 and 2017 we have sponsored the following projects through grant awards.

In-depth characterisation of Fabry patients with cardiac devices to predict risk of malignant arrhythmia and sudden cardiac death – Tarekegn Geberhiwot (Queen Elizabeth Hospital, Birmingham & Institute of Metabolism and System Research), Derralynn Hughes (Royal Free Hospital, University College London), Ana Jovanovic (Salford Royal NHS Foundation Trust, Manchester), and Richard Steeds (Queen Elizabeth Hospital, Birmingham).

The value of portable technologies in recording day to day patient monitored information in children and young people with Fabry disease: A pilot study – Uma Ramaswami (Royal Free Hospital, University College London)

Assessing the bioavailability of Genistein in patients with MPS II, MPS IVA and MPS VI – Gisselle Wilcox (Salford Royal NHS Foundation Trust, Manchester)

Impact of elosulfase alfa treatment on patient-reported outcomes in Morquio A Syndrome: results from the first year of an English managed access agreement

Christine Lavery¹, Simon A Jones², Derralynn Hughes³, Elaine Murphy⁴, Ana Jovanovic⁵, Christian J Hendriks⁶, Maureen Cleary⁷, Tarekegn Hiwot⁸, Suresh Vijay⁹

¹Society for Mucopolysaccharide Diseases, Amersham, UK; ²Willink Unit, Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester Academic Health Sciences Centre, University of Manchester, Manchester, UK; ³Royal Free London NHS FT & UC, London, UK; ⁴Charles Dent Metabolic Unit, National Hospital for Neurology and Neurosurgery, Queen Square, London, UK; ⁵Salford Royal NHS Foundation Trust, Salford, UK; ⁶University of Pretoria Department of Paediatrics and Child Health, Pretoria, South Africa; ⁷Department of Metabolic Medicine, Great Ormond Street Hospital, London, UK; ⁸New Queen Elizabeth Hospital, Birmingham, UK; ⁹Birmingham Children's Hospital NHS Foundation Trust, Birmingham, UK

Background

- Morquio A syndrome is an ultra-rare, inherited, multi-systemic disease which, if untreated, results in impaired functioning, mobility, and quality of life (QoL), and early death
- Enzyme replacement therapy with elosulfase alfa is the only approved treatment
- In England, access to elosulfase alfa is granted to all patients on a conditional basis through a managed access agreement (MAA) (Figure 1)
- Patients must fulfil four out of five response criteria to continue receiving treatment; one of the five criteria covers patient-reported outcomes (PROs)
- PROs support continuing treatment if stabilization or improvement are reported in two of the following three domains: QoL, depression, and pain
- PROs for those patients completing the first year of the program are reported herein

Methods

- All patients completed PRO assessments on entry to the MAA, and at 4, 8 and 12 months (Table 1)
- PRO questionnaires were completed by either the patient or their parent/carer depending on the age of the patient
- Patients or their parent/carer completed the questionnaires either over the telephone or during a face to face interview with a patient organisation representative
- QoL was monitored using the EQ-5D-5L tool and the caregiver assistance domain of the MPS Health Assessment Questionnaire (MPS HAQ)
- The Beck Depression Inventory (BDI); only applicable for patients ≥ 13 years) was used to assess mood
- Pain was measured using the Adolescent and Paediatric Pain Tool (APPT) for patients under 18 years of age or the Brief Pain Inventory (BPI) for patients aged 18 years and over
- Thresholds for clinically meaningful changes versus inherent assessment variability were established post-hoc by the MAA stakeholders (Table 2)

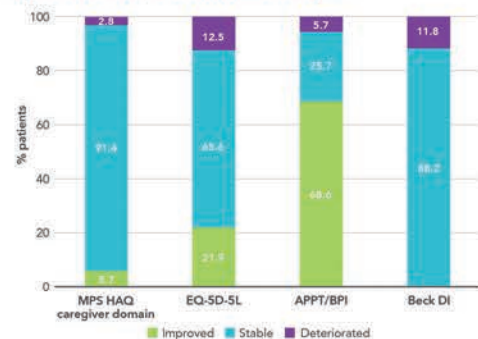
Conclusions

- Assessment of multiple domains was a critical component of the program due to patient heterogeneity and the importance of individualized patient management in Morquio A syndrome
- BDI is a new tool for use in this patient cohort and the exact meaning of changes in this measure will have to be further explored in the future. The current changes seen in the naive patients may be linked to the initial improvement in fatigue which is widely reported and to which patients may adapt over time.
- Based on PROs, the majority of patients met or exceeded the necessary level of treatment benefit established by the MAA stakeholders

Results

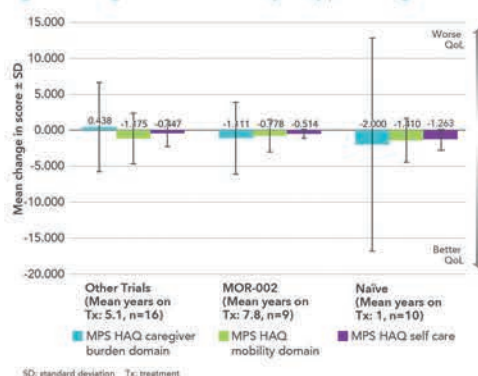
- As of March 2017, 25 children and 10 adults had completed one year of treatment under the MAA
- Ten patients entered the program treatment-naïve, the remainder came from the clinical trial program (mean years on treatment=6.08 [SD 1.36]; n=25)
- The assessment of PROs versus the agreed clinically meaningful changes are summarised in Figure 2

Figure 2. Patient reported outcomes at one year



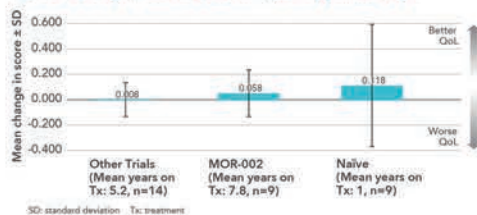
- Overall, PROs provided evidence supporting continued treatment for 33 of 35 patients
- Results for the individual measures are presented below, results for patients who took part in the MOR 002 study are presented separately as these patients have received elosulfase alfa for the longest period prior to the start of the MAA
- Mean QoL scores for the caregiver burden domain of the MPS HAQ are shown in Figure 3
- MPS HAQ mobility and self care domains were also collected but are not part of the MAA criteria. Changes in mean scores are shown in Figure 3

Figure 3. Change in MPS HAQ over 1 year by patient origin



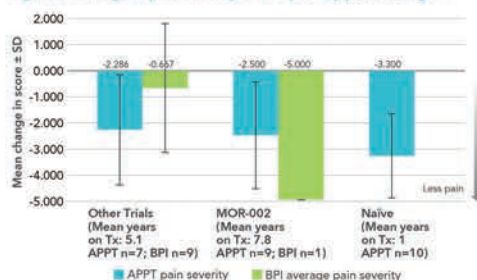
- Mean changes in EQ-5D-5L were positive in both patient groups (Figure 4)

Figure 4. Change in EQ-5D-5L over 1 year by patient origin



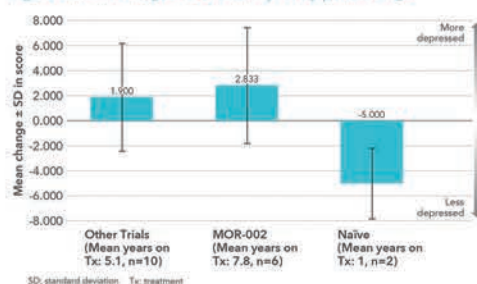
- Mean changes in pain scores showed a pain reduction of over 2 points as measured by the APPT and 1 point as measured by the BPI

Figure 5. Change in pain severity over 1 year by patient origin



- The mean changes in BDI score are shown in Figure 6. A score of 13 or under is considered as normal (i.e. no depression) on the BDI¹ and it is worth noting that no treatment naïve patients and only 1 ex-trial patient scored over 13 on entry to the MAA.

Figure 6. Mean change in BDI over 1 year by patient origin



References

- Smarr KL. Measures of Depression and Depressive Symptoms. Arthritis & Rheumatism (Arthritis Care & Research) 2003; 49(S5): S134-46
- NICE. Managed Access Agreement Elosulfase alfa for treating mucopolysaccharidosis type IVa. 16 December 2015. Available from: <https://www.nice.org.uk/guidance/hst2/resources/managed-access-agreement-december-2015-2238935869> (accessed on 19 June 2017)

Figure 1. The Managed Access Agreement criteria¹

Start criteria

- Confirmed diagnosis of MPS IVA
- Confirmed enzymatic test, elevated urinary keratan sulfate and mutation analysis
- Sign up to the 'Managed Access Patient Agreement'
- Full set of baseline assessments obtained for patients over 5 years of age

Exclusion criteria

- Patient is diagnosed with an additional progressive life limiting condition where treatment would not provide long term benefit e.g. cancer or multiple sclerosis
- Patient has a forced vital capacity of less than 0.3 litres and requires ventilator assistance
- Patient is unwilling to comply with the associated monitoring criteria

Stop criteria

- Non-compliance with assessments for continued therapy (non-compliance is defined as fewer than three attendances for assessment in any 14 month period)
- Patient fails to meet 4 of the 5 treatment response criteria (Table 1)
- Patient is unable to tolerate infusions due to infusion related severe adverse events that cannot be resolved

Patients who are taken off treatment will continue to be monitored for disease deterioration and supported with other clinical measures

Table 1. PRO assessment schedule

Assessment	Baseline	Month 4	Month 8	Month 12
MPS HAQ	X			X
EQ-5D-5L	X			X
BPI/APPT	X	X*	X*	X*
BDI	X		X	

*Day before and day after infusion

Table 2. Thresholds for PRO measurements

	MPS HAQ Caregiver domain	EQ-5D-5L	APPT / BPI	BDI
Clinically meaningful change	Change in point score of 13 or more	Change in value set of 0.2 or more*	Change in 12 month best score versus baseline of 2 or more (APPT) or 3 or more (BPI)	Change in point score of 10 or more

*The EQ-5D-5L threshold for the MAA is still under review, a 0.2 change in value was used for this analysis

Acknowledgments

- Patient questionnaires were conducted by MPS Commercial
- Medical writing and editorial support was provided by MPS Commercial and funded by BioMarin Europe Ltd.
- Data analysis and review were provided by BioMarin Europe Ltd. and BioMarin Pharmaceuticals Inc.
- This research was funded by BioMarin Europe Ltd. under the requirements of the MAA

MPS Commercial needs your help

We currently have three research studies that we need your help with.

Alpha-mannosidosis research study

We would like to get information on alpha mannosidosis, how it feels for you to live with the disease, and how this affects day-to-day quality of life for you and your family members. There are four questions that we would like your support to try and answer:

- How the symptoms and signs of alpha mannosidosis change over time?
- How alpha mannosidosis is diagnosed and looked after by the National Health Service (NHS)?
- How having alpha mannosidosis affects your day to day quality of life and health in general?
- How alpha mannosidosis affects any people that care for you, and your family members?

There are three parts to the survey:

- Part One: A short printed questionnaire which you complete and send back to us
- Part Two: An interview with a representative from MPS Commercial, during which you will complete a more detailed questionnaire. This can take place over the phone, in your home, or at another convenient location to you
- Part Three: After Part Two, we may ask you to complete another face-to-face interview with a representative from MPS Commercial. This can take place over the phone, in your home, or at another convenient location to you. This can either be arranged at the same time as part two, or on a separate day.

If you complete Part Two and/or Part Three of the survey, we will compensate you with a voucher worth £30 in recognition of your time and valuable contribution to the survey.

If you are interested in taking part in this research, please email Jackie at MPS Commercial – j.adam@mpspact.com.

MPS VI research study

We are developing a questionnaire to review the needs of individuals with MPS VI.

The research project will:

- Focus on an individual's educational journey looking at their academic attainment, support needs and overall experiences.
- Obtain informed consent to enable urinary GAG results to be collected to identify disease progression.

The research consists of a short printed questionnaire which you complete and send back to us (in the stamped addressed envelope provided) or can be completed over the phone with a representative from MPS Commercial.

If you complete the questionnaire, we will compensate you with a voucher worth £30 in recognition of your time and valuable contribution to the research.

If you are interested in taking part in this research, please email Jackie at MPS Commercial – j.adam@mpspact.com.

European MPS VII survey

This project is planned to take part across Europe during the second half of 2017 to determine the impact of the disease and the challenges and needs of individuals with MPS VII and their carers. As this is one of the rarest of the MPS diseases we are appealing to all those affected to get in touch and take part in the survey

If you are interested in taking part in this research, please email Alex at a.morrison@mpspact.com

Patient Access to Clinical Trials and Treatment (PACT) registered as MPS Commercial

Registered Address:

MPS House, Repton Place,
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Bucks, HP7 9LP, UK

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T: 0345 260 1087
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Board of Directors

Chair: Bob Stevens
Board: Derek Beards,
Georgina Smith,
David Patterson
Secretary: Christine Lavery

MPS Commercial is a Private Limited Company Registered No. 08621283.

MPS Commercial trades as Patient Access to Clinical Trials (MPS PACT), and is a wholly owned, not for profit subsidiary of the Society for Mucopolysaccharide Diseases (the MPS Society), Registered Charity in England and Wales No. 1143472.

MPS Commercial's social objectives are to reinvest any profits for the purposes of education, enhancing needs-led advocacy support, quality of life research and scientific research to the MPS community.

You can now reach MPS Commercial on their own dedicated number: 0345 260 1087

A fond farewell to Charlotte

It is with much sadness that we announce the departure of Charlotte Roberts, the Business Development Manager for MPS Commercial.

Charlotte started work with the MPS Society in July 2014 as Communications Officer.

She was instrumental during the campaign for funding for Vimizim, working tirelessly to help bring awareness to the plight of those affected by Morquio, and the initial decision of the NHS/NICE not to fund the drug.

After helping to establish the Managed Access Agreement for those needing access to Vimizim, the first of its kind, she became Business Development Manager for MPS Commercial in 2015. While in this role, she has overseen the continued operation of the Managed Access Agreement, research and surveys as well as the management of the Patient Access to Clinical Trials team.

During her time with MPS Commercial she has encouraged all of us to take an active part in growing the business and exposing us to new experiences.

We would like to thank everyone who contributed to the photo album we put together about the Vimizim campaign. We know that Charlotte enjoyed working with you all, despite the circumstances, and that she will miss you all.

I hope you will all join us in wishing Charlotte all the best in her new career.

Charlotte's new role is with BioMarin, and we share with you the invoice we submitted following Charlotte's appointment with them.



MPS Commercial
MPS House, Repton Place
White Lion Road, Amersham
Buckinghamshire, HP7 9LP, UK

0345 389 9901
mps@mpscommercial.com
www.mpsociety.org.uk/commercial

Invoice No: VACANT

Invoice Date: 10.08.2017

**BioMarin Pharmaceutical Inc
Attn: Accounts Payable
105 Digital Drive
Novato
CA94949
USA**

Attention: Paul Humphrey, Maria Meehan, Nigel Nicholls

Description

Supplied: One **highly** esteemed leader

Cost:

Tissues – to dry our tears
Time – to mourn our loss
Understanding – that we may not want to talk to you for a while
Forgiveness – for all of the bad things we currently think about you
Cloning machine – to get another manager of this quality
Chocolate – for comfort eating
Candles – to settle our shattered nerves
Alcohol – to dull our pain

Payment due 30 days from receipt of Charlotte
Return accepted – with open arms

This invoice is being sent on behalf of the MPS Commercial Team.

MPS Commercial is a Private Limited Company Registered No 08627065
MPS Commercial trades as Patient Access to Clinical Trials (MPS PACT) and is a wholly owned, not for profit subsidiary
of the Society for Morquio Syndrome Diseases (the MPS Society), Registered Charity in England and Wales No. 1143472.
MPS Commercial's social objectives are to reinvest any profits for the purposes of education, enhancing needs-led
advocacy support, quality of life research and scientific research to the MPS community.

Fundraising



Nicole got in touch to let us know she was inspired to organise a Hollywood themed party to fundraise for the MPS Society after hearing about similar events. The party is on 29th September so look out for photos in future magazines.

“I came to the family weekend organised by the MPS Society with the conferences and ball which was in Coventry and it was this that made me want to do my own charity event because of all the inspiring stories and seeing how the money can help people like myself.

It’s a Hollywood themed party at the Derriford Health and Leisure Centre in Plymouth. We’ve got dancers, a disco, a singer, a raffle and a buffet.

It’s coming together really well, everyone has been really helpful especially with donating prizes for the raffle! A lot of people want to get involved and are interested in the charity as they haven’t heard much about it before.

It’s been exciting organising it and has given me something to concentrate on but also stressful at times as there’s a lot involved.

We have managed to raise £600 already with just selling tickets and with donations people have given.”



Stolen scooter supports Sanfilippo

In the area of High Wincobank, Christine Lauanders had her mobility scooter stolen and amongst friends started to fund for a replacement scooter. After having her story spotted in Star magazine by Clark and Partners – a mobility firm and work place of Wayne Bond whose son has Sanfilippo – they then stepped forward to provide a free replacement for Christine and in return she donated what had already been raised to MPS Society, which was a total of £150.00





Who sells sea shells on the sea shore?

Elliot Norris, age 11, painted the shells he had found and sold them outside the house, his sister and cousins kept their money but Elliot wanted to donate his because of his friend Sophia Scott.



Birthday girl

Anjali, who turned 6 this year, didn't want any gifts at her birthday party and instead chose to raise money for the MPS Society because of her best friend, Divya Kishore. She raised an amazing £130.

“ Anjali's friend is the true hero

Fashion show

Pat Cutts and the ladies of the Torquay Inner Wheel Club have been raising money all year.

The MPS Society was Pat's chosen charity for her presidential year, after hearing Teresa Jeffery speak about Corey and MPS. They have had fashion shows, a Christmas fair and many other fundraising events throughout

the past 9 months and even Corey made his first public speaking appearance at one of the fashion shows!



Marina does it again

The lovely Marina who featured on the front of the last issue of the magazine has sent a cheque from her shop, Marina and Friends, for £3939.10. This brings the total Marina has raised to £177,295.59 from her shop in Bristol.



Sporty fundraisers

Brand new event next year...

On Sunday 4 March 2017

Created by London Marathon Events Ltd, The Big Half is a new one day festival centred around the half marathon distance, which aims to be truly global and uniquely local in a celebration of the wonderful cultural diversity of the great city of London.

We have now secured 10 places and would like to offer these to any challenge takers on behalf of MPS Society. Just get in touch with our fundraising team at fundraising@mpssociety.org.uk



My very good friend Richard Cooke (or Cookie as he's known around here) took it upon himself to undertake a massive cycle ride from July 19th to 23rd from London to Paris, in all about 300 miles, all for MPS. After hearing about my daughter Hannah who has MPSIIIA he wanted to help and has so far raised £3110.00 in sponsorship. Amazing! We couldn't resist the trip and surprised him at the finish line. Next stop... Barcelona...watch this space, I feel another trip may be in order.

Maggie Louis

Terrie Brown, Paul Brown, Ian Turner and Anne-Josette Maddox all took part in the 100 mile cycle that is Prudential RideLondon and collectively raised a superb £2094



Until my partner's diagnosis officially in March, I hadn't heard of MPS. Since we are keen to raise awareness and finances to help MPS Society support families, getting drugs and support. Especially to those families who are less fortunate than ourselves.

The Prudential bike ride in some ways felt more challenging this year, than it had previously. I honestly felt incredibly proud raising awareness and funds for such a small charity! The bigger charities often take the limelight, mainly due to prevalence! However one of the most heart-warming aspects of taking part in the London Prudential this year is that there is a myriad of small charities, who I felt wearing the MPS shirt supported me on route!

Anne-Josette Maddox

MPS became part of our world when our niece, Anabelle Shepherd, was diagnosed with MPS1 Hurlers in February 2012, just after her first birthday, her journey sadly ended on Christmas day 2012, she developed an infection months after receiving a successful lengthy procedure that gave her a chance of extending her life. Our support for MPS has continued as we feel very passionately about raising awareness and much needed funds for continued support and research.

Upon diagnosis, we felt we needed to do something to support and we decided to take part in the Great South Run, this was a particularly scary challenge for me as I'd never run more than 2 miles! However, Anabelle was being so brave during her treatment, any fears I may have had were nothing compared to what she and her parents were experiencing. Running has since become a very big part of my life and I have taken part in the Great South every year since in memory of Anabelle.

Again with the bike ride I had a moment of madness. Paul had started cycling the year before but I didn't even own a road bike when I put our names forward for charity places! The training was interesting and certainly tested the strength of our relationship at times!

We spoke with the landlady of our local pub and she agreed to support a charity evening. We put a plea out for donations for a raffle and were overwhelmed by the fantastic prizes donated. On the evening of 15 June we held a quiz, play your cards right and the raffle was drawn. It was a fun evening and we cannot thank everyone enough for their support with donations and a very big thank you to Ali from The Lawrence in Southsea who provided food for all that attended the evening at no cost, but encouraged them to donate for each plate eaten. In addition to the funds donated people were genuinely interested in learning about MPS and what it meant for our family and others.

The bike ride itself was an amazing experience and it was great that on the "pit stops" people were checking out the charity tops leaving us with a warm feeling that vital awareness was being raised as well as any financial donations we were receiving.

We raised €924 with a generous additional fund match of €350 from Vodafone as Paul is an employee, so a grand total of €1274. It is an honour to be able to continue taking part in fund and awareness raising events for MPS Society in Anabelle's memory.

Terrie Brown



Charity Golf Day

I had been toying with the idea of a golf charity day for ages, years really. This year I was made Captain of the Rising Sun Golf Society, a Society of a mixed bunch of golfers which a lot of clubs wouldn't entertain. However being Captain I get to choose the charity for a Charity Golf Day, which is the only privilege I have, oh I do have a green shirt with Captain emblazoned on it though.

A committee of four members and Lulu was convened, which at first didn't come up with anything spectacular, in fact we never reached the spectacular. It was decided there would be the obvious raffle tickets, sponsored holes, fines for ending up in a bunker, and an auction, it didn't sound too promising.

Every year the Society jet off to somewhere abroad, this year it was Spain. Here's a tip, if you want to start the cash rolling in, get 24 golfers in a Spanish bar and when all are merry say very loudly "time to cough up £50 to sponsor a hole and nobody can slide off to the loo". The next day we had a count up, £1100 had been pledged.

Most of the raffle prizes tended to be bottles of booze or golf bits. The auction, which did raise quite a lot of money was mainly golf days for 4 players at other golf clubs which were kindly donated by those clubs, Royal St. Georges at Sandwich was a gem at £400 which is half price.

We were lucky with the weather, although it's nearly always windy on our course, the club (Walmer & Kingsdown) being on top of the "White Cliffs of Dover" this gives fantastic views across the channel, as Bob Stevens found out.



“Lulu was over the moon with the amount raised, which to my mind was long overdue, our family has had a great deal from the MPS Society with Lulu and Phoebe having Fabry so we feel good within ourselves to be able to contribute something.”

I think 36 players teed off on the day, we had Society members with friends down from London, who turned out to be very generous. The golf ran a bit late finishing around 6.30.

During the day Lulu, Phoebe and school friend of Phoebe's were busy with the raffle tickets, which in the end raised £400. The funniest part of the evening was the auction, I ended up buying an office chair I didn't want, seems nobody else obviously wanted it, a pair of donated framed pictures of golf scenes went for £200, the bidder then said they "were rubbish and best save them for the next charity day," that didn't please the donor, much to the mirth of everybody else.



A great deal of banter went back and forth, probably too many beverages were had. The day had gone well, a good day's golf, a few ales, a good meal, a great deal of mickey taking and most important of all £3580 raised. One also has to bear in mind that I'm 72 years old and not the oldest in our society, so we did pretty good considering we thought we might get £1500.

Lulu was over the moon with the amount raised, which to my mind was long overdue, our family has had a great deal from the MPS Society with Lulu and Phoebe having Fabry so we feel good within ourselves to be able to contribute something.

Pat Quin

This year we have received some generous donations from artists, so we thought we would tell you a little bit more about their unique work.



Silver Wren and Oak donating Oak leaf pendant value £50

“I have always been inspired by natural surroundings and deeply love and care about our planet and her beauty. After experiencing two tough life changing events, I started making silver and copper jewellery as a therapeutic means of healing and to express how I feel nature nourishes us all. I make 3D sculpture birds and organic leaf pendants - all handmade and unique. Thanks ever so much for asking me to be part of this - it’s a worthy cause and I’m honoured to be part of it in some small way.” Anastasia Robson

www.etsy.com/uk/shop/SilverWrenandOak



Fired Works donating £25 Voucher

Fired Works is a studio and gallery of fused glass art by Marie Cavanagh, Chalfont St Peter Buckinghamshire

Marie produces various pieces from bespoke wall art made with glass powders, sheet glass and crushed glass right down to tiny stud earrings. Marie works mainly with fused glass, using the medium to create anything from functional items such as trinket trays and coasters to large pieces of wall art and splash backs. Each item is hand made so all of her work is unique and while similar items can be created no two pieces will ever be the same. If you would like something made specifically, Marie is always happy to oblige and loves a challenge!

www.fired-works.com

Adam Cope donating a small painting (to be confirmed, NOT the item pictured)

I became ‘a painter’ early on in my life, during in my teens. The painter’s métier has given form and colour to my life. It pleases me. I love it...the paints, the brushes, the slow making of a painting. I enjoy just looking at things. It seems to me more than ever that eyesight is an incredible thing. The visual...arts. Then about thirty years ago, I started teaching. It was a real revelation to me that learning could be just as creative as painting. Creativity is intimately linked to learning.



www.artists-atelier.com

Nati

Top prize

Fabulous three night stay for two including breakfast, in a one bedroom apartment at the Adina Hotel. The winner can also choose this year where to spend this luxury break, between either Berlin, Hamburg, Frankfurt, Nuremburg, Leipzig, Copenhagen or Budapest. Adina Hotels have also thrown in a complementary three course dinner on the first night for two people with wine and water. Transport is not included. Valid until 30 December 2020.

Second prize

Two First Class return tickets with Virgin Trains West Coast Line valid until 31 March 2018.

Third prize

The Staybridge Suites in Birmingham have given a fun one night stay for two people with breakfast. Transport is not included. Valid until 31 March 2018.

Tickets are just £1 each and by purchasing them, you not only stand a chance of winning, but also help to make a difference to the lives of those affected by MPS and related diseases. Why not let your friends, relatives, neighbours and work colleagues stand a chance of winning some prizes by telling them about the MPS Society and getting them to purchase some tickets too!

We will have sent you some tickets with the magazine but if you want some more ticket booklets just let us know on 0345 389 9901 or email fundraising@mpssociety.org.uk.

If for any reason you are unable to sell all the tickets we have provided, please do return them to the office so that they can be passed on to others to sell.

All monies, sold ticket stubs, the accompanying form and any unsold tickets should be returned to the MPS office by Thursday 30 November 2017 at the latest. The draw will take place on Saturday 2 December 2017.

Many thanks and good luck!

It's that time of the year again where you can stand a chance of winning magnificent prizes

Further prizes include:

- A wonderful voucher for Marks and Spencer for the value £150
- Small painting by Adam Cope
- Silver Wren and Oak – Bespoke Silver Oak leaf pendant
- Snopake – £50 Amazon voucher
- Tesco – £50 voucher
- The Entertainer – £50 voucher
- Ocado – £50 voucher
- Bluebeards Revenge – men's grooming products
- Clarins – Gift box
- L'Oreal – Beauty products
- James Galt Toys – Dino Play nest
- Redbush Tea Company – gift bag including notelets, soaps, hardback book and tea
- Everyman Cinema – two tickets
- Fired Works – £25 online Voucher
- Boxclever Press – 2018 Family Life Book Diary
- Bucks Leather – colourful cotton pashmina

Keep checking the website for updates of further prizes as they come in!

onal draw



Thank you to all our donors and fundraisers – you inspire us!

Carol Westland and friends Joan and Lyn raised €265.00 at their local school with a craft stall and items sold at a flea market/collection fair plus bits a pieces that she has sold to friends.

Bowen Masonic Lodge of Beaconsfield donated €400.00.

Robert Locke raised €410.00 in a fishing event.

High Close School in Wokingham raised €15.00 with a cake sale.

Colin Barrett raised €60.00 with a bicycle jumble sale selling bike parts.

Michelle Brooker collected €80 on MPS Awareness Day.

Rosemary and Harry Nurse raised €200.00 at a car boot sale that included donations from her brother, sister and friends and few bits of linen they had spare.

Personal Best Education and all the young people taking part in the spring NCS Programme raised €257.15.

Wayne Bond's company, Clark and Partners, donated €150.00 that was from the client who had their stolen scooter replaced by the company as well as €20 from a kind son of one of the customers.

Clark and Partners also raised €93.81 from their charity collection box in their showroom.

Weatherite Ltd raised €888.00.

Elizabeth and Richard Volk made a surprise visit to conference in July to celebrate the 35th anniversary with Christine and have donated €200.00 to the MPS Society in her name for all that the Society has accomplished.

Brian Keane and his colleagues at Molex in Shannon, Ireland have raised €1150 (including a contribution from their employer) at a Wear it Blue fundraiser as Brian is friends with Ethan who has MPS VI.

Joyce Parkes baked cakes to sell at her husband's work last month and donated €67.88 this month we have received another payment of €46.30.

Carole Cox raised €225.00 on her birthday because of her relation with the Watts family.

Remaining sponsorship of €85.00 was received for Andy Battle and friends who ran the Bath Half in March.

Towersey Morris have raised €600.00 in total this year through collections at morris dancing events and their performance at Christmas 'Mummers Play'.

Wood End Park Academy raised €132.50 in their charity collection.

Jasmine Price (mum to Riley and Jenson Harlock) placed collection boxes in the Garth Owen shop and raised €217.15 plus €52.47 through other donations.

James and Josie Hooper raised €230.00 in lieu of presents on their silver wedding anniversary on behalf of Jamie Macfarlane who suffers from Sanfilippo Disease.

Capital Group matched a gift from Aaron Espin with €100.00. Aaron had donated €50.00 in June.

Rosemary Nurse donated some neckerchiefs.

Daisy Mitcham raised €243.80 with help from the Blossom Street Gallery in York selling more Pom Poms!

Charles Read raised a further €141.50 with the ongoing book sales at work.

Andrea Payne, Dean Finn and Jim Dawson took part in the York Triathlon after a close friend lost her brother in law to Fabry. They raised €180.00.



You raised £60 by sending us your used stamps!

Donations

Joan Crespin, Adrian Morter, Brenda Walker, Heather Silvester, Mrs V Dawson, Roger Taylor, Jean Helen Davy, Ann Baker, Maria Murphy, Stuart Hale, Lauren Wallis, Emma Clewes

Regular contributions by Standing Order or Give As You Earn

Gail Simpson, Sarah Winzar, William Cavanagh, Barbara Harriss, L Brodie, A Sabin, A Ephraim, J Dalligan, M Malcolm, E Mee, Elliot Moody, S & D Greening, M Hahner, K Brown, Z Gul, M Fullalove, E Brock, Margaret Leask, G Reeves, E Parkinson, Gordon Ferrier, R Taylor, R Gregory, L Stillwell, R Henshell, K Bown, S Home, V Little, A Bansal, J Winzar, J Casey, E M Lee, K & J Hudson, Daniel Winzar, J & V Hastings, R & K Dunn, Marcia Tosland, Saville Norman, S Bhachu, C Cullen, S Brown, V M Lucas, E White, C L Hume, A Weston, A Sullivan, A Byrne, Dorothy Robinson, S Cadman,

J Wilson, A Tresidder, J Arnold, E Cox, K Osborne, S Robinson, N Thompson, C Garthwaite, J Ellis, I & V Pearson, C Gibbs, A Cock, A Dickerson, M Kalsi, P Summerton, A Weston, E White, C L Hume, A Sullivan, A Byrne, D Robinson, N Cadman, J Wilson, A Tresidder, J Arnold, Stuart Robinson, K Osborne, Mr Thompson, E Cox, B Weston, J Ellis, I & V Pearson, D & A Gunary, D Forbes

Donations via collection boxes, stamps, foreign coins, mobile phones, ink cartridges, jewellery, PayPal Giving, eBay for charities

Ann Baker, Mark Hughes, Lynne Grandidge, Enola Halleron-Clarke, Brenda Walker, Marilyn Eggleton, Ian Evans

In memory

Jean Maxwell, Luke Bown, Douglas Bennetts Cock, Robert Silcock, Paul Francis Carnill, Lisa Nurse, Kathleen Ghai, Pauline Hammond

Thank you also to all those who donated anonymously – we don't know who you are, but we think you're great!



In June, to help support their friend Clark, Angela Grimes and friends who she met through a postnatal group entered a 2.5km obstacle mud run in Dorking.

“We were very pleased to have totally smashed our target of £500 raising £1,065!”

Together with offline sponsorship the group raised a grand total of £1,125. Speaking of the fundraising Angela said: “we are very proud! We very much hope that the money will be put to good use supporting children like Clark.”



Ben wanted to do the triathlon as his sister Hannah has MPS VI. Ben realises that children with MPS diseases can't do all the things that he can do and he feels this is not fair and wanted to do his bit.

Thank you to those who donated via the Weather Lottery:

Mrs O Megoran
Mrs G Plummer
Mrs D Bown
Ms C Halleron
Miss L Lorimer
Mr A Dickerson
Mrs M Crespin
Mr M Hughes
Mrs T Brown
Mr A Selwood
Mrs J Edwards
Mrs C Lavery
Miss D Halleron
Mrs J Speed
Mrs L Bennett
Mr M Taylor
Miss J Petch
Mrs A Baker
Mrs S Swayne
Mrs K Weedall
Mr N Saville
Mrs J Stather
Miss D Horsley
Mrs J Hopper
Mrs L Brock
Ms M Hartwell
Mr M McCawille
Mrs V Eastwood
Miss L Hiller



Get your walking boots,
trainers or wellies
on and arrange a
WALK!

5 steps to the perfect walk, toddle or roll...

1 Plan
a route



2 Choose
a date

3 Tell your
friends
and family



4 Get
sponsors

5 Have fun



For your walk goodie bag email
fundraising@mpssociety.org.uk
Find out more about walkabouts at
www.mpssociety.org.uk/walkabout

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