MPS

Autumn 2005

Care Today, Hope Tomorrow

Society for
Mucopolysaccharide Diseases

National Registered Charity No. 287034

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

What is the Society for Mucopolysaccharide Diseases?

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

What are the aims of the MPS Society?

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

To bring about more public awareness of MPS and Related Diseases

To promote and support research into MPS and Related Diseases

How does the Society achieve these aims?

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

Telephone Helpline
Includes out of hours listening service

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

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

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

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

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

Information Resources
Publishes specialist disease booklets and other resources including a video

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

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

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

Cover photograph: Myles Broughton (MPS IHS).
Read more about Myles on page 8.

CARE TODAY HOPE, TOMORROW

MPS Magazine

MPS Society

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Registered Charity No. 287034

Newsletter Deadlines

Winter	1 Dec 2005
Spring	1 Mar 2006
Summer	1 Jun 2006
Autumn	1 Sep 2006

Become a Friend of MPS

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

CONTENTS

Welcome to the Autumn MPS Magazine. This edition is particularly full of stories and news from our members. We are always really pleased to include your articles so do keep them coming in!

Whilst writing, we would like to draw your attention to the **Leeds Family Day** being held on **Sunday 27 November 2005** at the **Craiglands Hotel, Ilkley** in West Yorkshire. The Society is holding a private lunch for its members and families. We are grateful to Masterfoods for kindly sponsoring the childrens' meals and entertainment at this event. The day will give members a chance to not only meet up with old friends, but also to make new ones! There will be a three course lunch and entertainment for the children.

All members living in North Yorkshire, West Yorkshire and South Yorkshire as well as the East Riding and Humberside will receive details by post. If any members living outside this area would like to participate please call Linda at the MPS Office for a booking form.

Antonia Crofts
Editor

CHIEF EXECUTIVE'S REPORT

Autumn 2005

With the summer holidays all behind us there is an air of anticipation and excitement in the MPS Office. In 1998 the Society relocated from a two-room office suite to our present offices, 46 Woodside Road, an old adapted house, when there were only five full time members of staff. Nearly nine years on we are ten staff, soon to be eleven, are now feet from a busy roundabout where everybody jumps every time there is a screeching of brakes and the only remaining place to keep the personnel files is in the toilet, so the MPS Society is on the move.

After much searching the MPS Society has found just the right site. MPS House, about one mile from our current office, will provide ample space on the first floor for the Society to provide all its advocacy and support services whilst the ground floor will offer a large meeting and library area. We want the MPS centre to be vibrant, motivating and welcoming, the heart and home for the MPS community whether that is affected children and adults, their families and carers or any of the many professionals in healthcare, education, social services and the pharmaceutical industry that work with us. Whilst MPS House is accessible with ample parking for visitors, it also has the potential to be made accessible to members in all parts of the UK through video conferencing with the advocacy team. The additional space will also enable the Society to utilise the services of committed volunteers for whom we currently just don't have the room. Hopefully we will be able to give you details of our new address in the next newsletter.

Enclosed with this magazine is your registration form for the Society's Alton Towers Fun Weekend 29 – 30 April 2006. We are delighted that we have been able to keep the prices the same as in 2004 and that the Society is able to subsidise the cost for members of the Gala Dinner and Children's Gala Tea and entertainment, the family disco on the Saturday night, Childcare Volunteer costs and entry to Alton Towers on the Sunday. Members may book an additional night (Sunday, it is Bank Holiday Monday on 1 May) through the Society at the time of booking and upgrade your Alton Towers tickets to two days. Last year over 100 members and their families had a brilliant time at Alton Towers so please do book early so as not to be disappointed.

Finally thank you to all of you who are busily selling Annual Draw tickets. Your help in selling draw tickets raises money towards supporting all our members as well as vital research. Whilst Christmas is still some way off I also invite you, your family and friends to support the Society with sales of Christmas cards, Christmas paper and calendars.

Christine Lavery
Chief Executive

new phone & fax number!

The Society now has a **new telephone number** and **fax number**. Please make a note of these numbers as they will be replacing our old telephone and fax numbers.

Telephone: 0845 389 9901
Fax: 0845 389 9902

News from the MANAGEMENT COMMITTEE

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Management Committee Meeting held 9-10 September 2005.

Individual Advocacy

The Chief Executive outlined to Trustees the difficulties facing Scottish and Welsh members accessing clinical assessments outside their respective countries and that both Scotland and Wales are now refusing to fund these assessments and ERT. In order to uphold members' rights the Society is considering challenges for failed due process in both countries. The Chief Executive also confirmed that the Society is undertaking a consultation with its members in Scotland and Wales in respect of diagnostic services, clinical management, treatment and the role of the MPS clinics.

MPS Education and Information Centre

It was confirmed to Trustees that the contract has now been exchanged and the completion date is 21 October 2005 subject to the schedule of works being completed on time. The Chief Executive confirmed that funding towards fitting out the ground floor Educational Centre had been received from two pharmaceutical companies and John Lewis. Further funding applications are still under consideration.

Jeans for Genes

Ann Green, Jeans for Genes Trustee representing MPS, gave an overview of developments particularly the changes to governance. Ann Green, Antonia Crofts and Christine Lavery attended the Jeans for Genes Fashion Show at the Inter Continental Hotel in London to launch Jeans for Genes 2005. The event received considerable press and media interest and attracted many celebrities. Trustees also thanked Bob Devine for all his hard work as the outgoing Jeans for Genes Trustee representing MPS.

International Symposium on MPS Diseases 2006

The Chief Executive advised Trustees that she and Sophie Denham had to date been unable to carry out the required risk assessment on the location of the International Symposium on MPS Diseases to be held 28 June – 1 July 2006 in Venice. It was agreed that until the risk assessment has been undertaken the Society is not in a position to set out its level of practical and financial support to members wishing to participate. (This of course does not preclude UK MPS Society members enrolling for the Symposium directly with the secretariat.)

Policies and Governance

The Trustees reviewed and agreed the following policies: Equal Opportunities, Conduct Policies for the Chief Executive, Staff and Trustees, Mobile Phone policy, Managing Abusive Phonecalls, UK Travel and Subsistence, Overseas Travel and Subsistence, Medical Research Grants, Redundancy, Relocation, Private Shareholding and Financial Interests, Media Handling, Age Discrimination, Maternity, Leave for Domestic and Personal Reasons and Flexible Working Policy. Following on from the Charity Commission risk assessment exercise undertaken at the last Management Committee Meeting the Trustees explored ideas for a Disaster and Operational Continuity Plan. Key risks were identified and the Chief Executive is to draft a plan to bring back to Trustees.

ANNUAL GENERAL MEETING

2005

The Annual General Meeting of the Society took place at the Northampton Hilton Hotel on Saturday 2 July 2005 at 7.30pm. 103 members, 15 guests and 8 employees attended. Apologies were received from Trustees Bob Devine and Paul Sagoo.

The minutes of the Annual General Meeting held on 1 May 2004 were distributed in advance to those members present and were accepted as true and accurate.

The Chairman, Barry Wilson, presented the Trustees Report. This is published in the Annual Report and accounts for the year ending 31 October 2004.

The Treasurer, Judith Evans, presented the statement of accounts for the financial year ending 31 October 2004, the details of which are also to be found in the Society's latest Annual Report. It was proposed and seconded that the Auditors, McLintock and Partners, Chester be appointed the Society's auditors for the financial year ending 31 October 2005.

There having been no other nominations received it was proposed by Barry Wilson and seconded by Rosemary Nurse that Paul Sagoo be elected to the Management Committee and that Wilma Robins and Ann Green be re-elected.

Under any other business the Chairman outlined the way forward for the MPS Society stating that within weeks the Society will be re-locating to premises that will allow the achievement of a major goal, that of creating an MPS Information and Education Centre integral to the Society's advocacy support services. This project is being funded by Jeans for Genes and will enable the Society to reach out to an ever-increasing number of individuals, families, partners, carers and professionals affected by MPS, Fabry and related lysosomal diseases.

Finally, the Chairman warmly welcomed the presence of most of the Society's research grant holders stating that in March 2005 the Trustees awarded record grants and subject to the success of the Jeans for Genes Appeal on 7 October 2005 it is hoped the research programme will continue to expand. The Chairman thanked members, guests and professionals for coming to the Conference and making the weekend so special.

Ellie Gunary

As Ellie moves on to pastures new, I want to take this opportunity to say thank you to Ellie Gunary for her contribution to the MPS Society both as a member and an employee.

Ellie came to MPS in 1999 to lead the advocacy team, and over the next five years she played a significant role in transforming the Society's individual advocacy work. The service Ellie helped develop is today the envy of not only many, many patient support organisations but also of the statutory agencies.

Throughout 2004 those members who felt they were losing the battle for enzyme replacement therapy (ERT) with their Primary Care Trusts will recall the efforts Ellie went to, to challenge due process on your behalf. For those on ERT today for MPS I and Fabry Disease and those waiting for ERT for MPS II and MPS VI you may not appreciate Ellie's fortitude in taking on PCT after PCT and winning the argument in almost every case.

As some of you will know Ellie has been on sick absence over the past six months. After a lot of thought Ellie has now decided to say Goodbye. On behalf of the members of the MPS Society we wish you well for the future

Christine Lavery

Announcements

Introducing Nikki McAuliffe

Hello all! I've just joined the MPS Society as part of the Advocacy Support Team after having recently graduated from university with a first class honours degree in Psychology after three year's hard work.

As some of you may know, I did some work for the Society last year during my summer break helping out on the siblings weekend at the Pioneer Centre as well as researching the epidemiological database, so as you can see, my current role is quite different! In my spare time I enjoy shopping, swimming and yoga, but having just moved house, I don't seem to have a lot of free time on my hands.

Having witnessed the good work that the Society does, I hope I can keep the standards as high as has previously been the case! I'm really looking forward to the challenges that lie ahead of me and getting to meet some of the MPS members in the not too distant future.

New Members

Mr Fullalove and Miss Rowe have recently been in contact with the Society. Their daughter, Libby, has a diagnosis of Sanfilippo Disease. Libby is four years old. The family live in the South East.

Gillian recently contacted the MPS Society and has a diagnosis of Fabry Disease. She is 64 years old and lives in Wales.

Deborah Ruffley has recently been in contact with the Society. Deborah has a diagnosis of Fabry Disease. Deborah lives in the North West.

Ingrid recently contacted the Society and has a diagnosis of Fabry Disease. She is 35 years old and lives in Wales.

Elaine recently contacted the Society and has a diagnosis of Fabry Disease. She is 58 years old and lives in Wales.

Julie recently contacted the Society and has a diagnosis of Fabry Disease. She is 39 years old and lives in Wales.

David was mistakenly diagnosed with MS in 1998 and is now trying to rebuild his life with his new diagnosis of Fabry Disease. He is very keen to meet fellow Fabry sufferers. David can be contacted at vic_badger@hotmail.com

Avril Stokes has recently been in contact with the Society. Avril has a diagnosis of Fabry Disease and she lives in the South West.

Nikki McAuliffe

Deaths

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

Daniel Allen who suffered from Sanfilippo Disease and who died on 5 July 2005 aged 15 years.

Edward Butler who suffered from Sanfilippo Disease and who died on 9 July 2005 aged 14 years.

Benjamin Lemon who suffered from Hunter Disease and who died on 25 July 2005 aged 13 years.

Laura Graham who suffered from Sanfilippo Disease and who died on 29 August 2005 aged 16 years.

Jamie Andrews who suffered from Hunter Disease and who died on 15 September 2005 aged 14 years.

Does your child have Sanfilippo Disease?

My son Rudy is 9 years old and has Sanfilippo A. He can walk a few steps, falls alot and seems to be in pain from his hips. Rudy has alot of weight and cannot swallow. Doctors in South Africa have no experience of Sanfilippo Disease and gastrostomy. I would love to hear from Sanfilippo families who have been through this experience. Write to: Rindie Fensham, 6 Heilbron Street, Crystal Park, Benoni 1515, South Africa.

Standing out from the crowd

Our cover star, Myles Broughton, MPS IHS, recently graduated with an HND in European Travel and Tourism Management.

'I knew personally that in order to get a job I would need an ace card up my sleeve. Applications are made by hundreds of people and you need to be able to stand out from all the applicants. Having MPS shouldn't be seen as a flaw, but a strength.'

Circumstances occurred at school which prevented me from taking my GCSEs. I went to Wakefield College and asked them what I could study at my level at that time. Wakefield College were very kind as they let me start at Foundation level in ICT (information communication technology).

The college had a career ladder that was always on my mind and I always thought it would be nice to reach the top of the ladder. So I went from the Foundation ICT to Intermediate Business and then I was stuck with what to do next. I went on to study key skills while I could figure out what I was really interested in. I have a lot of interest in flying, going on holidays and how it's arranged, so I decided to study AVCE Travel & Tourism which covers a vast area.

By this stage I was on the third level of the college's career ladder and studying really hard. The pressure was really on when I started the enzyme replacement therapy trial. This is due to the treatment basically taking a day out of your week and then recovering from the pre-med through the following day as well. You've got five days and may be a bit more to study and live life to the full in this situation. While studying try your best and no one can ask anything more of you.

I barely passed the AVCE Travel and Tourism course, and then thought what to do next. I really wanted to get the HND European Travel and Tourism Management qualification and reach the top of the college's career ladder, but I knew it would be a lot more difficult than the AVCE course. I like a challenge and so applied anyway!

Two years on, I completed and passed the course! I went to my graduation ceremony at Wakefield Cathedral. My options now are either to get a job or go to Uni. In the future I plan to go for my Masters but this will be a long way off yet. I'm currently applying for jobs and going to interviews confident now that I've collected a whole variety of knowledge needed for the travel industry.

Many congratulations to Myles from all at the MPS Society. Well done!

MARKING SUCCESS AGAINST THE ODDS

In the Winter 2004 edition of the MPS Magazine, we featured Aisha's Story. This was an article written by Aisha's parents about her and her family's experience of living with Morquio Disease, MPS IV. We were so pleased to learn that Aisha has since been nominated for the Young Person of the Year Award for Leicestershire. Read on to learn more...

After a very difficult year last year for Aisha her school nominated her for the Lord Mayor's Young Person of the Year Award 2004/05 for Leicestershire. It is an award which recognises youngsters who have succeeded against the odds. Out of thousands of applications Aisha was one of 20 finalists. The ceremony took place at Leicester Town Hall Square in the Lord Mayor's Chambers on 5 May 2005. Aisha was amongst the inspirational youngsters who had overcome adversity to live happy and successful lives. Aisha was the youngest nominee as all the others were over 10 years of age.

There are no words to express the way we felt as parents on that day. It was a very special day. Her grandparents flew in from Malawi especially. Also present were Aisha's class teacher, Mrs Vivian Evans and Mrs Diane Felstead (her one to one at school). Aisha's school, Whitehall Primary in Leicester, had nominated her for all the difficulties that Aisha faces and overcomes with her disability. The encouragement and support that we have received through the school has enabled her to deal with the difficulties that she faces.

Later that month, the school held an assembly for Aisha which was probably even more special for her as her school friends were present and she was given the Head Teacher's Award. She did a little speech which read:

Good morning everyone:
Thank you for nominating me for the Lord Mayor's Award. It made me very happy to go to the Town Hall Chambers. I would like to thank everyone at school for bearing with me last year. Thank you also to Mrs Evans, Mrs Felstead, Mrs Adam, Miss Brown (school SENCO) and Mr Fewster (Headteacher) for arranging everything.

Since these awards Aisha has had her KS1 Sats and has done extremely well. Her report has surprised us all for all the effort that she has put in since the halo has been removed.

Do you have a great story to share with us?
Send an email to
newsletter@mpssociety.co.uk
or phone
0845 389 9901

MPS Regional Clinics

Scottish Clinic

As those of you living in Scotland will no doubt be aware, there has been no Scottish MPS Clinic this year. The MPS Society are currently in the process of consulting with all our members living in Scotland regarding their diagnosis, clinical management and treatment and the future of the MPS Clinic in Scotland. All these people should have received a letter from the MPS Society concerning this. It is important that we receive feedback from you all as to what is working well in Scotland, what improvements could be made and what you all value in terms of clinical management including new therapies. We would also like your input over who you would like to see at these clinics. If you are a member who has not received our letter, or you would like to put your views forward, then please do not hesitate to contact the Advocacy Support Team.

The advocacy team (left to right): Sophie Denham, Neisha Hall, Linda Norfolk and Nikki McAuliffe

Birmingham Clinic

This is to let members know that Dr Ed Wraith will be unable to attend the next Birmingham Clinic on 26 October 2005. The clinic will be going ahead as planned but if you are a patient of Dr Wraith's and would like to see him in a Birmingham Clinic then please could you contact us at the MPS Office. Additionally, if you have an urgent medical need and would like to see Dr Wraith then please contact the Royal Manchester Children's Hospital. The MPS Society's Advocacy Support Team will be in attendance at the Birmingham Clinic on 26 October 2005 and would be happy to offer support or information to people where needed. We will look forward to seeing you.

If you would like to contact the Society's advocacy team telephone 0845 389 9901 or email advocacy@mpssociety.co.uk

MPS Regional Clinic Dates 2005

Birmingham
Wednesday 26 October

Bristol
Tuesday 22 November

Cardiff
Friday 25 November

Northern Ireland
Thursday 17 November

Tricky travel but super science

Bernie Drayne battles the buses to take daughter Roma, MPS IV, on a learning and sport-filled day out in Belfast.

For our day out in Belfast we made a rule to use public transport. The Translink website offers a guide for disabled travellers showing services available at each train station, ranging from disabled people's parking and audio loops to ramps and low information desks.

The guide is wide-ranging but does not tell you on which bus and train routes the newer, accessible stock operates. So, if you are a wheelchair user, it is advisable to board at main bus terminals where you can wait for low floor services.

Once in the city centre, we caught a cab to take us to the recently opened W5 science museum, part of the Odyssey complex.

The complex is bright and spacious and there are plenty of staff willing to assist if necessary. There was access to all areas and the disabled toilets were clean. There are several restaurants, all of which are navigable except for one which was crowded and not suitable for wheelchairs.

W5 is an interactive science discovery centre that will appeal to all ages. Exhibits feature light, sound, heat, electricity, magnets and much more to stimulate the senses. It would take hours to visit everything – even if you do not become engrossed in a tug of war, lie detectors, building houses or visiting the shop. Most exhibits are at a low level and were easily accessible to my daughter from her wheelchair.

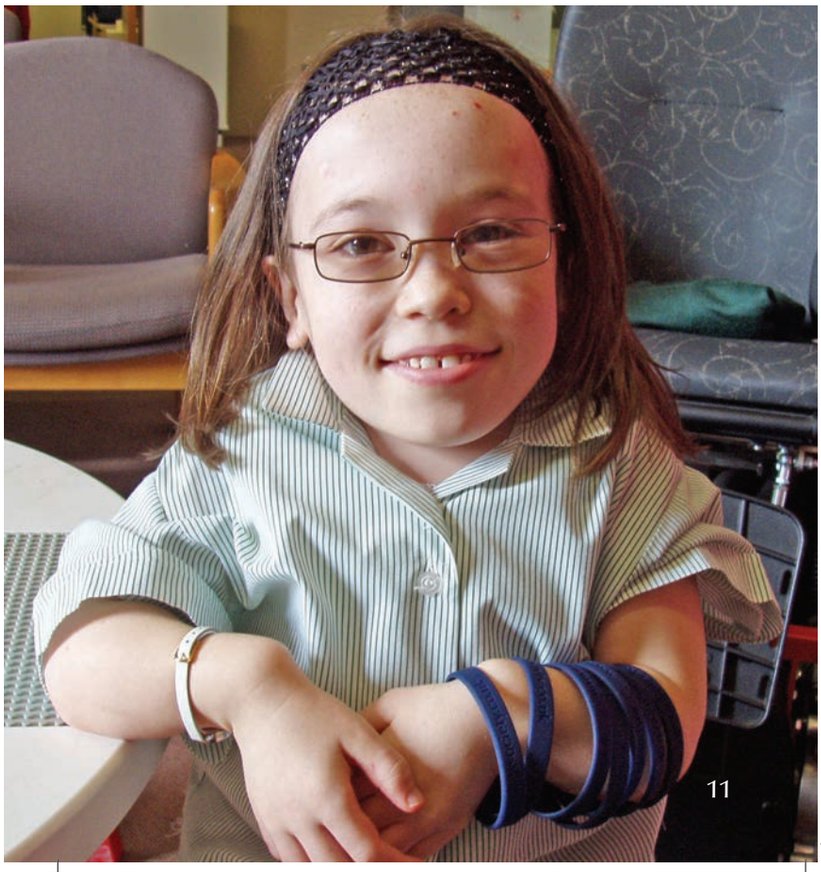
Tired, but happy, we were picked up from the Odyssey car park, which has many disabled car parking bays, and whisked off to Casement Park in Belfast to watch the Gaelic football semi-final between Armagh and Derry.

We had arranged this in advance so we had tickets for a nearby car park for disabled fans. On arrival we produced our passes and were led to the front of the queue. This was really useful, considering the 30,000 strong crowd. Our fully-ramped spectator's area was designed so that no one would be able to stand in front of us and block our view and the stewards were very helpful, moving anyone on who obstructed us.

For more information, including a Holidays, Play and Leisure factsheet, visit www.cafamily.org.uk/nireland or telephone **0808 808 3555.**

This article appears courtesy of Disability Now Magazine. **www.disabilitynow.org.uk**

Do you have any stories, photos or interesting news for the MPS Magazine? If so, email us at newsletter@mpssociety.co.uk



Scottish MPS Family Day at

ALMOND VALLEY by Nikki McAuliffe

On Saturday 3 September 2005, Clare Cogan and myself arrived at the Almond Valley Heritage Centre in Livingston, a small village just outside Edinburgh city centre. This was to be the setting for the MPS Scottish Family Day. Seven families joined us, some familiar faces as well as some who had never been on a Society organised day out.

The day started off in the soft play area giving the children somewhere to let off a bit of steam after their journey from their homes to the centre. This proved to be popular with all the children. I seem to recall a lot of ball throwing! Next on the agenda was a trailer ride around the park, which meant that all the children got to see a variety of farm animals without having to move from the comfort of a chauffeur driven tractor.

We then moved over to the bouncy castle, which we had managed to book so that the families had it all to themselves. This too, seemed to go down well and once the children had finished bouncing, there was just enough time before the picnic lunch to have a play in the adventure playground or go and look at some of the smaller animals.

After lunch in the picnic barn, everyone piled onto the small train and got to admire some of the beautiful Scottish scenery. This gave everyone the chance to let lunch digest before the children went off to play and look at some more of the animals that are looked after at the heritage centre, before heading off home.

I must take this opportunity to say how lovely it was to meet all of the families who came to Almond Valley as well as a huge thank you to all the staff at the heritage centre for being so accommodating and helping to ensure that the day's activities ran so smoothly.

Photos this page clockwise from bottom right: James Morrison; Jordan Mount (MPS I); Lewis Broadley (MPS III) and his family; Megan Broadley and Aaron Brown.






Photos this page clockwise from top right: Norma Broadley, Craig McDougall and Lewis Broadley (MPS III); Luke Morrison (ML II); Megan Broadley; Craig McDougall (MPS II); Connor McDougall (MPS II); Megan Broadley and Lewis Broadley (MPS III), Dawn, Morgan and Craig (MPS II) McDougall

Starting school is a big experience in any child's life. So, when Jordan Mount started school on Tuesday 16 August there was a mixture of emotions in Jordan's house that morning. This was a day his parents thought they would never see as Jordan was diagnosed with MPS I, Hurler Disease, on 29 March 2001 when he was eight months old. Jordan's mum tells their story...

STARTING SCHOOL

If you would like help from the Advocacy Support Team please do give us a call on **0845 389 9901**. Alternatively, if you have a story which you think other readers may benefit from, email us at **newsletter@mpssociety.co.uk** or phone the MPS office.

After two bone marrow transplants and a lot of isolation we were eventually able to mix Jordan with other children his own age. At first we joined our local mother and toddler group. Jordan was very shy and sometimes we had to sit with his coat on for a while. Eventually, however, Jordan came to the conclusion that playing with the other children could actually be fun.

In September 2003 Jordan got a place at Bankhead Nursery. We were allocated five afternoons and at first Jordan found it hard to separate from us. It had been decided between ourselves and the education authorities that Jordan should have one to one support whilst he was at nursery. As Jordan's speech was delayed we also had access to a speech therapist. Once a week someone from the early years support base would visit Jordan in the nursery to help him with things that he struggled with.

Jordan settled in and was not long in winning over all the staff in the nursery. The children all loved Jordan and he made lots of new friends. Jordan has an amazing personality and is such a likeable wee boy that everyone he meets ends up adoring him. He did struggle in some areas, mainly physical activities, but Jordan found that if he could not do tasks the proper way then Jordan would do it his own way – 'Jordan's way'. Jordan did well at nursery and his speech improved. His confidence soared but we still found that with bigger groups, Jordan was reluctant to take part.

In November 2004 the educational psychologist visited myself and Jim at home. We were advised on picking a school for Jordan. Jordan's older brothers had both attended St Columbkilles Primary School which was our local school, but we were advised to look at a school with a support for learning base. We were advised not to send Jordan to a mainstream school as the classes were large and she felt Jordan would struggle.

At first I was upset at this suggestion as I had always thought that Jordan would go to school with Dean (Jordan's brother). Declan, Jordan's eldest brother, was now at local secondary school. We were offered one school but after visiting, we didn't think it would be suitable for Jordan. We contacted our education authority and asked that we were given a list of schools with a support for learning base within the schools.

We then visited two schools in East Kilbride, about twenty minutes drive from our house. The schools were very good and I had a really good feeling about both. Our first choice had been a school called Murray Primary but this school had many stairs and none of the infant classes were on the ground floor. So, instead, we opted for Greenhills Primary School. Initially, the education authorities refused to budge because there was a more local school which they wanted Jordan to attend.

We phoned the MPS Society and spoke to Clare who helped us get Jordan into the school we had chosen. Clare also helped us start a record of needs for Jordan as no one yet had bothered to start one. We were told that, in Scotland, records of need were coming to an end, but we insisted that Jordan should have one anyway until something else was put in place. Many thanks to the MPS Society!

So, Tuesday 16 August has arrived and standing in front of me is this handsome little boy. Where has my baby gone? Jordan looks so grown up that I'm finding it hard to stop the tears.

We bundle him into the car and begin the twenty minute journey to school. Greenhills Primary is a mainstream school with a support for learning base. Jordan is in Room 5 and there will be six other kids in his class. They all require support and there are two staff in the class. The teacher is Mrs Walker and the support assistant is Miss Friery. At school Jordan will be offered speech therapy and an occupational therapist will visit the school. This would not have been guaranteed in our local school.

Jordan went into school on his first day with no bother. In fact, I was more upset than he! On the first day the teacher wrote in his home/school diary that he settled in well and had toast for his snack. Jordan also enjoyed playing with the cars, playdough, sand and blocks. He was great at matching colours and, all in all, had a great first day at school. His teacher also mentioned that she was looking forward to seeing him the next day so Jordan must have been good!

Archie goes to school

Last Christmas, four-year-old Archie Rudham, who has MPS II, started Bramcote Church of England School, and who knew how it would work out? **Sarah Hayward** tells us what happened.

Head teacher, Liz Youngman, is determined to include children with special needs in mainstream schooling. She ensured Archie could come to the school, despite the complications caused by Hunter's. It was uncertain whether Archie would settle in and relate well with the other children.

Due to limited verbal communication, Archie expresses his enthusiastic friendship through physical hugs, kisses and somewhat overwhelming embraces! Likewise, he vents his frustration, through what is commonly known to most in the school as a "bang". Despite misunderstandings, Archie's class has lovingly embraced him. Every playtime the children gather round him to help him play. Beth, Max and William help him perform functional tasks as well. They encourage Archie to line up, follow them to the toilets, wash his hands, sit down for drinks and so on.

Beth (6) supports Archie at lunchtimes, repeating teacher's instructions to him, and encouraging him to sit quietly and sensibly like herself.

Likewise, six-year-old Max, realises the importance of setting an example. In his own words, he helps Archie when he's just about to "bang", "by showing him how not to bang, by being good and not being naughty".

When asked how Archie makes her happy, Beth replied, "He smiles at me and he makes me laugh". Her answer reflects the feelings of many others in the school.

Archie's most favourite place in the school is the courtyard. This is an area set in the school quadrangle, filled with toys for younger children.

When Mrs Youngman asked for older children to help look after Archie in the courtyard over lunch time, the response was so huge, she had to set up a rota, and even turn some down!

Following this experience, the older children were asked what they like doing with Archie, what they love about him, and how the experience benefits them.

Callum Burrows and Robin Jootun (10) usually work as a pair. Both comment on what they love about Archie most – his laugh, the nicknames he gives people and the way he uses his own little phrases. Robin finds it hilarious when Archie says, "Rob, on your bike". Robin also loves the

Photos this page, clockwise from top right: Archie & Chae-Youn try a puzzle together; Archie & Kyra; Beth helping Archie with a book; William, Ali & Archie with sand.

way Archie "knows what everyone should do and how they should do it."

Callum, "or Halli" as Archie would have it, describes Archie as "such an enthusiastic boy, for someone who has the disadvantage of Hunter's." He is very encouraged by working with Archie and believes that helping Archie is "the perfect taster for people who want to work with children".

As with all his helpers, Archie also has great fun with ten-year-old Georgia. She describes their interactions: "I chase Archie with the cars and bike; he loves it! He calls me George! I love it because whenever he sees me he has to say, "hello George!" The way he acts is cute as well, because he always mumbles to himself, not saying anything, just gobbledygook! Now I work with Archie I know more about people with his sort of disease. I love working with Archie!"

Different children play different games with Archie. Annah Hammond, who also plays with Archie at home, has great fun "putting on outfits to make him laugh". Similar to so many children's comments, she says: "Looking after Archie has shown me more about what he enjoys, so me and others can make lunchtimes fun for him. Helping and playing with Archie has been great."

Photos this page, clockwise from bottom right: Ruby, Archie, Suzy & Annah - let's dress up; Archie, Jordan & Georgia enjoy the sand pit; Jordan talking to Archie in his beloved car.

The Society would like to thank Sarah Hayward and all the children who kindly agreed to feature in this article.

Ruby Turner (10) and Suzy Davis (11) usually pair up to work with Archie. Ruby explains that she was unsure about helping with Archie at first, but continues, "I started to enjoy him so much when we played". Suzy also makes her enthusiasm for Archie unmistakably clear: "I think that children with problems should have just the same chance to do things as any other kid… I love everything about Archie!" Suzy and her Mum wrote this poem:

Archie came to my school one day
He stayed a while and had a play
He slowly stayed for longer each time
And now he's a special little friend of mine.

I love to help Archie he makes me smile
His giggle is great – you should stop by for a while
Archie is kind, and gives us all funny names
I'm Scooby or Doody, not often the same!

Our school is better for having Archie around
He's happy, he's busy, and special I've found,
He sometimes gets tired and cross with us too
But a rest on the cushions will usually do.
So thank you, thank you to Archie for teaching me so much,
Thanks for giving us your special touch.
I'll miss you.

Suzy explains how she has benefited from the experience. "I really want to be someone like Mrs. Hayward who looks after people with problems. Looking after Archie has made my decision even stronger." And her final words to Archie: "Thank you Archie for brightening up our school and always smiling. I'm really going to miss you when I move to my secondary school!"

These sentiments are clearly shared by Alex Peters (11): "I will miss him when I leave so I hope he enjoys having other people to look after him… I WILL MISS YOU ARCHIE. BYE BYE."

The children's comments clearly show that Archie is an accepted and well loved member of the school. He has settled in, and he has formed beautiful relationships. He's happy to be here, and everyone is happy to have him. ■

A parent's experience of

managing cataplexy and mild epilepsy

Karen Otway, mother of Emily (MPS III), was invited to speak at the MPS Conference in June 2005. Here she relates her family's experiences.

We are the Otway family, my husband Patrick, daughter Emily (9) affectionately called Ellie, son Sam (7) and Lucy (20 months).

On 9 July 1996, our eldest daughter Emily was born five weeks early following a difficult pregnancy and after a complex delivery. She spent several weeks in Special Care. What followed was what I now think of as the 'honeymoon' period, oblivious of all that was to come.

As a baby Emily was a cheeky, blonde ball of fun and extremely easy to care for but we were aware that Emily's development was slow and that all was not well. In March 2000, after what seemed an eternity of tests and investigations, a diagnosis of Sanfilippo Disease, MPS III, Type A came and subsequently our lives changed forever.

Ellie has been lucky so far and on the whole has remained in good health and, until this year, fairly calm spirits. Although if any of you have had the pleasure of meeting her at the conference weekend, the noise levels have notched up a fair few decibels!

It has also been during this last somewhat turbulent year that we have noticed a progression in Ellie's seizure activity and the emergence of what we now know to be called cataplexy.

During last summer I noticed that often, when I was feeding Ellie, she would giggle. It wasn't her usual laugh, quite often out of the blue and almost inappropriately her eyes would flicker and appear to roll back into her head and her face would sometimes twitch. I would be unable to feed her as her tongue would fall forward but this would only last seconds and then she would stop and recover instantly and carry on eating. This was quite different to her other seizures and would leave us confused as to whether it was a 'fit' or not.

If it happened when Ellie was mobile or if I was changing her standing up, she would giggle just as before, but became like a 'rag doll' with almost no muscle control at all. She would stop laughing for a few seconds and regain some control but, if she should laugh instantly, she would collapse again which made it very difficult to move her. All we could do was support her and try to remove the stimulus that was making her laugh, which usually involved her 7 year old brother pulling funny faces, or worse!

This went on for several months until I read an article by Dr Ed Wraith on cataplexy in Sanfilippo Disease and following an appointment with him we had our diagnosis. This condition is often confused with epilepsy but is in fact a neurological condition causing loss of muscle tone in response to laughter, stress or a frightening experience. However, the main difference that we have found is that the recovery is instant.

At almost the same time we noticed Ellie's absence seizures starting to last longer where she would stare, not blinking at all, for maybe only a minute and sometimes she would drool. What was more noticeable at this time was that Ellie's recovery was extremely slow. She would be dazed and difficult to feed or engage in any kind of communication, and this could last for several hours.

A PARENT'S PERSPECTIVE

On an increasing number of occasions Ellie had started to have drop seizures. She often screams, again quite unusually, and then suddenly drops to the floor where we have noticed very slight twitching of her limbs and on these occasions Ellie will stay there for up to 15-30 minutes asleep before recovering. When this happens complete recovery can take anything between 12-24 hours.

So, what have we done about all this?

We felt Ellie was becoming quite vulnerable with all this varied and quite complex seizure activity so we decided to develop some information to accompany Ellie wherever she goes that explains the different seizures and cataplexy regarding signs, symptoms and treatment in order that we feel Ellie's safety can be maintained.

Along with this we have made obvious changes to Ellie's room in that we have had a padded bed and padded parts of the walls. We have beanbags on the floor and raised the height of the bottom part of her stable door so if she is leaning over when she has an episode she can't hurt her face or neck.

We have tried hard to obtain some sense of balance regarding Ellie's freedom and enjoyment of life and maintaining her safety. We did try a helmet and knee and elbow pads, but as most of you with Sanfilippo children will know, more injury was caused to innocent parties with Emily throwing these objects than it was a help to her.

Ellie remains happy in the most part despite all of this and although she is starting to show obvious signs of slowing down, retains her own determined little personality and therefore contributes immensely to our family life.

Dr Maureen Cleary talked to me very early on about living for today and most of all trying to focus on what Ellie can do now and not what she can't do or has lost because if you do you can miss some precious moments. We have tried to remember that in every aspect of Ellie's care – get the advice and treatment, make sure everyone knows who needs to know and then we get on with being a family.

I once read that some of the best novels ever written remain unfinished! We don't know how long we are to be blessed with Ellie in our lives but I hope that an account of her life would be one that you couldn't put down. ■

Living with Sanfilippo

How has it affected our lives?

Peter Hawkins was invited to speak at the MPS Conference in June 2005. This is a transcript of his speech.

My name is Peter Hawkins. My wife, Jean, is not here today because she is preparing for our middle daughter's wedding on Friday. We have four children, three girls, aged 34, 32 and 28. Our eldest daughter, Tracy, is a carrier of Sanfilippo Disease but fortunately her daughter is not affected by MPS. Our fourth child is Andrew. He has Sanfilippo Type A, is 22 years old and still walks and talks, albeit with limited vocabulary.

My brief today was 'Living with Sanfilippo: How has it affected our lives?' but more likely, 'How hasn't it'. Andrew was born on 5 March 1983 very quickly at the local Wokingham Maternity Hospital. Only one midwife was on duty as the rest were off sick. Our initial joy of having a boy was curtailed when I noticed he had breathing difficulties. The midwife checked and said he was ok and then promptly also went off sick! This was ominous but there was worse to come… The doctor checked Andrew and said he was ok. He was wrong. Andrew was rushed to Reading Hospital that night with a collapsed lung. He spent nine days in intensive care after which time, although jaundiced, he had recovered sufficiently to come home. Andrew then decided that for the next nine months there was something he could do without – sleep!

By the age of two Andrew had an enlarged stomach and head. He was somewhat backward. Andrew went to playschool and then on to school where he caused chaos (along with another pupil called Victoria who I shall mention later). After 15 months he went to a special needs school. This was fine at first until they changed their policy on 'problem children' and Andrew ended up with a broken arm.

We took Andrew to a procession of doctors. We regularly saw our paediatrician who measured his stomach, his head, and then she measured my head! One day she surprisingly asked my wife 'Why do you want to know what's wrong with him?' Although she later apologised, this question was sufficient to seriously shake Jean's faith.

By 8 or 9 Andrew's behaviour was sometimes violent and aggressive. He floored me on two or three occasions by unexpectedly hurling a plastic bucket into the small of my back. He threw a garden hose at me – at the time he was in the garden and I was in the house and in between us was the patio door, which amazingly didn't break. In shops Andrew would crawl under clothes racks and in crowds he would push doors into people (not funny when they are over six foot tall).

Andrew has always required toileting and has a very limited diet. He lives mainly on Edam cheese, sausages, chips, beans and toast.

When he was 10 I went to the local GP and asked 'What's wrong with him?' She didn't know and asked for any suggestions. I mentioned about testing him for food allergies. I paid £100 to find out he was allergic to nearly everything! Another doctor had noticed a chromosome abnormality and for two years this was a complete red herring.

We eventually took Andrew to a doctor, Dr Pike, in Oxford. He followed a hunch and carried out a few tests. We received the results at Reading Hospital. On arrival we were confronted with a young female doctor sat between two men, one stood on either side of her. There was Jean, Tracy, Andrew and me. The doctor said 'We think he should live into his twenties'. Jean visibly shook as I stuttered 'What did you say?' 'Oh yes, it's incurable, it's called Sanfilippo. Talk to so-and-so, they'll explain'.

We were subsequently put in touch with the MPS Society as we slowly came to terms with reality. After Andrew's broken arm, with the Society's valuable support, Andrew was transferred to a special needs school in Surrey with a personal 1:1 support.

Andrew has created several embarrassing moments. Andrew loves planes, unfortunately expensive 'Virgin' planes. You can't take Andrew on a crowded beach but the sun is apparently helpful to the brain function in his condition. We break the bank and try to go to Florida (Gulf Coast beaches are not over-populated). Whilst there some years ago, we went to a local restaurant. One of Andrew's traits is that if anything happens to him, regardless of the cause, it's always 'Your fault'. Sitting next to him at the table, I must have caught his arm or leg. Without warning, he punched me right on the nose! In the restaurant everyone just stopped eating and stared. You could hear

a pin drop. Jean grabbed Andrew and left. In a muffled voice, whilst holding my nose, I asked the waiter for the bill and everyone else resumed eating and talking again!

Andrew is very stubborn. He was about nine years old (mental age 2-4 years) when we went to Studland Bay in Dorset. We left his mother sat on the beach and went to the far end of the beach to explore rock pools which are set on the shoreline in rocks well above the level of the sand. After some time, I said to Andrew that we should be getting back to mummy and he replied 'No.' I started to return, hoping that he would follow. Instead, he came up behind me and pulled down my shorts to my ankles. As a result, I am stood naked, almost on a stage, knowing that thousands of eyes are focused on me! I stared into space, pulled up my shorts, started walking and, thankfully, Andrew followed.

I regret to say that some friends and family have been unable to allow for his problems. Although he was still small, due to a friend blaming him rather than trying to understand him, we ended up moving home. Again, when he was small, my mother inferred that Andrew's problems were due to his mother, Jean. As a consequence, she has not met my mother face to face for fifteen years!

Following an MPS Conference, I was enthused to explain Andrew's problems to people. When in our local chip shop with Andrew, he brushed against a man in the queue. I apologised by saying 'Sorry, he's special needs'. 'What's wrong?' came the reply. I said 'It's terminal. He's a Sanfilippo'. The man asked 'How did he get that?' and I explained that it was a genetic condition. He replied 'Well, then it's your fault isn't it?' I found myself passing my chips back to the girl, whom I knew, behind the counter and moving swiftly towards him. He bolted at the last moment.

Inadvertantly, I realised that day that I had learnt a valuable lesson – some people cannot handle the embarrassment of their lack of compassion.

Regrettably, many friends fade away but some friends and family do not. Out of the blue, Victoria, the girl who caused havoc with Andrew when he first went to school arranged a service at the local church in his honour. She found out the truth about Andrew when, after diagnosis, he made the headlines in the local paper. The church was full, not only with his old school classmates, but many others, few of whom we knew. It was a very humbling experience.

My eldest daughter, Tracy, her husband and two friends, ran to the top of each of the three highest mountain peaks in England, Scotland and Wales in 24 hours. They raised over £3,000 for MPS. My soon-to-be son in law just ran in the London Marathon raising over £6,000 for MPS. This was despite requiring hospital treatment just two weeks before the race and being told not to compete!

When Andrew was sixteen, I took the opportunity to take early retirement and move to Dorset. Andrew changed schools and again got 1:1 support, although not as good

Andrew

as before. He now goes to an adult day centre where he was supposed to get 1:1 support. I've constantly queried this and have now finally had confirmation that the council have been deceiving us. A current ongoing fight which is being fought with the support of MPS.

Andrew has certain characteristics which have to be catered for. He is difficult to wake and can be aggressive. Each morning I wake him for Homecare. I use 'Sooty'. He'll punch me but he won't hit Sooty. I also do a poor ventriloquist act for Sooty. I've done it for three hours on a beach on the Great Barrier Reef. I did it to amuse Andrew while my wife fulfilled her life-long ambition to swim on the Reef. I spoke to Sooty on his 'shell phone'. I've also done this with Sooty on an aeroplane but, unfortunately, lots of other kids appeared before long!

Andrew can also be funny. Once when we were driving along in the pouring rain Andrew shouted out from the back seat 'Abandon Ship!' His own words for flooding are 'It's flugged'. Great for a different computer password!

We try to amuse Andrew as best we can. We drive him here and there to get him out of the house. He likes cups of tea, sausages or cheese sandwiches, but taking him to visit people is a problem as he constantly needs the toilet. You have to ensure you take a urine bottle and place it on the toilet seat in case he gets there before you. If there is no bottle, Andrew will hose down the walls and will then proceed to strip naked!

Andrew's obsessional behaviour is getting worse. He constantly picks up bits whilst ripping his clothes to pieces. In bed we often have to put socks on him as he sometimes rips his toenails half off.

The future will be a problem. As we age, his condition will worsen. Jean and I already have problems with slipped discs. I did mine lifting Andrew when he fell several years ago. Somehow we must persevere and I am sure that we will.

Andrew

Oh Andrew, oh Andrew
When you were born that day
We wondered if you'd make it
Whether you'd stay

But stay you did
And life's never been the same
It's since been a real roller coaster
With you in the frame

From nine months without sleep
To then watching you grow
One minute a treasure
The next a right 'so and so'

From sunshine to rain
With the immortal words 'Abandon ship'
You've made your mark
In life's hazardous trip

You've given us great joy
In each and every day
Life could never be the same
Whatever people might say

You're unique, you're a one off
A sight to behold
You're a fighter, then a comedian
A real pot of gold!

So despite Ol' Sanfilippo
Who gave you a name
Despite that label
You've proved to be game

You're proved you're a person
An individual by right
And long will you persevere
To fly your kite

Let's face it in life
No one gets out alive
And as long as you continue
We will all strive

To love and treasure you
And all that you do
You're our son and we thank you
For being Andrew

Peter Hawkins (Andrew's father, see page 20) sometimes writes rhymes. Here are two he wanted to share with MPS readers...

A Tribute

By Peter Hawkins

From her own tragedy in losing a son
Was born a cause that she had begun

She found there were many with similar woes
No help, no guidance, to help deal with the blows

A state so uncaring, so bureaucratic and trite
It needed someone not afraid to put up a fight

Someone who'd challenge and not accept the word no
Someone who'd fight them blow for blow

And so she began her life-long quest
Reaching out to the world, putting the system to the test

Seeking recognition of the problems faced by so many
Giving help and support when before there wasn't any

Now there's doctors, carers and a Society for all
Where once there was nothing, now they're on call

Without her what would happen
Where would we all be?

Now finally honoured, she deserves it
Christine Lavery, MBE

If you have a story, information or a photo that you would like to appear in the MPS Magazine contact us now on
0845 389 9901
or email
newsletter@mpssociety.co.uk

10 WISHES FOR JEANS FOR GENES CHILDREN

Jeans for Genes has teamed up with the Starlight Children's Foundation for its 10th Anniversary Year 2005 and has granted ten wishes to Jeans for Genes children.

Back in January the MPS Society was asked to nominate two children and invited members to apply for a wish on behalf of their child or children suffering from MPS, Fabry or a related disease. The children had to be aged 4 – 18 years, indicate the wish themselves and not have received a wish from another wish-granting organisation in the past. The child and family were also required to participate in media coverage of the wish. Over 40 nominations were received from MPS members and we are sorry it is only now that we can tell who was successful.

Congratulations to **Oliver Moody** aged 10 years who has Maroteaux Lamy disease (MPS VI) and lives in Yorkshire and **Roma Drayne** aged 12 years from Northern Ireland who has Morquio disease (MPS IV). They have both been chosen to receive a Jeans for Genes 10th Birthday Wish.

As I write Oliver is in Milan, Italy, visiting the Ferarri factory that builds the Formula One racing cars and there may be other surprises in store. We hope to have Oliver's Jeans for Genes wish story for the next newsletter. We will also have to wait until the next newsletter for news of Roma's wish as she hasn't had it yet.

All children meeting the criteria and wanting a wish granted can apply to the Starlight Children's Foundation and other wish giving organisations. You can read more about the Starlight Foundation on page 29 of this magazine. Here are some other wish-granting organisations you may like to approach:

Dial a Dream
Tel: 020 8530 5589
www.dial-a-dream.co.uk

Happy Days Charity
01582 755998
www.happydayscharity.org

When You Wish Upon a Star
Tel: 0115 979 1720
www.whenyouwishuponastar.org.uk

Make a Wish Foundation
01276 24127
www.make-a-wish.org.uk

10th Birthday 2005

For more information about Jeans for Genes visit **www.jeansforgenes.com** and check out the MPS Fundraising Newsletter.

'Sanfilippo Angel'

Louise Lewis, mother of Georgia, MPS III, gives a personal account of Georgia's diagnosis and its effect on their family.

Driving home, the rain pounding relentlessly on the windscreen only enhanced the sombre mood inside the car. I felt numb, sick and angry all at the same time, a roller-coaster of emotions dominating my entire body.

Just beside me, my husband was driving, staring, fixated on the windscreen. We couldn't speak, just embroiled in our own living hell, the outside world passing by in a blur. I wanted to reach out and help him, but I felt paralysed and helpless, not even able to help myself. Only 15 minutes earlier, we'd been sat in the consultant's room and told that our little girl was going to die. As simple as that, and there was nothing we could do about it. Nothing, except to watch and wait and just ensure that she is comfortable and not in pain. Feeling out of control and wanting to die, this was a living nightmare. This surely must be the worst day of my life.

However, the worst was yet to come – I had to tell my parents that they were going to lose their grand-daughter, I had to tell my sisters and friends. Looking back now, I was on 'auto pilot', I would calmly tell friends and family that my little girl was dying, while people around me would be distraught with grief. However, I coped and quite well, I thought too, I didn't cry for weeks on end, it was all very surreal, I just knew I had to get on with it and tell everyone who needed to know.

Prior to this appointment, there was a lot of history, involving endless hospital and clinic visits, all kinds of professional people involved, GPs, consultants, psychologists, all doing their best to find out what was wrong with our little girl. Of course, we knew something was wrong, but nobody knew exactly what, which was extremely frustrating.

Georgia was born on 11 July 1995, by Caesarian section, due to her being in a breech position, weighing a healthy 8lb 2oz. In the early stages of pregnancy, a test for Down's Syndrome had concluded that my baby had a 1 in 54 chance of having the disease. We were told that this was extremely high, and after much thought and deliberation, decided to go ahead with an amniocentesis to get an exact result. Whilst waiting for the results, our life was on hold, in a state of limbo and after much soul-searching we decided not to go ahead with the pregnancy if the result was positive. Luckily, the result was negative and we were to have a healthy baby girl. We were ecstatic and both sat crying in the waiting room, clinging to each other, so relieved that our anguish was over. Looking back now, with hindsight, it hadn't even begun…

From a baby to toddler stage, Georgia was considered 'normal' and did everything as she should've done and reached the essential milestones. I already had a son, so I knew what to expect. Our concerns with Georgia began when she was approximately two years of age. She wasn't talking as well as my friends' children, she was terribly frightened of people and was extremely clingy. This was often put down to the fact that she may be very shy and she had an older brother, who would do the talking for her! Our concerns grew and so did the endless appointments and tests. For three years this went on, with nothing conclusive and during this time, Georgia was deteriorating. I could write a book about our life and experiences during this difficult time.

In time, a Consultant at the University Hospital of Wales, who had seen Georgia on previous occasions, wanted to carry out further tests, which would involve a skin biopsy under general anaesthetic. We had to wait six weeks for the result, as it involved growing skin cells to obtain her DNA. This was a traumatic time for us, as it was during Christmas 2000 and we had decided not to tell our family, but to persevere and enjoy Christmas for everybody's sake. It would've been pointless to spoil this time for everybody. Waiting for the results of the test seemed like an eternity and all this time, I knew deep down that something was seriously wrong. I was right. All the uncertainty ended on that fateful day on 7 January 2001. It was the new millennium, the start of a new year, but our world came crashing down upon us.

Georgia was diagnosed with Sanfilippo A. This is an extremely rare genetic disease, one of a group of diseases called Mucopolysaccharide, or MPS. Mucopolysaccharides are long chains of sugar molecules which are used in building connective tissue. Typically, when the body is finished using these sugar molecules, it breaks them down with enzymes and disposes of them. Children with Sanfilippo Disease, however, are missing the enzyme to breakdown the molecules and instead store them in cells in their body. The storage of these molecules then causes progressive damage. Babies show little sign of the disorder, but as more and more cells become damaged, symptoms start to appear.

MPS III

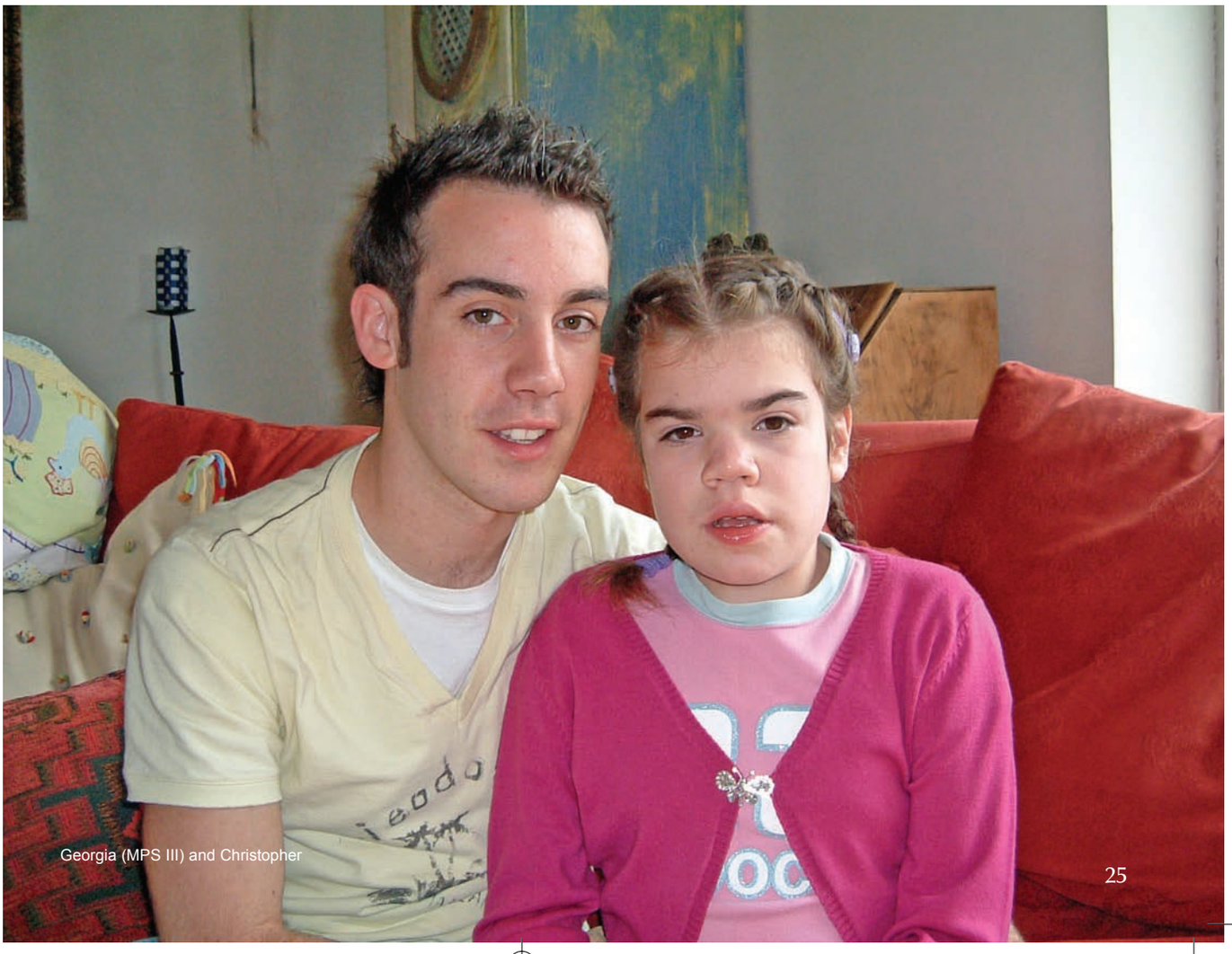
Finally, after many years, we knew what we were dealing with. In a strange way, it was such a relief, as not knowing what was wrong is far worse. Generally, there are three stages of Sanfilippo progression. In stage 1, it is noticed that the child starts to lag behind and develops difficult behaviour. In stage 2, the child may become extremely active, restless, and often have very difficult behaviour. Some children have sleep disturbances. Many like to chew on their hands, clothes, or other accessible items. Language and understanding will gradually become lost. Some children never become toilet trained and those who do eventually lose the ability. In stage 3, the child will begin to slow down. They have more difficulty when walking or running and fall often, eventually losing the ability to walk altogether.

Sanfilippo is a terminal disease that has no cure, and Georgia is not expected to reach adulthood. I know of one family, whose daughter had Sanfilippo and died when she was 11 years old.

Georgia is now 10 years old and she is now in stage 3. She has lost all her speech and has really slowed down. She can no longer run and although she is still mobile, she is very unsteady on her feet and cannot walk far as she gets too tired. Also, she has lost many other functions, for example, feeding herself and in time may need to have a gastrostomy as she will be unable to swallow. However, she is beautiful, adorable, funny and extremely happy. We know if there is something wrong with her, as she stops laughing. To hear her laughing is such a joy and something that I cannot imagine being without.

Throughout all of this, people ask how we cope. To be honest, I really don't know, but the strength comes from within and from the support of family and friends. However, the most important thing in our lives is Georgia. She is our angel, our shining light. She is our inspiration and she has made us realise what is important in life, and now we just have to treasure her for the time that we have left with her. ■

If you need support after receiving a diagnosis please phone MPS on **0845 389 9901**



Georgia (MPS III) and Christopher

JESSICA AND TERRI HAMBLY – A SUCCESS STORY!

Paul Hambly tells us about his two daughters, Jessica and Terri, both of whom suffer from MPS IHS. Following on from the Summer 2005 MPS Magazine, in which we featured an article about the Hambly's which appeared in the Hull Daily Mail, their father Paul explains what has happened since.

We have finally moved to Hull Royal Infirmary for our infusions. The nurses and staff are very nice, the girls have a room all for themselves and the day is a lot shorter and easier for them. We start the infusion at 12.30 and are finished around 5 – 5.30pm. Once we've been for a few months, they are going to start the infusion a little earlier.

We have also moved to a large four-bedroom house, the downstairs is open plan and so has lots of room for the girls to run about.

I read in the last magazine about Dawn McDougall and the problems she had getting adequate accommodation for her two boys.

Helen and myself had similar problems when we tried to get the house we are in now. The only adaptation it has is a through floor lift and a walk-in shower. The engineer who came to service the lift said that he didn't know they still made that type of model any more! When we agreed to take this house we were told it was either this or nothing, so we took it.

However, after last year's Jeans for Genes campaign when we had pictures on the front page of the Hull Daily Mail, things have certainly changed for the better.

Jessica and Terri Hambly
Photo appears courtesy of Hull Daily Mail Publications Ltd.

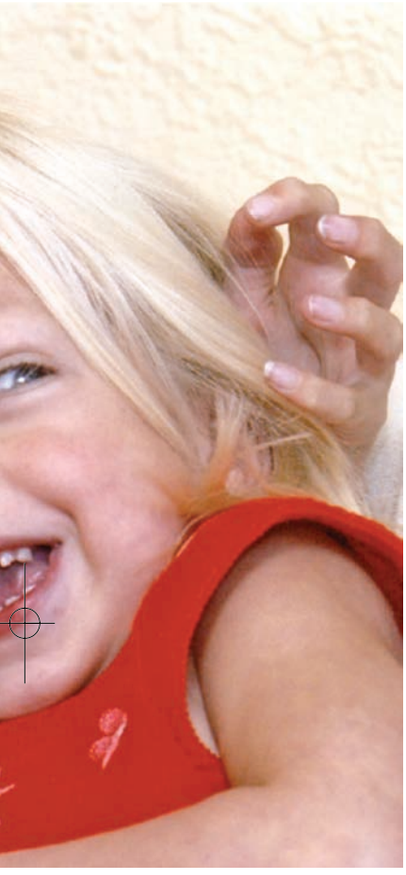
Armed with letters of support from the MPS Society and Dr Wraith and a copy of the Hull Daily Mail, we went with our Family Support Worker to see our Housing Manager. Within one week we were offered, and accepted, a four-bedroom house with a stair lift. After having talked with our Occupational Therapist, the Housing Officer agreed to other modifications and improvements.

After only seven months we were able to move into the property in May this year. The Council and other services have changed the layout of the house to open plan, have fitted a brand new bathroom and shower, fitted a new kitchen and brand new fire doors. They have replaced the heating system to gas central heating (thanks to letters of support). The two gardens are enormous, they need a lot of work but have great potential and we have been told we can have ramps fitted.

Although no one has said anything to us, Helen and myself believe this all happened as fast as it did because of the publicity in our local paper. Even though it was not our intention to benefit personally from the Jeans for Genes article, it certainly seems to have happened. So, my advice to everyone who is thinking about doing something in the media for Jeans for Genes this year is definitely do it as, for once, everyone seemed to benefit from it. I know hundreds of people donated money to the collection in our local "Kwik-Save".

10th Birthday 2005

For more information about Jeans for Genes visit **www.jeansforgenes.com** and check out the MPS Fundraising Newsletter.



Housing and Adaptations

A Professional's Perspective
from the Community Team Learning Disabilities

If I were to provide a blow by blow account of the work we have undertaken to support a family in obtaining housing or adaptations it would be a book, not an article. We have in turn been disappointed and frustrated and at times angry. But rather than focus on the negatives, I feel it would be more fruitful to focus on the positives.

The particular family we have supported are amazing themselves but they have also been amazingly patient and understanding. All the health professionals in both Children and Adult Services and the staff in the voluntary sector who have contact with the family are totally committed to giving them the best support we can. If we could build or adapt a house we would, if we could give someone else money to build or adapt a house we would, but we don't have the skills or the money.

Along with the family we have been trying to ensure that they are seen as a priority by those that can build or adapt a house. As with everything, there are also a lot of other people who think they are a priority. Who would want to sit in judgement when money and provision are limited?

In addition, some adaptations and needs are easier to meet than others and can be achieved quite quickly and at minimal cost. The needs of our family are in our opinion unique and their requirements cannot be met easily or cheaply. Additionally, people did not recognise or understand the disease, why would they? We had to educate as well as explain the requirements.

Bearing this in mind we knew we would need to go to the top of the tree and approach Heads of Services. We also recognised that others may be able to open doors that would be closed to us. To this end our team and the Specialist Community Nurse from the Family Support Team set up meetings, wrote letters, made constant phone calls. We also, with the family, got others to do the same, local politicians, the Community Health Council, the Local Health Board and many others all wrote letters of support.

The MPS Society and Clare Cogan specifically helped support the family, attended a meeting and wrote a letter to the Heads of Service with information and prognosis of needs including an example of a building plan.

The support and commitment of the team manager is essential, as the case is so complex, that other work has to be reprioritised in order that attention can be maintained on the case. My manager also acted as back up in sharing the case, to avoid loss of continuity in case of sickness or annual leave. Clinical supervision and good communication meant that we kept each other up to date. Time can pass so quickly, we could not afford to lose time by taking our eye off the ball.

All meetings resulted in action plans with clear objectives, accountability and timescales. As case co-ordinator making sure that these plans were in place and adhered to meant that everyone was clear of their role and responsibilities. As we also worked alongside Children Services, co-ordination and teamwork was essential.

To any professional or carer beginning this process my advice is to get support from the MPS Society as early as possible. Build up a network of support who are willing to make phone calls and write letters. Keep a record of all attempts to make contact, these were useful when we needed to apply more pressure. Finally make sure you have a really tenacious attitude. Without it we would still be waiting for someone to respond to our phone call. We were determined that we weren't going to go away or give up easily. It has not been an easy process but eight months from when we started, we now have the attention and hopefully the understanding of the people who can make a difference.

If you would like help with an housing issue contact the Advocacy Team at **advocacy@mpssociety.co.uk** or phone **0845 389 9901**.

A dream come true

Ryan Seeber, MPS II, had a day to remember when the Starlight Children's Foundation made it possible for him to spend a day fishing with his hero, John Wilson. Bob Seeber, Ryan's father, tells us about Ryan's dream come true.

Left to right: John Wilson, Ryan (MPS II) and Bob Seeber

My son, Ryan, was six in April this year but it was just after his third birthday that he was diagnosed with Hunter Syndrome, a degenerative illness affecting mobility and learning abilities. Part of the illness means that Ryan has a shorter attention span than most except when it comes to fishing with me or watching his hero, John Wilson, on the 'Go Fishing' programme on television.

When we heard about the 'Starlight Children's Foundation' we realised that there was a possibility that Ryan could have a day fishing with John Wilson and this was his number one wish of his three wishes when we applied towards the end of last year. There was a prompt response from the Foundation who then contacted John. His response was that he was generally very busy but would be more than happy to make time to see Ryan. He did, however, suggest that we waited until the following summer when fishing would be better.

Final arrangements were made at the beginning of June to visit John at his house in Norfolk on 8 July 2005. Accommodation was for two nights at a country hotel about a mile from John's house so we could get there the day before in our own time.

On the morning of the memorable day we rang John to get directions to his lake and as we drew up he appeared complete with a tray of fresh coffee having walked down the short distance from his house.

Ryan, for once, was absolutely lost for words as all he had known was that he was going fishing. After several silent moments, from Ryan that is, he managed to utter the words 'John Wilson' in total awe and disbelief.

From then on it was all go. John had already set up a couple of rods. One was to fish for carp and the other was for smaller fish such as roach and rudd. He had baited up a couple of areas the day before so it didn't take long before Ryan started to bring in a number of fish caught on sweetcorn.

We all had fun and Ryan's mum, Rachel, took plenty of photographs and some video footage. One of the pictures even ended up in the Sunday Express in John's weekly angling column.

After we had an enjoyable lunch in the local pub, with John keeping us all entertained, we returned to catch some more smaller fish up to about 10oz. After about an hour Ryan had begun to tire and we had just about given up on a bigger fish when the carp rod bent over and the line started peeling off the reel at a fast rate of knots as a carp picked up the bait and was hooked.

John carefully controlled the fish out of the lilies before handing the rod to Ryan whilst John landed a stunning carp of about 11lb. What a great end to the day! John was marvellous throughout and very understanding of Ryan's behaviour. The only problem now is that Ryan expects John to be there wherever we go fishing!

As a postscript we also met up with John at the Game Fair where Ryan gave him a birthday card and Ryan had John's latest book signed. John also wrote an article in the Sunday Express a few weeks later and sent us a lovely letter and some of his photographs to say how much he had enjoyed the trip.

This was one of the most memorable days for Ryan, and probably for me too, all thanks to the Starlight Foundation and the kindness of John Wilson.

Joyful Ryan is hooked

I must admit, following recent events, to feel extremely elated. Not from what I caught, but from what six year old Ryan Seeber from Harrogate managed to extract from our two-lake fishery in the course of an hour after lunch at the local hostelry. The day was arranged some time back through the Starlight Children's Foundation for young Ryan who sadly has a very rare disorder called Hunter Syndrome.

Because he is always watching my programmes on TV, his parents Bob and Rachel thought he would like to meet me. The pleasure, as is so often the case, turned out to be entirely mine. There is immeasurable joy to be gleaned from studying a child's face as he or she marvels at the silver and gold flanks of small roach and rudd, which are ridiculously prolific in my lakes.

Within minutes of the family arriving, young Ryan was hauling them out one after another on float-fished sweetcorn, with his Dad helping to steady the rod. I rummaged through the garage especially to find this particular rod because I made it purposefully, several years ago, for my grand-daughter from 6-wt fly rod blanks. Being super lightweight with a forgiving, all-through action, it means they are easy to control the float rig and strike effectively. Ryan really enjoyed catching roach and rudd between two and ten ounces.

On account of Ryan continually mentioning 'carp' I put out a heavier 'sleeper' rod with an 18mm boilie hair rigged on the business end, close to some lilies. Within half an hour of returning from lunch and casting it out again, the reel started to sing like a canary. Quickly I lifted the rod and put Ryan's right hand around the handle while he cranked away furiously with his left. The result was a lively, bronze-backed common carp pushing double figures. Ryan, understandably, became hyper-excited and was almost lost for words.

What a truly lovely, memorable day – and I didn't catch a thing!

This article, written by John Wilson, appeared in the Sunday Express, 7 August 2005.

STARLIGHT

Brightening the lives of seriously and terminally ill children

Starlight Children's Foundation brightens the lives of seriously and terminally ill children by granting wishes and by providing entertainment in hospitals throughout the United Kingdom. All of Starlight's activities are aimed at distracting children from the fear, pain and isolation they can often feel as a result of their illness.

Starlight was founded 18 years ago and in its first year helped just four children. Starlight now helps over 400,000 children every year, whatever their illness, and this number continues to grow.

Starlight's aim is to grant a wish to every child diagnosed with a serious or terminal illness in the UK and to provide entertainment for all children whilst they are in hospital.

Starlight Wishes

A Starlight wish is more than just a special treat to look forward to - it gives a child a wonderful choice to make all by themselves at a time when most decisions need to be made for them. A Starlight wish also remains a positive focus throughout long periods of treatment. Starlight always involves the whole family in the wish to help strengthen family bonds and to give everyone happy memories to share.

Starlight Wishes are limited only by a child's imagination, from swimming with dolphins to going in a helicopter; from meeting a favourite sports team to recording a song…

Anyone can refer a child for a wish, simply telephone **020 7262 2881**, email wish@starlight.org.uk or visit **www.starlight.org.uk** for more information.

The Children's Hospice Association in Germany

Elizabeth Volk, Founder Member of the Children's Hospice Association in Germany, gives a personal account of the Association's development. Part two of this article will appear in the Winter MPS Magazine.

Presently there are five children's hospice houses in existence in Germany, with two more in the planning. As children only spend a maximum of four weeks per year in these houses, our German Children's Hospice Society has recently branched out to create mobile services in order to go out into the homes of the families to stand by them during those many many remaining weeks of the year. Already these services have been established in five different locations and more are in the planning. Ultimately, the goal is that each and every family in Germany will be able to profit from these services in the areas they live in. This project has just been launched and there is still a long way to go to reach that goal.

But in addition to the financial aid we extend to all of the existing houses and the mammoth project of the mobile services, our Society also offers a whole range of seminars to meet the various needs of our families. We offer separate seminars for the sick children, the healthy siblings, parents and partners, grandparents, and the entire families. We offer fun weekends, canoe trips and camps for the siblings, as well as canoe trips for the fathers. These seminars are popular, and illustrate the need to relax and have fun in an otherwise often fairly stressful life. After the death of a child, we offer grief seminars which families may attend for as long as they may care to. Very significant are our seminars for the professional care-givers, be they the teachers in the schools our children attend, hospice personnel, or hospice volunteers.

This autumn we will be hosting the first national "Children's Hospice Days" in Cologne, with seminars and workshops for both professionals and parents. We hope for a huge turnout. The Society is more active than ever. We've come a long way but it seems like there is impossibly much yet to be done. This year our Society just celebrated 15 years in existence.

On a much more personal level I recently reduced my volunteer work with the Children's Hospice Society considerably, and so it seems like a good time to take stock and try to convey a very personal account of what the children's hospice has meant in our lives over the past 15 years. I would like to mention not only our own experiences, but also touch on the very human issues at the core of how our hospice work has developed. Because our family history with Children's Hospice goes back to its conception in Germany, certain links to the history of the development of hospice thought often go hand-in-hand here.

Although our twins Hendrik and Emily have since died, I think back on the day of their diagnosis as the worst day

Elizabeth Volk stands to the right of Christine Lavery. This photo was taken at the International Working Party on MPS Diseases, Frambu, Norway and the article is taken from a speech given by Elizabeth Volk.

of our lives. It was the day that the paths of our lives careened in a direction diametrically different from everyone else we had known until that time. We mourned for the futures of our beautiful children, the lives we would not be living. When I think back to that day, I find it remarkable that I can say and believe that that particular day was the worst of our lives, and NOT the days the children died, six weeks apart from each other, over 14 years later. I sense that this has been largely the result of our growth with Children's Hospice which makes this so. I am aware that this must not necessarily be the case for everyone who uses our services, but often others tell me the same thing about their own perceptions. It seems to be a thing to marvel at - that living with the imminent deaths of one's own children and gradually coming to face a lot of uncomfortable things head-on over the years can give you strength, and even lighten the load. Just imagine!

As the Children's Hospice Society slowly gained momentum and members, we noticed that our needs were very immediate, though we were not yet able to build the first house. Mostly in the beginning, we were just a handful of MPS parents getting to know each other better and better. Seminars were offered and we advertised among the other self-help groups and new families began to come to further seminars.

Despite other diseases being so different, we often found we had much in common with these families. As is the case with all other self-help groups, one of the goals of our Hospice Society is to put people in contact with one another, to give families a chance to help themselves and to help each other at the same time. I would still put a premium on this valuable aspect, and of course, this is paramount to the MPS groups as well.

After the Society was founded, it was inevitable that sooner or later those first children were going to die. When this did actually happen, for some of us it was a startling recognition that the children really do die after all. Bear in mind, we were young families at the time and these were the early years, around 1990. For others, it was a recognition that we needed each other and a comfort to be part of a larger group. For all of us, it was a knowledge that death was just going to be a topic we were going to have to take a closer look at.

Thinking back on those first unforgettable children who died before we got the house going, I realise just how much our lives, but also our attitudes have changed and our hearts have grown since then. Our sick children did not come to this earth with the sole purpose of teaching us a lesson, but it doesn't hurt those of us who go part of the journey with them to try to learn something along the way.

There have been a lot of funerals over these past years. Although it certainly does not get easier, or make any more sense in most respects, still something has happened for me personally. With each child who has died, each story I have heard, each funeral attended, more and more of my own initial fears of death have been allayed. Each time a child dies there seems to be some lesson which may be learned from the magnanimous example of this young life.

One truly monumental aspect of what our Society has now come to consider a focus almost seems too banal to mention these days. Probably all our groups focus on this in the meantime, but still I want to emphasize that the consciousness of the healthy siblings had to first be awakened. These are the adults of the future, who often yet as children ultimately stand at the graveside of a brother or sister beside their parents. An awareness of what we on the outside can do to stand by these children had first to grow. A perfect stranger can do nothing, of course, but generally these days in our Society, the children know and trust others who can step in and try to support the structure of the parents involved with their own grief while the child or children stand beside them, often overwhelmed. I wouldn't say that it can't happen that a child stands completely alone with no support at the funeral of a sibling, but in our Society, it certainly doesn't need to be that way anymore.

For years now, we have also considered the healthy siblings a priority of our Children's Hospice Society. It is a major and continuing aim that our parents be sensitised to the fact that while the one child is involved in living out his short life, the other children are involved in the vital processes of their own childhoods which will shape them into the adults they will hopefully someday become.

We offer seminars for our siblings to simply get together and play. But there are also adults there who kids can talk to who understand, who the kids can open up to without fearing they are betraying their parents, and who will not betray their confidences to their parents. I will always be grateful that our children had people who stood beside them and behind them to strengthen their backs. Have you ever read the book, Winnie the Pooh? Listen to this:
Piglet sidled up to Pooh from behind. "Pooh!" he whispered.
"Yes, Piglet?"
"Nothing" said Piglet, taking Pooh's paw. "I just wanted to be sure of you." (A.A. Milne)

Wouldn't it be nice if we could be assured that each person we know had someone they could be sure of?

Over the past fifteen years of our existence the sensitivity to what our families need has grown considerably. Individual families who find themselves with an immediate financial concern related to the child can approach us for help, for example. The costs for the stays at our Children's Hospices are anyway borne by the health insurances and other state resources, supplemented if need be by funds from our Society, and we make sure that costs for attending seminars should not necessarily be a hindering factor, especially for our larger families with many children.

Recently the need has been felt for the Children's Hospice Society to clarify the terms we use to describe our work, to make our needs and concerns known as precisely as possible so that we parents can be as well represented as possible. In a time when law-makers cast more and more glances at the exploding costs in the health sector, it is imperative that we make known in no uncertain terms what we are about. ■

Polish MPS Conference

Barry Wilson, Chairman of Trustees at the MPS Society and his wife, Barbara, recently represented the UK MPS Society at the Polish MPS Conference. This is how it went...

Well, for reasons far too numerous to mention, we took Christine Lavery's place and represented the UK MPS Society at the Polish MPS Society's family weekend. You would think after spending many years in the Royal Navy I would have learnt not to volunteer!

My wife, Barbara, and I set off on the morning of Friday 8 July 2005, catching first a plane from Birmingham International Airport to Munich and then a second plane from Munich to Warsaw. I was quite confident at this stage because I knew Barbara could speak Polish. Actually, she felt that it had been so long since she last used the language, some twenty years or more, that she was concerned that she may not remember enough to communicate in a reasonable way.

As we emerged from the Arrivals Lounge in Warsaw airport, we spotted a young lady waving a large piece of paper with 'Wilson's' written on it. This was a good start and as it happens, an indication of how she would look after us for the rest of the weekend.

The young lady's name was Malgorzata Kaflik but she asked us to call her Megan. Megan and the husband of the President of the Polish MPS Society Sylwester Matulka, had been dispatched by Teresa (the President), to collect us from the airport. Megan asked, in excellent English, "would we mind waiting for a short while as they had one more guest to collect who would be arriving soon". About half an hour later Mrs Diana Sucbah from Croatia arrived and so off we set on our 30 kilometre journey to destinations unknown.

We drove through the city of Warsaw until gradually the tall buildings were replaced with smaller ones and eventually, just trees and green fields, as far as the eye could see. Actually, I may have missed some landmarks because during the journey, Diana started talking to us and as she would be the first to say, she could talk for Croatia. At last we turned into a lane that had a building at the end of it. This particular building was to be the scene of several very plain but nice meals that were offered to us during the weekend and it was one of many buildings that were spread over several hundred acres of land, sitting in a huge forest on the edge of a very large lake.

Now, bearing in mind that we had assumed the conference would take place in a Warsaw city centre, hopefully, air-conditioned hotel, the contents of our suitcase reflected that, and we were a little concerned, to say the least!

Sylwester kindly drove us about half a mile further on to our accommodation, which turned out to be a large three-story block of identical rooms, no air conditioning and the bare minimum of facilities. We were given our key, room 25 and off we walked up to the first floor to be confronted with a door that said room 25/26? Most confusing! One of our keys fitted the door and in we went, only to find four more doors, one having room 25 on it, another 26 and the other two doors turned out to be the shared toilet on the one side and the shared shower on the other. On entering our room, we found two small single beds with what turned out to be very firm mattresses, a coffee table, a desk with television on it, one chair and a very small built-in wardrobe, Oh, and two hand towels. We hadn't brought any towels with us! I mention all this because we had clearly come unprepared! Our suitcase contents did not match our accommodation. Ed Wraith was just down the corridor from us.

Upon arrival, we were immediately offered a simple supper and afterwards, tried to say hello to as many families and speakers as we could. The introductions included meeting the Polish MPS Society's President, Teresa Matulka. It turned out I had met Teresa on at least one previous occasion.

While we stood outside in the wonderful warm evening, talking to Teresa, Professor Grzegorz Wegrzyn (the Professor is currently involved in research which is sponsored by the UK MPS Society) walked by with members of TKT and Genzyme and asked if we would like to join them for drinks at a nearby hotel. After one small drink and some excellent conversation, Barbara and I decided to retire for the evening and walked back to our accommodation block.

The following morning, after a shower and 'drip drying', we went off to breakfast where once again were joined by Megan. We sat at the same table where we dined the previous evening. This seemed to be the correct thing to do. Megan gave us advice on the day's proceedings and asked if there was anything I needed for my presentation. I was looking forward to giving my presentation, particularly now that I knew there would be simultaneous translation in place. Not knowing if there would be translation facilities available before we left the UK, Barbara managed to contact a good friend of hers who has an uncle, who happens to be a priest, living in Poland who agreed to translate my speech. We were therefore able to take copies with us, which it turns out was greatly appreciated by the families.

I should explain at this point that the Polish families had already been on site for a week and most of them were there for two weeks. They were treating it as their annual holiday. In addition to enjoying the sunshine there were all sorts of workshops arranged to allow the families to see doctors, nurses, counsellors etc.

Well, the conference started with Teresa welcoming all the families, speakers and other guests. She explained the format of the next two days. Then without further ado, she introduced me as the first speaker. With a little nudge of encouragement from Barbara, I took the microphone and contrary to my normal delivery, I managed to speak a lot slower in the hope of giving the translator a fighting chance! The last word delivered, and I was pleased to say that the audience appeared to approve. The next few hours followed a similar pattern, with speakers from several parts of the globe passing on information directly and indirectly connected with MPS.

The meeting finally closed at 1700hrs, just in time we were told, to go for tea. During tea, still at the same table of course, we were asked by Megan if we could be ready and at the dinning room by 2030hrs for the gala dinner. We arrived in plenty of time and were invited to sit at the top table. Barbara and I felt honored that the Polish MPS Society had given us the same standing as such well known people as Dr Ed Wraith, Professor Grzegorz Wegrzyn, Dr Barbara Czartoryska, Professor Shunji Tomatsu and others. An indication of how well the UK MPS Society is thought of in other countries.

It was an excellent evening with lots of good food, great company, in spite of, in my case, the language barrier. All this, despite the fact that not a drop of alcohol was anywhere to be seen! Barbara told me that wouldn't do me any harm and would be "good for my soul"! Before we had been allowed to eat, the Polish MPS Society called various 'dignitaries' out to present them with a little gift. Our name turned out to be on the list and we were presented with a bone china, plate, cup and saucer with their Polish MPS logo on it. I replied with a few words of thanks and we were duly given the Polish appreciation, clapping in synchronism. Somewhat very tired and very hot, Barbara and I said our goodnights and went to bed to "recharge our batteries", ready for the next day's conference.

Although much smaller than our weekend conferences, the Polish conference format was very similar to ours. The main difference seemed to be in relation to their volunteers. Apparently they had started off the week with a full compliment but by the Saturday, most had realised just what hard work it was to cope with MPS children and had given up and gone home. This off course, meant that both parents couldn't attend the conference so one at least, had to look after the children. Some parents even brought their children into the conference.

More meals, more conversations with the families and still our shadow Megan was with us, ensuring we didn't get lost and were where we should be at the allotted times etc. Megan was an absolute angel and Barbara and I have decided to adopt her and join our other band of adopted daughters of whom we think the world.

The conference closed at approximately 1300hrs. We had lunch and then spent the next four hours saying goodbye to all the wonderful new friends we had made. With promises of communicating in the not too distant future with some of our new friends, Sylwester and our shadow Megan drove us back to Warsaw airport. Sylwester kindly took us via Warsaw city center, so that we could get a feel for the city. With real feelings of sorrow on having to leave our new friends, we walked off with our luggage to departures.

The journey home was via Frankfurt where there was a small delay but we eventually arrived in Birmingham at 0020hrs. By 0100hrs Barbara and I were safely tucked up in bed, feeling very tired but extremely pleased that the weekend had gone so well. I think it fair to say that we had conducted ourselves well as representatives of the UK MPS Society and the Polish MPS Society had treated us royally as guests at their family conference. So from Barbara and I, thank you for the privilege we had in representing all the UK MPS members and to the Polish MPS members for being so kind and friendly. ■

ERT for Lauren

by the Graver Family

We first heard about ERT in May 2003 while attending our local clinic in Bristol. Prior to this it was something that we had heard of but didn't really know anything about. When Dr Wraith mentioned it to us and the possibility that Lauren may be eligible to receive it we were very excited. Lauren had undergone a BMT back in March 2001 which unfortunately had been unsuccessful so we had resigned ourselves to the fact that there was nothing more that could be done to help her. We went back to Cornwall full of hope and optimism that we would soon be able to start the treatment.

As many people are aware, funding for ERT has been a bit of a postcode lottery and as it turned out our local PCT was not willing to fund it. We were lucky in the fact that our consultant did everything for us – approaching the PCT and putting forward our case etc. As time went on we put it to the back of our minds as Lauren was doing remarkably well and we didn't feel the need to push it forward any faster.

Eventually though the inevitable happened and things started to deteriorate. Lauren is PEG fed and since she had her gastrostomy, her stoma has never really healed up and has always leaked and been a problem. We tried numerous types of MIC-KEYS to see whether this would help, we saw consultants and spoke to several hospitals around the country to get advice but by the end of last year her MIC-KEY was popping out up to 4 or 5 times a day. Added to the problem was the fact that she had been screened and come back as having MRSA around the site. Lauren has most of her nutrition via her overnight feed and after her bolus feeds during the day she had to lie down otherwise her feed just poured out from her site.

Lauren attends our local mainstream school and we have been very lucky that they have worked with us to try and overcome these problems and keep her at school. By now though they were getting extremely frustrated with the situation from Lauren's point of view and the apparent lack of a solution. None of the staff who look after Lauren are medically trained but they were willing to learn and to help Lauren and sort her out every time the MIC-KEY came out. We are very grateful to them for everything they do for her above and beyond what they should, but that is another story!

Last autumn our consultant told us that as from April this year funding for ERT was to be controlled centrally rather than locally and not to give up hope as it may work in our favour.

We requested an appointment to see Dr Wraith in April this year because we had reached a point where we didn't know what to do next. He, along with Dr Jardine, examined Lauren and told us that our only hope of getting her gastrostomy problems sorted out was ERT because her liver and spleen were so enlarged. They decided to put Lauren forward to the Funding Panel as a special case as she didn't meet all the criteria for the treatment.

ENZYME REPLACEMENT THERAPY

Lauren's case was due to be discussed on 9 June and I was a bag of nerves waiting to see whether we would hear the outcome that day. I was next to useless at work and by early evening had convinced myself it was not good news. The next day we took Lauren's younger brother Alex to the Royal Cornwall Show as a treat while Lauren was at school. Unbeknown to us, our consultant had been trying to contact us with the outcome. We finally got a message from her at 5 o'clock to tell us that they had agreed to fund her for six months. We were overjoyed and frantically phoned around our families with the good news. The treatment was to be done at Treliske, our local hospital, which is an added bonus as there is no travelling involved each week.

Immediately prior to commencing the treatment Lauren was spending most of her time, when not at school, lying down watching videos and looking at books. She used to be able to bottom shuffle around but was unable to do this anymore. Sitting up for any length of time would mean her clothes would be saturated from her leaking stomach and she was also finding it increasingly difficult to support herself, especially her head. When sitting up she was bent over and very unstable. Her stomach was extremely distended and solid to touch and her stoma was enlarged and 'stretched'. She was often irritable and didn't like to be moved around or have her clothes changed. She found it very difficult to hold anything as her fingers were very clawed and her grip was weak. Her limbs were stiff and at times we think she must have been experiencing a lot of discomfort. Our once cheerful, smiley little Lauren had gone.

After that things seemed to step up a gear and we then received the phone call asking us to go for our first infusion on 6 July.

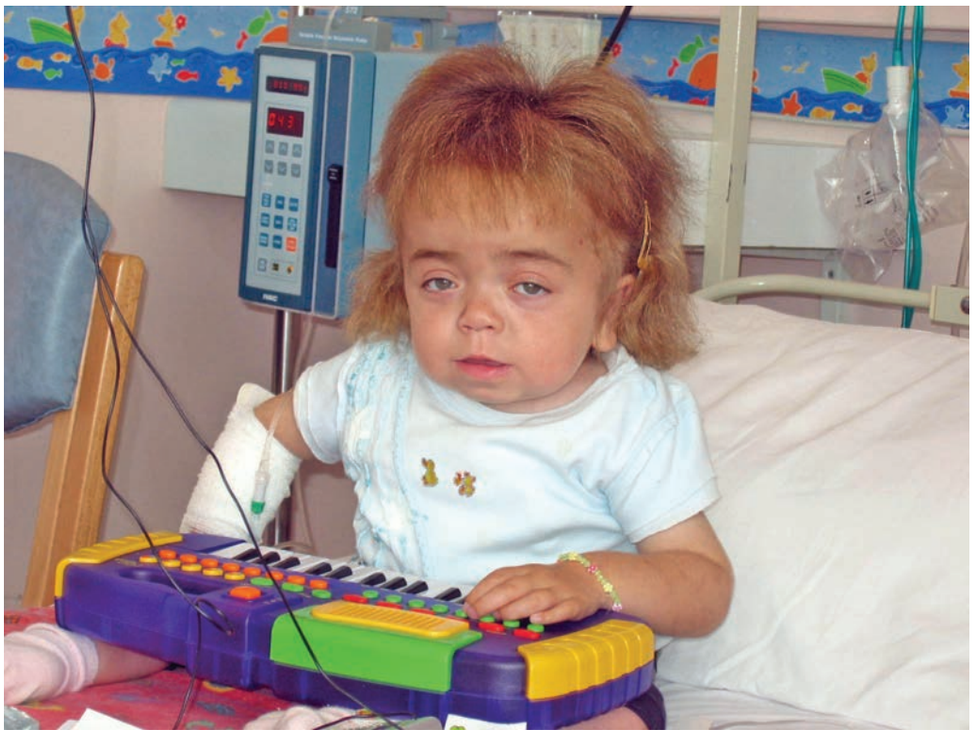
By the third infusion her stomach was visibly less distended and the doctor said that her liver and spleen seemed to have shrunk. After her fourth week she was bottom shuffling along the carpet – something she hasn't been able to do for a long time. She was also sitting unaided on the floor to do a jigsaw and trying to sit herself up from a lying position. At the time of writing this Lauren has had seven weeks of ERT. The changes we and other people have noticed in her are amazing. We would never have expected such changes in such a short time. Her fingers aren't as clawed and stiff, she is able to grip things much better. Her gastrostomy site is much better but it is not something which is going to sort itself out overnight.

Generally she has much better movement in all her limbs and doesn't seem to be as uncomfortable. She is back to singing her nursery rhymes and is much happier and contented in herself. For the first time since her brother was born nearly 2 years ago she is able to sit up and play together with him. All in all she is a very different little girl. Just this week she said to me "Mummy, I want to walk". She must be feeling a lot better in herself to want to try and stand and walk and hopefully now she will be able to go back into her standing frame and build up her muscles again. She is getting her quality of life back and is showing much more interest in wanting to get involved in everything that is going on around her. Since starting her treatment Lauren hasn't stopped talking! She's at it 24/7 to everyone and anyone who will listen to her!

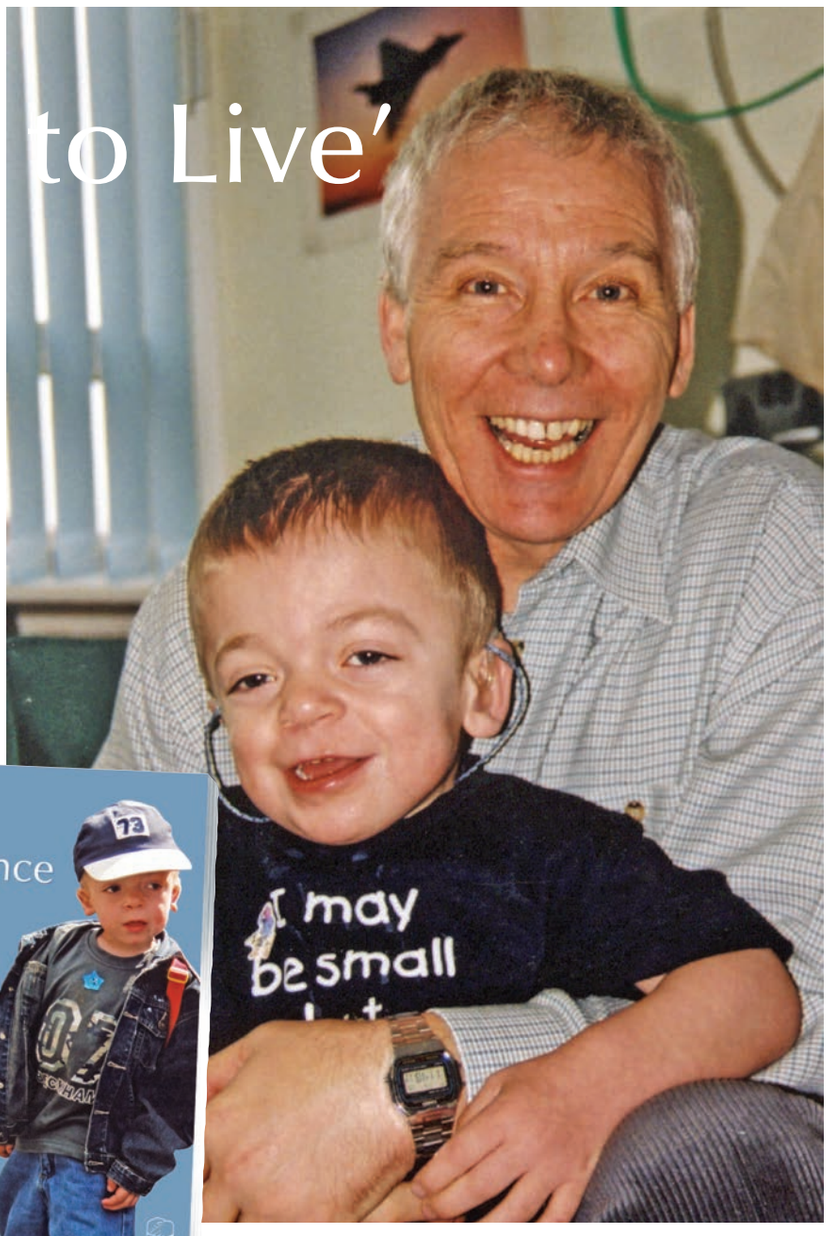
She is showing more of an interest in food and is now eating ice lollies, chocolate buttons and anything else that "hasn't got lumps in".

Lauren takes the weekly Friday visits into hospital in her stride. She sees it as a social outing and as long as we keep her supplied with plenty of sticker books and "milky buttons" she is happy! She loves chatting to all the staff who pop in to see her and is very good at bossing them around!

We cannot thank enough everyone who worked so hard to get Lauren her ERT namely Dr Gilbertson our local consultant and of course Dr Ed Wraith – our saviour. We can only hope and pray that after six months the powers who be will make a decision to carry on funding it.



'A Chance to Live'



The MPS Society has worked with Richard Dunn to publish a second edition of his book 'A Chance to Live'. The book is an inspirational account of how one family found the inner strength and outward courage to tread a trepid pathway. This story takes us from the devastating diagnosis of MPS I, Hurler Disease, on their much-loved son and grandson, through bone marrow transplants, to the creation of a future together with Isaac.

To order your copy
contact the Society on
0845 389 9901
or visit
www.mpssociety.co.uk

Bike for Sale

Special needs bicycle suitable for 3-9 year olds.

Comes with an attachment which allows parents to pull/push and steer (not pictured). Pedals can be adapted to fit most foot sizes with velcro fitting to ensure child's feet stay in touch with pedals and pedals remain in contact with the child's feet. Lap belt, side and back supports will ensure a safe comfortable and enjoyable ride. Adjustable seat up, down, forward, back and adjustable sides and back.

Good Condition
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DIRECT PAYMENTS? by Clare Cogan

A Direct Payment can be defined as money being given directly to an individual or family who live with disability so that they can pay for the support which they need themselves. This is achieved through the employment of an individual or individuals by the person who requires the support or the parent or buying in a service directly. Originally Direct Payments were only available to disabled adults, but following a change in the law in 2003, parents of disabled children became entitled to receive a Direct Payment for their child or children.

A Direct payment comes from the Local Authority. It is the responsibility of the Social services Department, i.e. the social worker to assess a child or individuals needs and agree the level of provision to meet those needs. This can be through services provided direct by the Local Authority, for example, overnight respite care but since the change in the law in 2003 Local Authorities are obliged to make Direct Payments available and social workers should inform you of your option to receive a Direct payment. If this is the option which is chosen then a person or the individual or families choosing can be employed to provide the support which the child or individual is assessed as needing. It is important to note that you have to have undergone an assessment of need with the Social Services Department before you can be offered a Direct payment.

The purpose of a Direct Payment is to allow individuals, and families to take control of the support and assistance they may require when they are living with a disability. Allowing people to receive the money directly is hoped to transfer the control, and consequently increase choice. One of the advantages of a Direct Payment is its flexibility. For example, if it was identified that you needed support with your child in the morning and evening for help with personal care, getting ready for school etc., then there is more flexibility to arrange for this person to attend at a time which is convenient to the family to meet identified needs. Sometimes, the provision of Home Care direct from the Local Authority to undertake a task such as this can be more inflexible as carers often have a list of people to attend to and may have to be at the home at a time which is not convenient to you or does not fit in with the timing of the school bus etc. However, sometimes the provision of a larger scale unit, for example, an overnight respite unit, can be an advantage rather than one individual working for you in the home, and often a combination of services, from both the Local Authority, and from Direct Payments is found to be the most supportive.

As outlined above, Direct Payments are an alternative way of funding a service which meets an assessed need. Many people use their Direct Payment to employ somebody to do the job that is outlined in the care plan which should be received following the social worker's assessment although on some occasions, individuals or families will use agencies. It is the individual or families responsibility to open a separate bank account in which they will receive the Direct Payment, and, if they choose to, employ their own carers, advertise, interview and recruit these people. In some cases, families or individuals may already know of someone who could help them, and the provision of a Direct Payment gives them the opportunity to pay them. Social Services Departments have a nominated hourly rate which they pay but added onto this is money to cover sickness and holidays which are the responsibility of the person receiving the Direct payment to cover. Depending on the number of hours which you have been given, national insurance or tax may have to be deducted from your carers payment. Most Local Authorities have set up support schemes in their area which can support people with drawing up adverts, interviewing people and dealing with the day to day issues which may arise with the provision of a Direct Payment. It is important to be aware that there is a considerable amount of paperwork involved in this scheme, including the possibility of a regular financial audit carried out by the Local Authority but the Direct Payments support group should be able to assist you with this. Any Direct Payment Scheme which is available should be able to provide the advice and help as well as the money.

Direct Payments Schemes are still a new initiative, although some have been operating longer than others in various Local Authorities. The important issue to be aware of is that following an assessment of need, an individual or child with a disability should be offered the option of a Direct Payment and have the right to choose. Despite the high level of responsibility that can occur with a Direct payment, its advantages include allowing increased flexibility, control and autonomy, to allow individuals or families to meet their needs in a way which bests suits them, their lifestyle and culture.

There are a number of sources of information and guidance provide about Direct Payments included below:

A Parent's Guide to Direct Payments – this includes advice on being an employer and how to advertise for a post, carry out interviews, undertake checks and references. This guide is published by the Department of Health and can be obtained from the internet, http://tinyurl.co.uk/kjf6 or are available from Department of health, PO Box 777, London SE1 6XH, Tel: 08701 555455.

A Guide to Receiving Direct Payments in Scotland is available from: Direct Payments Scotland, 27 Beaverhall Road, Edinburgh, EH7 4JE, Tel: 0131 5585200, Web: http://www.dpscotland.org.uk.

Direct Payments for Families is a guide that informs you of the Direct Payments scheme, how the scheme benefits families with disabled children and how the barriers to accessing Direct Payments can be overcome. Web: http://www.circlesnetwork.org.uk.

genzyme

Clinical benefits of Fabrazyme

Genzyme announces positive opinion from European authorities regarding clinical benefit of Fabrazyme

Genzyme Corporation has announced that the Committee for Medicinal Products for Human Use (CHMP) at the European Medicines Evaluation Agency has issued a positive opinion in response to Genzyme's request to include new evidence of clinical efficacy in its product labelling for Fabrazyme® (agalsidase beta) in the European Union.

According to the CHMP, the data, drawn from the largest randomised, double-blind placebo-controlled study ever conducted in Fabry disease, confirm that Fabrazyme administered at a dose of 1mg/kg of body weight every two weeks shows significant clinical benefit. Following review of the data, the CHMP has recommended to the European Commission that approval be granted to Genzyme to add the following information to the label:

Results from the trial demonstrated that the 'rate of clinical events was substantially lower among Fabrazyme-treated patients compared to placebo-treated patients, risk reduction = 53% in the intent to treat population, (p=0.0577), and 61% in the per protocol population (p=0.0341). This result was consistent across renal, cardiac and cerebrovascular events'.

'The results of these studies indicate that Fabrazyme treatment at 1mg/kg every other week provides clinical benefit on key clinical outcomes in patients with early and advanced Fabry disease. Because this condition is slowly progressive, early detection and treatment may be critical to achieve the best outcomes.'

The CHMP's recommendation will now be forwarded to the European Commission for final approval in all EU member states, Iceland and Norway. These data are also currently under review by the U.S. FDA.

Dr Stephen Waldeck, principal investigator of the study from Hope Hospital in Manchester, UK said: 'This trial provides the first randomised, placebo-controlled evidence that Enzyme Replacement Therapy impacts the clinical outcomes that cause morbidity and mortality in Fabry disease. The ability of Fabrazyme to address these critical organ systems increases our confidence that we can alter the course of the disease progression with early and adequate intervention.'

Prof. Christoph Wanner, University Hospital Wurzberg, Germany, added: 'The reduction in risk of clinically meaningful events seen in this trial highlights the need for early identification of patients with the disease, comprehensive management of symptoms, and prompt initiation of treatment to provide the greatest possible benefits.'

About the trial

The multinational, multicentre, double-blind, placebo-controlled trial investigating the impact of Fabrazyme on clinical outcomes began in January 2001 at 26 medical centres around the world. It was the largest study ever conducted in Fabry disease, enrolling 82 male and female Fabry patients with mild to moderate renal disease. Patients were randomised 2:1 at each trial site to receive Fabrazyme at a dose of 1mg/kg every two weeks or a placebo. The trial's primary endpoint sought to determine whether Fabrazyme would reduce the rate of occurrence of certain clinically important events that mark the progression of Fabry disease, namely renal, cardiac, cerebrovascular events or death.

About Fabrazyme

Fabrazyme was approved in the European Union in 2001 and in the United States in 2003, following completion of a 58-patient Phase 3 trial that demonstrated the ability of Fabrazyme to clear globotriaosylceramide (GL-3) from the blood vessels of the major organs affected in Fabry disease.

About Fabry disease

Fabry disease is caused by the deficiency of the enzyme alpha-galactosidase A, which leads to the progressive accumulation of lipids, primarily GL-3, within cells of the kidneys, heart, brain and other organs. Clinical manifestations of the disease include renal failure, heart disease, stroke and debilitating pain. It is estimated that 1 in 40,000 males has Fabry disease, whereas the prevalence in the general population is 1 in 117,000 people. The average life expectancy of patients with the disease is 43 years.

About Genzyme

One of the world's leading biotechnology companies, Genzyme is dedicated to making a major positive impact on the lives of people with serious diseases. Founded in 1981, Genzyme has grown from a small start-up to a diversified enterprise with more than 7,000 employees in locations spanning the globe. With many established products and services helping patients in more than 80 countries, Genzyme is a leader in the effort to develop and apply the most advanced technologies in the life sciences. The company's products and services are focused on rare inherited disorders, kidney disease, orthopaedics, cancer, transplant and immune diseases, and diagnostic testing. Genzyme's commitment to innovation continues today with a substantial development program focused on these fields, as well as heart disease and other areas of unmet medical need.

Fledglings is a not for profit organisation which helps parents and carers of a child with special needs of any kind to find simple, affordable solutions to practical problems.

Fledglings offer a free product search service to locate toys, clothing, developmental aids and other items which may stimulate the child's development or give relief to the carer. Many such products are kept in stock and they can obtain many more from the wide range of manufacturers and suppliers with whom they have contact. Once Fledglings have located a suitable product, they are usually able to supply it and do so at a competitive price.

Fledglings can help anyone who cares for children with special needs; parents, grandparents, childminders, foster carers, playgroup leaders, nursery nurses, health visitors etc. Fledglings also provide a range of other services including a newsletter and they advise on possible sources of funding for large, expensive but essential items.

For more information visit www.fledglings.org.uk. Please note that delivery times may vary depending on the level of demand.

Incy Wincy specialise in swimming products for children, from armbands to swim jackets and swim nappies to sun protection swimwear. They sell quality products at a reasonable price and particularly focus on products for children with special needs. Incy Wincy take orders by phone on **0118 3773 581** or online at **www.incywincy.net**.

disabledfriends.com

Disabledfriends.com is a global and fully interactive virtual portal for disabled people to allow them to create new friendships and build relationships within their own community and further afield; a central area to discuss issues that affect disabled people in their daily lives, as well as a place to provide support, offer advice and assistance to others. This is all achieved through topic and location talk rooms worldwide, discussion boards and forums, as well as a message, news and entertainment centre.

Disabledfriends has been online since March 2005 and has over 2,500 members in over 65 countries. We are growing at 140 new members per week. There is a lot of activity on the site, people messaging each other, talking in the global rooms, placing comments on our discussion boards and using the entertainment centre.

For more information visit
www.disabledfriends.com

EURORDIS

European Organisation for Rare Diseases

EURORDIS, the European Organisation for Rare Diseases and the MPS Society welcomes the adoption by the European Parliament on 7 September 2005 of the Regulation on Medicinal Products for Paediatric Use.

This long-awaited Regulation responds to a striking public health need as children who are still developing, respond differently to medicines from the way that adults do. About 50% of patients with rare diseases are younger than 19 years old. We estimate that two-thirds of these children are being prescribed unlicensed or off-label drugs. The risks of inefficiency and/or immediate or long term adverse reactions are clear.

Since December 2000, EURORDIS and the MPS Society have been actively advocating in favour of the prompt adoption of a European Regulation on Paediatric Medicines. EURORDIS has been working with national administrations, the European Commission and the pharmaceutical industry for the last five years in order to ensure that the interests of rare diseases patients and their parents were taken into account in the debate on paediatric medicines.

Today, EURORDIS congratulates all involved parties for the adoption of the Paediatric Regulation and is glad for the patients and their parents, who can now hope for the rapid development of medicinal products specifically intended for a paediatric use. The adoption of the Paediatric Regulation represents a crucial step for all the children of Europe!

EURORDIS also strongly welcomes the participation of patients' representatives within the Paediatric Committee established by the Paediatric Regulation.

EURORDIS' recent position paper on the regulation of medicinal products for paediatric use can be found on EURORDIS' website at www.eurordis.org

Society for Mucopolysaccharide and Related Diseases

National Registered Charity No. 287034

Become a Friend of MPS

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

What are the benefits of becoming a Friend of MPS?

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

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