Newsletter

The Society for Mucopolysaccharide Diseases

National Registered Charity No. 287034

Autumn 2002

Enzyme Replacement Therapy for Hunter Disease is on the Horizon

Rossendale Free Press

PARENTS' SHOCK FILM SHOW

Jubilee babies' silver lining

120+ JOBS ... HOMES SECTION 55-60 ... DRINKERS WARNED pg 3

Family planning giant street party for son doctors say is terminally-ill

PARENTS LIVING IN HOPE OF CURE FOR LITTLE JACK, 2

Cryers Panasonic

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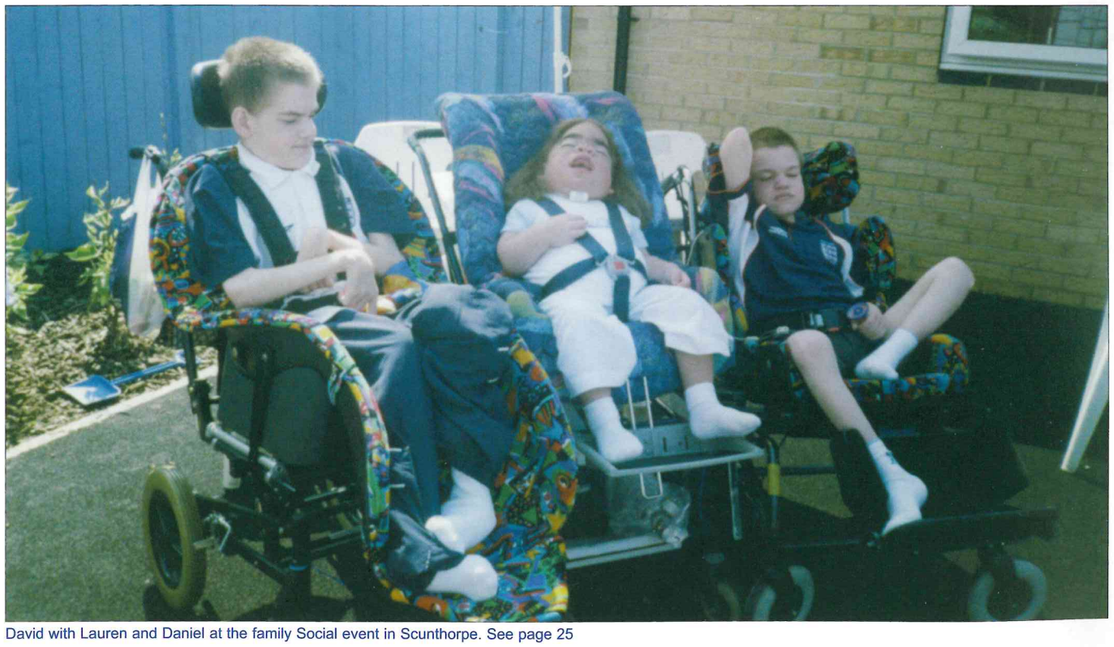


David with Lauren and Daniel at the family Social event in Scunthorpe. See page 25

Front Cover: Jack Heath

Director's Report

Christine Lavery

With the successful International Symposium in Paris behind us the Society has turned its attention to the planning and implementation of a programme of conferences and social events for the next 18 months. This programme will also incorporate events to celebrate the Society's 21st birthday year which starts in June 2003 with the Society's weekend conference 20th - 22nd which will be unique in running parallel sessions on palliative care, independent living and Treatment and New Therapies. There will also be a day conference in Scotland and Northern Ireland.

A series of Christmas parties are planned for November and December in regions throughout the United Kingdom. The locations and dates are given in the Newsletter. A lot of effort goes into arranging these parties so we really do hope that as many members as possible will come and join us.

The number of newly diagnosed individuals and families joining the Society has continued to climb. To meet the increasing demands of the individual advocacy service the Society is pleased to have appointed Alison West as Development Officer. Antonia Crofts has also re-joined the staff team as Administrative Assistant for the next eleven months prior to going to Teacher Training. During August we said goodbye and thank you to Fiona Woodcraft and Mounira Hadj-Rehouma. Both came to the Society on short-term contracts, Fiona to co-ordinate the International Symposium and Mounira to develop the Society's publications and achieved a lot for the MPS Society. Mounira's work continues with the recent publication of the Fucosidosis booklet.

Jeans for Genes took place on Friday 4th October 2002 and this year Jeans for Genes looks like it is going to raise even more money for research and to provide support to those affected by MPS and other genetic diseases. In January 2002 the Trustees approved a Jeans for Genes grant to part fund a consultant specialising in MPS and Related Diseases at the Hospital for Sick Children, Great Ormond Street. I am delighted to tell you that Dr Maureen Cleary has been appointed and will take up her post at the end of October 2002. Some of you will know Maureen as a consultant paediatrician in Manchester and we know Dr Wraith and his team as well as the MPS families that were under her are sorry to lose Dr Cleary. We also wish Dr Cleary well as she starts married life and her new post at Great Ormond Street.

Finally I would like to take this opportunity to thank all of you who have supported the Society over the past months organising fundraising events for MPS. Fundraising is a vital source of revenue which enables the Society to develop and deliver support and individual advocacy to more than 1000 families in the UK. Included with this Newsletter is the Society's first fundraising Bulletin, "Ten Steps to Fundraising." We hope this will prove useful to all of you in your fundraising activities for the Society and don't hesitate to ask for more copies for family and friends.

News From The Management Committee

Christine Lavery

The Trustees met in July to consider the following matters.

Election of officers

Chairman - Barry Wilson
Vice Chair - Judy Holroyd and Bob Devine
Treasurer - Judith Evans

Enzyme Replacement Therapy (ERT)

The Trustees received from the Director an update on ERT for MPS I, II, VI and Fabry disease. The proposal to form a users working group was accepted.

International Symposium, Paris

The Management Committee received feedback from Trustees and the Director. It was agreed unanimously that the International Symposium was a great success. The Trustees also thanked Fiona Woodcraft for all her dedication and hard work bringing this Symposium to fruition.

MPS Office

The Trustees agreed to look at several options for the MPS office when its lease expires in February 2003. They include renewing the lease on the current premises, moving to another rented property and purchasing an office premise with potential for an MPS resource and information centre. The Trustees are considering all the options and seeking financial advice.

Support and Advocacy

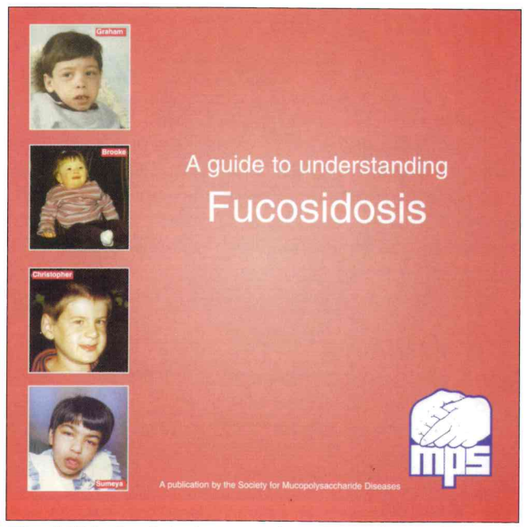
The Trustees as well as the ongoing individual advocacy agreed a range of support activities for 2002/2003 incorporating the Society's 21st birthday year which starts in May 2003.

Jeans for Genes

The Director informed Trustees that Jeans for Genes now has eight Trustees. Bob Devine and Christine Lavery being the Trustees for MPS.

Fiona Woodcraft

Congratulations to Fiona and her new husband Simon on their recent wedding on 24th August. We wish them both the very best of luck in the future.



The MPS Society has produced a new booklet on Fucosidosis, this is part of the Society's plan to update our range of disease publications.

Date for your Diary

MPS National Conference
20-21 June 2003
Northampton Hilton

Parallel conference sessions on:

Palliative Care
Achieving Independence
Treatment and New Therapies

Christmas Parties 2003

Helen Heard

CHRISTMAS IS JUST AROUND THE CORNER!!

MPS SOCIETY CHRISTMAS PARTIES 2002

YORKSHIRE AND EAST COAST
SUNDAY 1 DECEMBER
KALEIDOSCOPE CENTRE
SCUNTHORPE
12.30 - 4.30 PM

HAMPSHIRE AND DORSET
SUNDAY 1 DECEMBER
HILTON, SOUTHAMPTON
12 - 4 PM

WALES
SATURDAY 7 DECEMBER
COPTHORNE HOTEL, CARDIFF
12 - 4 PM

NORTH EAST
SUNDAY 24 NOVEMBER
HARTLEPOOL HISTORIC QUAY
12 - 4 PM

EAST ANGLIA
SUNDAY 24 NOVEMBER
PARK FARM COUNTRY HOTEL
NORWICH
12 - 4 PM

SCOTLAND
SUNDAY 24 NOVEMBER
ALMOND VALLEY HERITAGE CENTRE
LIVINGSTON VILLAGE
WEST LOTHIAN
1 - 3 PM

NORTH WEST
SUNDAY 8 DECEMBER
THISTLE HOTEL
HAYDOCK
12 - 4 PM

SOUTH WEST
SUNDAY 8 DECEMBER
HILTON, BRISTOL
12 - 4 PM

HOME COUNTIES
SUNDAY 8 DECEMBER
HILTON, BRACKNELL
12 - 4 PM

SOUTH EAST
SUNDAY 8 DECEMBER
HILTON, MAIDSTONE
12 - 4 PM

MIDLANDS
SUNDAY 1 DECEMBER
HILTON METROPOLE
BIRMINGHAM
12 - 4 PM

NORTHERN IRELAND
THURSDAY 28 NOVEMBER
HILTON, BELFAST
5.30 - 8.30 PM

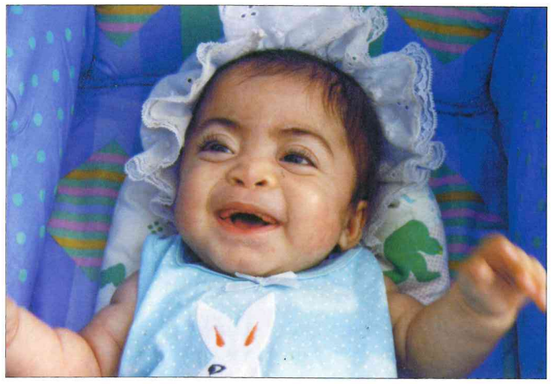
Each member of the MPS Society will be entitled to book for ONE Christmas Party at the subsidised rate. If any member wishes to attend a Christmas Party outside their area, could they please phone the MPS Development Team and ask for a booking form for the event they wish to attend.

The Afzal Family

Babita Afzal

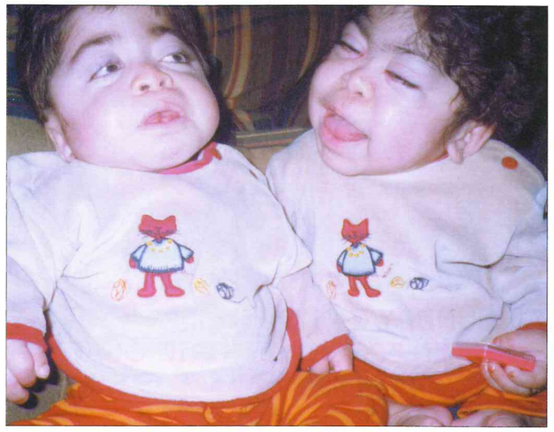
Living with two daughters who have MLII is the most heart rendering experience for a mother and also for the rest of the family who get so attached to the disabled children. At home every one has become a part of each other and we all seem to have forgotten that the two girls are disabled and will have to go one day.

I have two daughters who have ML II, Azaria who is 5 ½ and Alisha who is 4 fi. Both go to a special school in London called JFK. Azaria and Alisha are in the same class and are really adored and loved by their class.



Azaria can sing nursery rhymes, loves music and has a few certificates for being the class lead singer. She can also bottom shuffle and reach for her toys. She loves chocolate and cakes a lot.

Azaria also looks out for her sister Alisha who can't really sit or talk but can hum along. They look alike and they dress the same, just like twins. They have an elder sister Anika who is 11 years old and healthy. She looks after her sisters so much: she puts them on the bus, plays with them and teaches them new things all the time. Then I have two healthy boys. Mustapha is 2 ½ years old. He also likes to play with Azaria. He wouldn't let anyone near the girls or their toys. Our last son is 3 months old. We all love him. Azaria sings songs to him all the time. Finally, the children's father is Afzal and he loves us all.



Every day, going into their bedroom in the morning scares me and I always pray that nothing has happened to them. I wish they both live longer as they enjoy a good quality of life. They go out a lot: to respite at Little Haven Children's Hospice, to a lovely day nursery called Richard House daycare and during the holidays they go to play-scheme. They have been to Carol concerts, to the MPS Christmas party at 10 Downing Street and to Tweenies' live shows which they love watching. They go to the bowling alley, seaside…they have even been to the Millenium Dome.

When we go shopping Azaria loves the attention she gets… by the way, the girls have even had parts in the school panto as baby Jesus and Mary. Needless to say that they hardly ever spend a Sunday at home!

We look at them as special angels who are at our home for a holiday. One day they will go back to God, to their own home to rest.

The Gremo Family

Janet Gremo

Hello I'm Janet Gremo. I live with my husband Rob and our two children Stephen 13 and Nathan 10. Nathan has Sanfilippo disease. Our story is similar to lots of others but at the same time unique to us. Stephen was a lovely baby who had read the textbook and so knew what to do and at what age to do it, then came Nathan. Nathan was born by emergency caesarean, which was not a positive beginning and all the usual Sanfilippo problems followed, although at the time we didn't know why he had the constant ear infections etc. The worst problem we have suffered has been his inability to sleep; it has at times been tortuous.

Despite all the problems we kept trying to help him talk and use the toilet, but had only a limited response. Unfortunately our G.P. didn't know the disease and Nathan doesn't have too much of the Sanfilippo look so his diagnosis didn't happen until a month before his eighth birthday, two and a half years ago. It changed everything about the way we live our lives and continues to require us to think about ways to solve problems as they occur.

A big change happened when we attended the conference nearly a year ago. We had finally accepted what had been thrust upon us. The biggest revelation at the conference, which was actually obvious, was that Nathan will continue to grow and when he is eventually immobile he will be a big teenage boy, like some of the children we met. Although we knew, we hadn't thought about it. We then realised we needed to move house as the small terrace we lived in would eventually be much too cramped and there wasn't any room to extend. The hunt was on to find a suitable bungalow. That was the easy part. We then had to acclimatise Nathan to an unfamiliar place, so we visited it a couple of times before we moved in.

The moving day arrived which happened to coincide with staff training week at the hospice, so no help there! Nathan went off to school and we rushed around packing things as the removal men were taking them. It was a time to be thankful that Nathan had already destroyed most of our bits and pieces so not too much left to pack! Then another hiccup as the building society hadn't transferred the money and we didn't collect the keys until 30 mins before Nathan got home from school. We raced to help unload the van as Nathan appeared around the corner. It was very worrying as we weren't sure how he would cope. We needn't have been anxious because he adored the bungalow and as the removal men sorted the beds out, Rob put up the safety gates to keep Nathan safe, he and I went into the lovely big garden and tried out most of the leaves for taste, well he tasted them and I risked getting my fingers bitten hooking them out of his mouth.

The relief was enormous it was going to be okay. Not everything went smoothly from then on. Nathan took a while to settle into a nightly routine even with the sedative that he has, and as we were all tired from unpacking there were a few difficult weeks. Luckily there was one saving grace, which we resorted to, too frequently, we have a fish and chip shop 200 yards away and it's Nathan's favourite food but I'm afraid I've definitely gone off it.

The summer holidays are nearly over as I write this and I can honestly say this has been the easiest six weeks we have had for a long time, probably with still having things in packing cases we are living the minimalist life that is guaranteed to make living with a Sanfilippo child in the hyperactive phase much easier . Despite all the work and anxiety I know we have done the right thing and hopefully we shall all be able to live life without too much disruption in the future, just enjoying our children while we can.

The Lloyd Family

Sheryl & Stuart Lloyd

The last 8 months have been a roller coaster of emotions for both Stuart & myself. I won't go into it all in detail, but Ben our 4 year old son, soon to be five, was diagnosed on January 18th this year with Sanfilippo type A. 5 days later, Jake, our 2 year old was diagnosed also.

All parents reading this will obviously know how we felt, to be told that our seemingly 'normal' looking boys could have this terminal illness, to which there is no cure - that sick, numbing feeling that is with you from the moment you wake, 'til the moment you go to sleep at night.

All I wanted to do was talk to somebody whom was in the same situation as us, so we didn't feel isolated or alone. I contacted MPS and we soon had 2 link families to befriend. Alton Towers was the first time we came into contact with people in 'the same boat' as us. We talked and for the first time even laughed. They seemed positive, and were getting on with life - maybe there was some sort of hope for us.

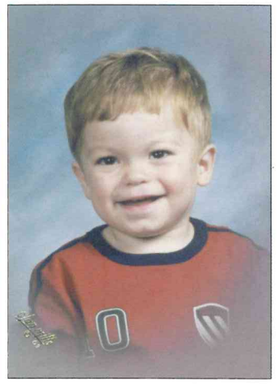
Talk about jumping in at the deep end - Paris was our first conference, the future stared at us in the face. People were smiling at us, not staring. The children were having a great time and Ben & Jake didn't stand out, they fitted in. There were no awkward silences, we were all the same, families with 'special' kids. We just about held up in Paris, but on arriving home we cried hard. It hurt, the reality of it all was kicking in.

Today, 8 months down the line, we see so many changes in our beautiful son Ben, typical of Sanfilippo children. Jake has just been for his 2 year developmental check and is showing some delay in his development and has been recommended for speech therapy. This is where it all began for Ben.

We have people to turn to now, we would like to thank those people from the bottom of our hearts, because they have made us see that life can go on, you can still have a laugh and get drunk. We went to Gail and Nick Barnets' house on bank holiday Monday, mum and dad of Faye - Sanfilippo type A. It turns out they live only a stones throw from us. John and Jo Allen were there too with Bethany, Sanfilippo type A. We had piggy back races, ran round the mop and even timed each other racing on Faye's special bike - how sad is that?

We got through so much booze you would turn green, but we were all there together sharing precious time - we'd definitely do it again!!!!

To any newly diagnosed family, we'd just like to say, it does do you good to talk and it does help. Gail and Nick, Jo and John have been our rock over these last few months and we'd never have got through it without their support. We have also spoken to Jo Richardson with Ben from Rugby and Sandra Doherty from Northern Ireland and we have also just been linked with another family with 2 children with Sanfilippo from the North.



Being parents of these special children, makes you sit up and realise just how precious their little lives are and that you have to compress everything you can into a short time and give it all you've got and NEVER EVER take them for granted.

If anybody would like to contact us, please feel free to do so, by e-mailing: stuart.lloyd@tiscali.co.uk

Bethany's Big Surprise

Sophie Denham

Eaton Automotive Fluid Connectors Operations run a charity event every year where they raise money for charity. At the time when events were being discussed John, Bethany's Grandfather was working at the company and told them about Bethany's condition and the MPS Society.

Eaton Automotive Fluid Connectors Operations wanted Bethany to benefit from the charity event and asked John and Joanne; Bethany's parents what Bethany would really like or need. Like every child Bethany wanted to have a bike that she could ride.

Eaton Automotive Fluid Connectors Operations held a charity golf day, which was a great success and a lot of money, was raised for charities.

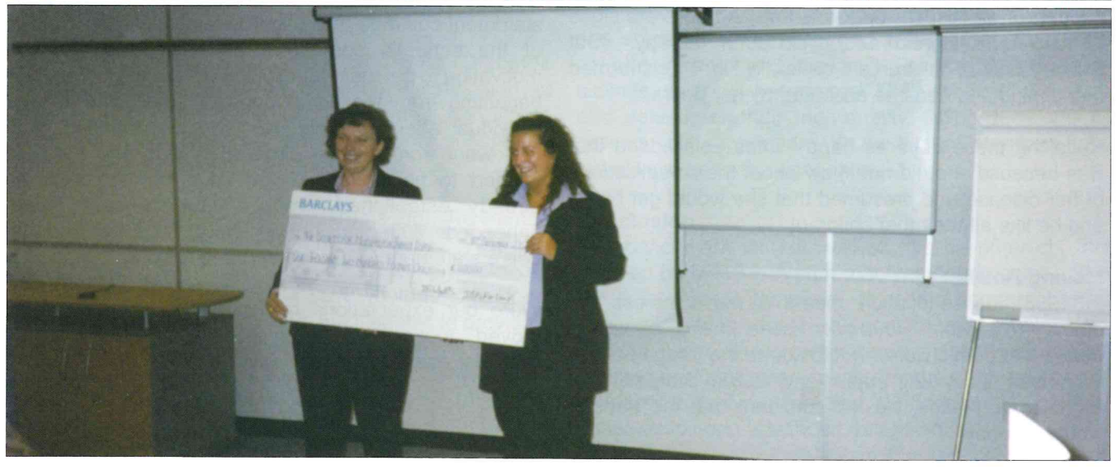
On Thursday 19th September I went along to Eaton Automotive Fluid Connectors Operations on behalf of The MPS Society to be present as Bethany was presented with a specialised bike, which she would be able to ride.

The company also presented me with a cheque for the Society for £1200.00, which had been raised on the day.

A further cheque for £100 was also given to the Society from Moss Plastic Parts Ltd who are a supplier for Eaton Automotive Fluid Connectors Operations.

The MPS Society would like to thank Eaton Automotive Fluid Connectors Operations and Moss Plastic Part Ltd for their kind donations.

We also hope that Bethany has many hours of fun on her new bike.



Lois's Life

Linda Pack

Introduction
Lois has just celebrated her 14th birthday. It is 10 years since her second and successful bone-marrow transplant.

Lois was just 2 and a half when diagnosed with Mucopolysaccharide Disease Type 1 - Hurler Scheie. Ed had the onus of providing the diagnosis and a prognosis that left us with some sort of hope. As Lois's parents, Howard and I regard ourselves as fortunate that her condition was amenable to bone-marrow transplant and that her younger brother Alex was a compatible donor.

Having been to a conference last year, it is reassuring to know that Lois is receiving the benefits of worldwide research and technology, controlling the symptoms of Hurler's and also treating the effects of two bone-marrow transplants. Much expertise has gone into adjusting her skeleton, her height and soon her sexuality. So how are we helping her to cope psychologically?

Primary Education
Lois's memories of her early years are mercifully vague and there is nothing to be gained in encouraging her to recount them. To say that she was desperately ill after total body irradiation would be an understatement. Her survival is a testament to her character and fortitude. A particularly difficult time for the family came when Lois was discharged home following almost 18 months in hospital, much of it in isolation on the Bone Marrow Transplant Unit. She had rarely met her brother who was now a robust 2 year old. The joyful anticipation of being a united family under one roof was intense. I soon realised how wrong I was to presume that son and daughter would share the same joy! They disliked each other on sight. My son wanted nothing to do with the vomiting child who was getting all the attention, and very little to do with me either. The dust took some time to settle!

Her first happy memories are of attending Primary School - starting with a statement detailing full time support of an NNEB. Lois was frail and bore the signs of long term steroid and cyclosporin therapy. But Primary School children are generally kind. Lois formed friendships and became accepted by her peers.

Recalling these days as happy times Lois added that was because she did not know about the complications of her disease and presumed that she would get better and be like all the other children!

Moving House
In 1998, job re-location meant a move across the country to Lincoln. Our new house in a small village meant that both children would attend the village school. With less than 100 pupils and Lois's Statement of Educational Needs, we felt confident that the transfer would be easy.


Lois's brother, Alex

Lois's statement detailed that most help would be needed to facilitate her mobility, ensuring that all info was accessible to her and to develop her IT skills.

It quickly became clear that the Headteacher had no intention of employing any extra help for Lois. When pushed, he said that he would ask someone to come and listen to her read. As Lois's reading skills were never in doubt, we indicated other areas where the statement could be more appropriately utilised. Neither of the school's computers were working and the motivation to get them functioning did not exist. Far from benefiting from the advantages of small classes and individual attention, Lois was now just marking time. She went from a wide circle of friends and full time support to becoming isolated as a newcomer to an already established group. Our concerns went unheeded. With hindsight we should have ensured that Lois's statement was honoured and used as intended.

Were our expectations realistic - that someone who moved, looked and even sounded different would integrate easily into a new environment?

Secondary Education
Our local comprehensive school is acknowledged to be one of the best in the country. With over 1500 pupils, most of them arriving by public transport, it is vastly different to what Lois has been used to. The school has an experienced co-ordinator of the SEN Department, and Lois's statement was tailored to fit her needs.

Transferring schools also coincided with the beginning of a programme of orthopaedic surgery. Lois was taught at home for several weeks following each operation. Taxis were provided to transport her to school whilst she was unable to walk to the bus stop. In 2000 the option of treatment with Growth Hormone was offered. With her younger brother already towering above her, Lois decided she would like to be taller, and began to give herself daily injections.

Social Integration
Life had settled into some semblance of normality punctuated by spells in hospital, recuperation and return to school. Lois had no special friends. Whilst getting on with her classmates, she considered a lot of them silly and often messing about. Was she so disparaging because she was excluded from this fun? As she walked slowly, the other children from the village left her behind at the bus stop. Lunch breaks at school were spent in the SEN department where it was warm and she could use the computers. It also provided an area where Lois did not need to socialise - it was comfortable and safe.

Our regular meetings to discuss Lois's progress at school and statement reviews focussed on her mobility needs and the good academic results she was achieving. But I was also interested in how she was relating to the other children. Lois had been asked to complete a Quality of Life Questionnaire during a visit to the Endocrine Unit. The answers confirmed my concerns. Lois was not the contented child who easily adapted to treatments and procedures. She spoke of frustration at her lack of mobility. She felt that as soon as she had recovered from one operation she had to have another.

At school she prefers classes when she is totally independent of her classmates e.g. PE. She has no wish to socialise with other children.

Lois has recently been diagnosed with Diabetes Mellitus, currently requiring twice daily estimation of blood sugar. She feels depressed that her life is complicated by injections and now a low sugar diet. Most of all, Lois wants to be normal.

Normality has been our aim as parents too. We wanted Lois to benefit from mainstream education. Was this denying her real needs in order to comply with our ideals? We presumed that a SEN would help to "level the playing fields" of school life, but it seems in providing more support, we have relieved Lois from the need to socialise and integrate as she withdraws to activities and areas that she feels able to cope with.


Lois as a brides maid

The Future
Lois has been making her own decisions regarding treatment for some years now. We are already discussing future operations to remove staples from one leg and a metal plate from the other. Her desire to stay away from hospitals is not an option.

Lois would like to take advantage of Further Education. Her choice of jobs would involve work with animals but she is unsure in what capacity. Her brother Alex is now 11 and in perpetual motion. The contrast between siblings could not be greater. They are both cherished and valued for their individuality. Our decisions as parents, although made with the best of intentions have not always been the right ones.

Conclusion
The benchmark for Lois will always be other children affected by M.P.S. A path that was nearly ours, and is too painful to contemplate. It seems ungrateful to be suggesting even greater support for children like Lois, but she is one of a growing number whose needs both physical and psychological are unique. We will need to prepare ourselves for the challenges of puberty, relationships and adulthood that lie ahead.

BMT Clinic, 6th September 2002

Alison West - Development Officer

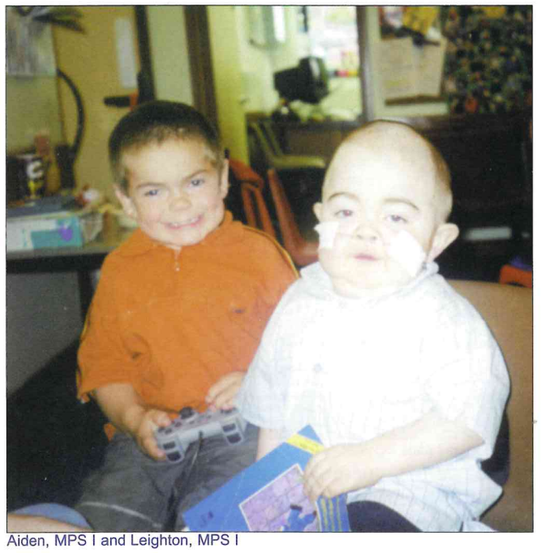
This was my first BMT clinic, and I was very much looking forward to meeting the individuals and families and getting involved. While I managed to introduce myself and spend some time with the families, everyone seemed much more interested in exploring the new extension to the RMCH's Willink unit!

The day was less chaotic than I had been led to believe it would be, but there was no doubt that the children's energy levels far surpassed my own. The toys and games were shown no mercy and the playstation proved immensely popular (with anyone under 13 that is. No-one older had any idea how to work it!).

Ellie and I were there, representing the MPS Society, and greeting everyone who arrived for the clinic. It was lovely to welcome so many of you and we were pleased to see how well everyone looked.

It is always interesting to find out how children and adults with MPS are doing post transplant and we supported the individuals and families present with DLA applications, achieving home adaptations, achieving grants for equipment and by listening to any medical concerns.

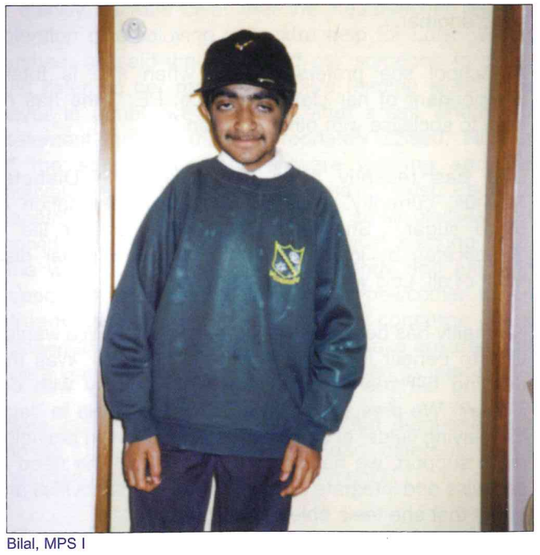
I would like to express our thanks on behalf of everyone at the clinic to the whole Willink team, especially Bernie, Gill, and Sue, and Ed Wraith and the medical team. We remain indebted to their commitment and expertise in running this clinic.



Aiden, MPS I and Leighton, MPS I

Mohammed, MPS I and his family

Jacob, MPS I



Bilal, MPS I

Birmingham Clinic, 12th July 2002

Sophie Denham

Ellie and I attended what was another successful clinic at the Birmingham Children's Hospital.

For me it was the first opportunity to meet many of the MPS families and individuals who live in and around the Birmingham area.

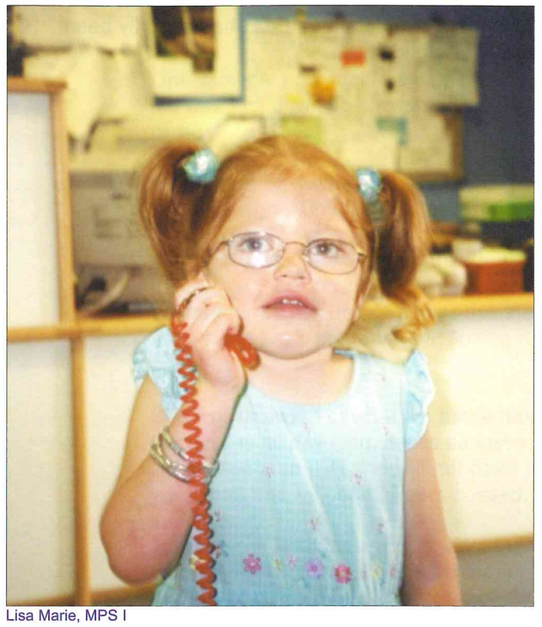
Although the waiting area was small and space was limited the day ran relatively smoothly and everyone said how they had benefited from meeting Dr Ed Wraith, Dr Chakrapani, Ellie and myself.

We have received many evaluation forms highlighting the lack of room at the clinic and are currently looking at other facilities within the hospital, which can accommodate the clinic comfortably.

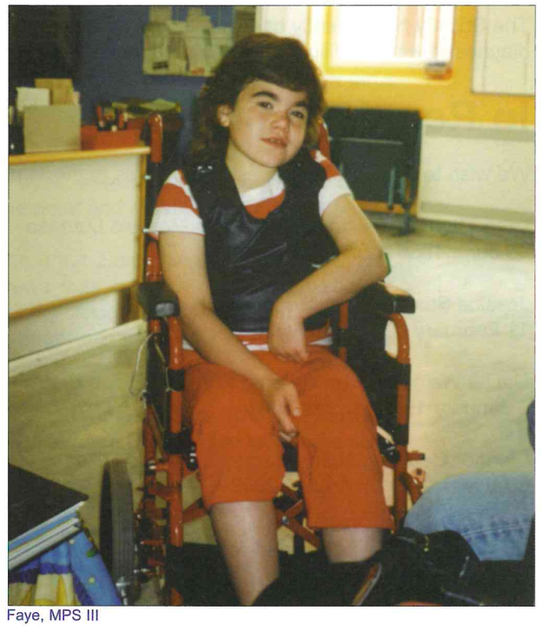
I would just like to thank all those who attended the clinic and I look forward to meeting you at future clinics.

Our thanks also go to Joy Wright for her help in organising the clinic and to Dr Ed Wraith and Dr Chakrapani for once again another successful clinic.

We are very grateful to Dr Ed Wraith for travelling the country and enabling individuals and families to access expert medical consultations in their regional areas.



Lisa Marie, MPS I



Faye, MPS III

David and Jamie, MPS II and their brother

New Members

Paula's son, Reece, has been diagnosed with MPS I Hurler disease. Reece is 2 years old. The family live in the South East.

Dave and Maria's daughter, Jasmine has recently been diagnosed with Sanfilippo disease. Jasmin is three years old. The family live in the South West.

The Society has recently been contacted by Marjorie Banks who has Fabry disease. She lives in the North West.

Jon and Teresa's son Toby has been diagnosed with Hurlers disease MPS I. Toby is one years old. The family live in Essex with Toby's older sister Bethan.

Sandra and Trevor's daughter, Laura, has been diagnosed with MPS IV Morquio disease. Laura is 10 years old. The family live in the North East.

Jeanette and Philip's daughter, Kayleigh, has been diagnosed with MPS I Scheie disease. Kayleigh is 12 years old. The family live in Wales.

The Society has recently been contacted by Sian and Joe Purchell whose daughter, Elsa, has recently been diagnosed with Sanfilippo disease. The family live in the South of England.

In Remembrance

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

Lynn Thompson who suffered from Morquio Disease
16 June 1965 - 27 May 2002

Jessica Stuart who suffered from Hurler Disease
11 February 1991 - 8 July 2002

Daniel Wainman who suffered from Sanfilippo Disease
8 January 1988 - 8 July 2002

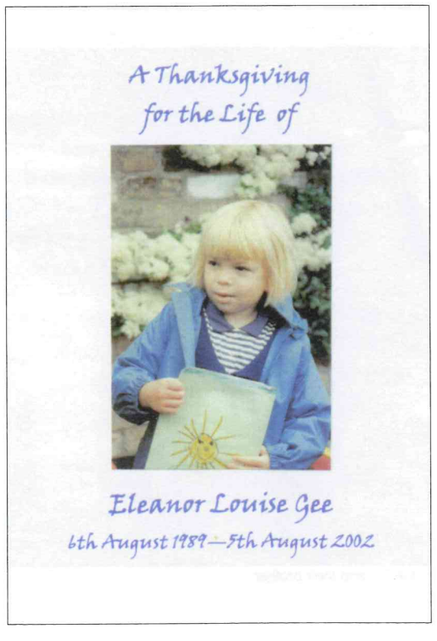
Eleanor Gee who suffered from Sanfilippo Disease
6 August 1989 - 5 August 2002

Eleanor Gee

Remember Me

To the living, I am gone
To the sorrowful, I will never return
To the angry, I was cheated
But to the happy, I am at peace
And to the faithful, I have never left
I cannot speak, but I can listen
I cannot be seen, but I can be heard
So as you stand upon the shore
Gazing at the beautiful sea, remember me
As you look in awe at the mighty forest
And its grand majesty, remember me
Remember me in your hearts
In your thoughts and the memories of
The times we loved, the times we cried,
The battle I fought and the
Times we laughed
For if you always think of me, I will
Have never gone

Author unknown



Jessica Rose Stewart

Mr & Mrs Stewart

Sadly we lost our daughter, Jessica (MPS I) on July 8th 2002. She was 11 years old.

As a family we had been preparing for her death since her diagnosis at 9 months, when we were told that she had 2 years at the most. Once we had been to see Dr Ed Wraith (Dr Chookyegg to Jessica), we were told that Hurler children have a life expectancy of between seven and ten years (dependant on the Hurler mutation).

Life with Jessica was one long learning curve; she proved to us all that a determined spirit can win through. It was because of her determination to get as much out of life as possible, that I began to plan for her funeral. Some people thought it a very good idea, as in grief you don't always do what you want to do and then later regret how things went. Other people thought I was morbid to plan such things while Jessica was fit and well. I'm glad I did plan as on the day, as sad as it was, everything went brilliantly and, as a family, we regret nothing.

I would like to share with you a poem I started to write soon after Jessica's diagnosis, it helped me come to terms with what we had been told. Over the years I have changed and adapted it, as Jessica passed significant milestones. The last change was from present to past tense.

Jessica Rose

One winter's morning there was snow on the ground, a chill wind blowing, a baby girl's life was just dawning. Joy she brought, with lots of tears and smiles over her miracle of life.

Nine years of waiting, to see her bright blue eyes, bare head and soft skin, such a beautiful sight. Tiny fingers, toes, a stubby little nose and rosebud lips.

Time passed, fed on breast milk and love she grew. Something was not quite right though as she was not developing. The news was bad, she was not well, our contentment was shattered, was this the beginning or the end? Our little girl was to die before she lived, the tears that flowed were of anger and sorrow for what should have been.

She was unaware of what her future held and played without a care. So full of laughter and smiles all day long, it was almost too painful to bear. Please God let her die now, in her sleep, so that she will not have to suffer, so that we won't have to suffer. Can we cope? Will we cope? Years passed.

Jessica, our tears turned into laughter, our odd looking child, so very beautiful to us. Our flesh and blood, a gift not a burden. You don't have to be perfect to be beautiful.

God gave us a very special child, all smiles and giggles. What was lacking was made up in her determination and sunny nature, her wicked sense of humour and infectious laugh. Everyone you met, you won over with your bravery and courage. You were such a nice person to know. You suffered in relative silence, struggled with things that we took for granted.

When your beautiful eyes went dull - their light forever gone, did you give up? No, you adapted and carried on. When your ears faltered and your world became silent, did you give up? No, you carried on in your own determined way. Little Miss Bossy Boots!! You were generous and kind to others, especially to your little sisters whom you adored. They understood your struggle, so they helped, teased, laughed and cried with you. The memories they have of you will be with them always.

You were a credit to us JESSICA ROSE - a gift from God to be treasured forever and always in our hearts.

When the time came for your forever sleep, we say our goodbyes in a celebration of music and flowers, hoping that you will be happy with God, only he knows, how much we love you, JESSICA ROSE.

ENT Problems In Mucopolysaccharidosis

Ian Bruce and David Luff, specialist registrars in ENT North Manchester Children's Hospital

Children with Mucopolysaccharidosis (MPS) may present to the doctor with a variety of medical problems due to the fact that the partially broken down mucopolysaccharides that characterise this condition are distributed widely within the body. This article aims to explain those complications that occur in the 'EAR, NOSE and THROAT'. We have not considered each type of MPS separately and it must be understood that every child who is diagnosed as having MPS may not encounter the problems detailed here. In writing this we hopefully aim to give you the knowledge needed to successfully discuss any ENT problems with your doctor before a definite management plan is agreed.

THE EAR

There are 2 main types of hearing loss. Conductive hearing loss (CHL) signifies a problem in the mechanism of transmission of sound energy to the hearing sensory organ. Sensorineural hearing loss (SNHL) occurs when there is a problem with either the sensory organ (cochlea) or the auditory nerve transmitting signals to the brain.

Normal transmission of sound across the middle part of the ear is most efficient when the pressure on either side of the eardrum is the same. The Eustachian tube runs from the back of the nose to the middle ear behind the eardrum. Its job is to match the pressure in the middle ear to that in the surrounding atmosphere. If this tube is blocked or not functioning correctly a negative pressure may develop in the middle ear. If this continues then the eardrum becomes sucked in and will move less in response to sound. Fluid from the cells lining the middle ear will build up in the middle ear to form a thick 'glue' preventing the tiny bones that span the gap between the drum and the cochlea from transmitting the sound as easily. This can be explained by imagining how much more energy it takes to jog on the spot in a swimming pool than on the poolside. In order to treat this condition a small cut (myringotomy) can be made in the eardrum under a general anaesthetic, the fluid can be sucked out and a grommet can be fitted. A grommet is shaped like a 'cotton reel' with a hole running through it and a flange on either end. If a grommet is placed in the eardrum the space behind is now linked to the outside world and the negative pressure that can lead to glue ear cannot be created. Grommets are temporary and with time they will fall out. It is hoped that they will last 6-18 months, subsequent to which further grommets may be needed. A SNHL is not managed surgically, but the child may benefit from the use of a hearing aid to amplify the sounds reaching the cochlea.

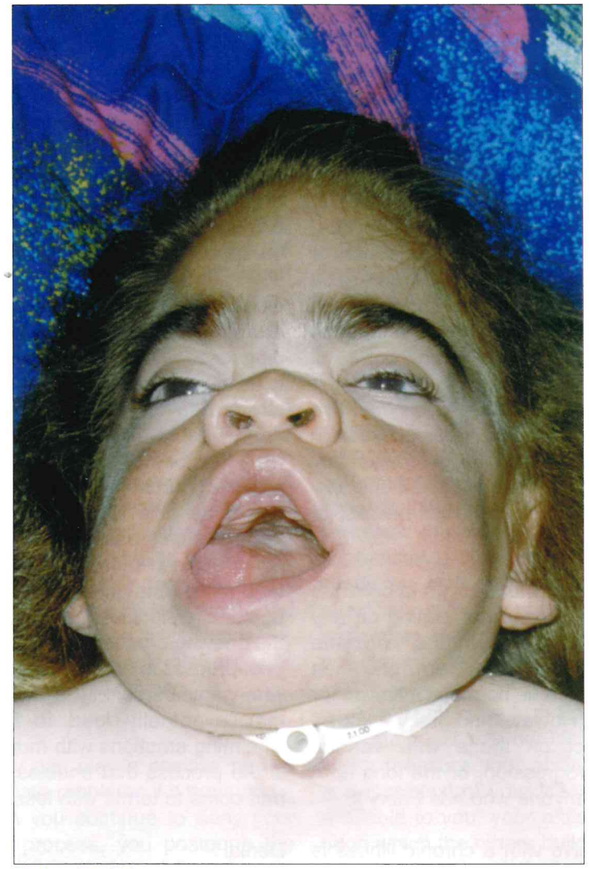
THE NOSE

The lining of the nose may become swollen narrowing the space through which to breathe. This is often accompanied by frequent infections of the nasal lining resulting in chronic nasal obstruction and an almost continuous 'runny nose'. The most significant problem encountered is the overgrowth of the adenoids. The adenoids are swellings of glandular tissue that are found at the back of children's noses. The adenoids are generally large in childhood but have shrunk and disappeared by the teenage years. In MPS they can enlarge to a significant size leading to a 'blocked nose' and may cause obstruction of the opening of the Eustachian tube into the nose. An operation called an 'adenoidectomy' can be performed under a general anaesthetic to scrape the adenoids away if necessary.

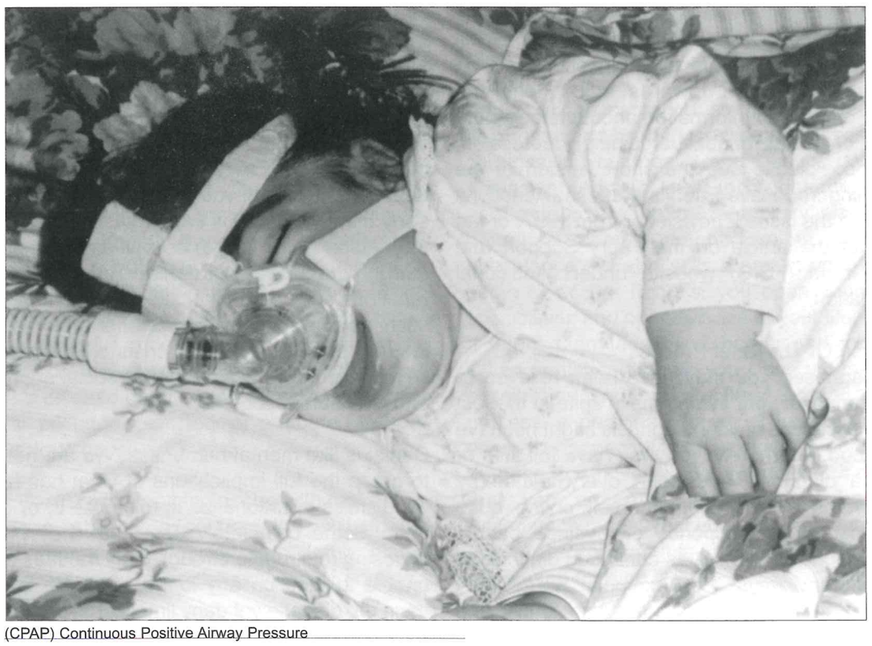
THE THROAT

Partially broken down mucopolysaccharises may be deposited throughout the airway leading to the lungs. This alters the appearance of the airway and may narrow it significantly. The tonsils are also composed of glandular tissue, like the adenoids. Enlargement of the tonsils found on either side of the back of the mouth, along with this airway narrowing and adenoidal enlargement may lead to noisy breathing. This airway narrowing is often worst at night and the parents may notice that the child is restless and snores loudly. If the airway is completely blocked for a short period, then the child will stop breathing over that period. If severe this is called Obstructive Sleep Apnoea Syndrome (OSA Syndrome). If the child stops breathing then the levels of oxygen in the bloodstream will fall. This is used by doctors as a way of assessing the severity of the problem. You may be asked to bring your child into hospital for a 'sleep study' during which a probe will be placed on your child's finger to record the levels of oxygen in the blood whilst they sleep and observations of chest and abdominal movements during any episodes where the oxygen level drops will be recorded. An operation to remove enlarged adenoids and tonsils may help to improve the obstruction. If this fails or there is no enlargement of the tonsils and adenoids, OSA may necessitate the wearing of a special mask that provides continuous positive airway pressure (CPAP) whilst asleep. This positive pressure helps to splint the airways open preventing them from collapsing and obstructing. Usually this is enough but if these measures are not sufficient it may be necessary to perform an operation called a tracheostomy. This is an operation to create an opening from the skin at the front of the neck down into the windpipe and this tunnel is held open by a tube called a tracheostomy tube. The child will then breathe through the tube, with air passing from the front of the neck into the lungs, thus bypassing the site of the obstruction in the upper airway. This is a significant undertaking and much discussion will be undertaken between the parents and the doctors. Following the surgery there will be an intensive period of education for the parents in all aspects of care of a child with a tracheostomy and only when both the medical staff and the parents are happy will the child be allowed home.

The advent of specialist MPS clinics such as that held at the Willink Biochemical Genetics Unit at the Royal Manchester Children's Hospital has meant that all of the Paediatric Subspecialties involved in the children's care have developed a great deal of experience in the management of complications of MPS. We hope that this article has helped to explain our involvement as ENT doctors and we would urge you to always ask questions!



Tracheostomy Tube



(CPAP) Continuous Positive Airway Pressure

Coping With Fabry's Disease: Denial vs. Acceptance

Dr Barbara A. Wehmann

Thirty million Americans have a chronic disease, an illness that cannot be cured. Like many other diseases, Fabry's may be diagnosed at any time in your life from the prenatal period to well into adulthood. Though chronic diseases differ, those with such illnesses and their loved ones are united by feelings such as shock, disbelief, denial, anger, fear, anxiety, depression, loneliness, and despair.

Learning to live with a chronic disease is a process of continual complexities and changes, as well as physical and emotional challenges. Changes may be minor, or they may be profound, but they eventually will have to be accepted. Coming to terms with all of the issues related to Fabry's is an intensely personal journey. Your struggle to cope may get harder before it gets easier, but it will get easier in time. Coping with Fabry's requires doing what so many chronically ill people do each day; live with their disease the best they can. Your success in accepting Fabry's, meeting the continual challenges, and adapting to the ongoing changes will determine how happy and satisfying the rest of your life will be despite the fact that Fabry's is one part of it.

It is important for those dealing with Fabry's to continue to improve coping strategies instead of putting off any progress toward adaptation until the enzyme replacement treatment is available to them, and then expecting that their life will become normal. Optimism for the future is very important. However, no one yet knows either the ultimate impact on those with various symptoms and stages of progression, or the long-term effects of the treatment on anyone who has Fabry's.

In many ways, learning to live with a chronic illness is similar to the process of grieving. This article will focus on two stages involved in the grief process, denial and acceptance. Both positive and dysfunctional denial will be discussed. In the next article, suggestions will be given for moving forward towards adjustment and acceptance. For detailed information about the complete grief process of denial, anger, bargaining, depression, and acceptance, please see the short version of the article, "Learning You Have Fabry's Disease: Reactions and Feelings" in the Issue 2 newsletter from 1997, or the long version of the article on the Fabry Support and Information Group (FSIG) website under "Additional Online Information."

The Importance Of The Grief Process

When first diagnosed you may have been relieved to end the uncertainty of your symptoms in spite of the fact that the full implications of your diagnosis might not have been understood. Alternately, you may have felt little or no relief at a diagnosis and been overwhelmed by sadness, perhaps even finding yourself crying quite often. There is no set response style since people respond to a diagnosis, as well as continued diminished health, in different ways. For example, some may choose not to talk about things even with family and friends, while others want to talk to as many people as possible.

Whatever the initial reaction to a new diagnosis or symptom, health complication, or negative lifestyle change, if you have the cognitive maturity to understand both what has happened and what may occur, resulting feelings can be remarkably similar to intense grief. Your attention eventually will shift to the things that have been or may be lost. The process of acknowledging and coming to terms with our losses is known as grieving, the emotional suffering felt after a loss of some kind.

Grief is usually thought of as something experienced during times of death. However, when you are diagnosed with Fabry's, or when your health continues to diminish, major loss is experienced. This traumatic stress has an effect very similar to grieving. When Fabry's is involved, you grieve because of what you have lost, or may lose, including health, normal routines, future opportunities, and all the things you might no longer be able to do. You may experience additional losses as time passes such as being unable to work, or being avoided by some family members or friends. The type of loss may change over time, but any loss that is personally significant may cause you to re-experience the grieving process. It is important to remember that the grief process is normal, predictable, and even necessary when any personally significant loss occurs. Grieving may eventually lead to the replacement of over-whelming emotions with more manageable feelings and is the process that enables you to finally acknowledge and come to terms with loss.

Denial

A diagnosis of Fabry's represents a significant life change. At first, you may deny the diagnosis and think, "Not me" or "This can't happen to me." Those are common reactions in the early stages of any chronic illness. Even when you have known about the disease for quite some time, when you experience a new symptom, health complication, or negative lifestyle change, you also may face denial. You are not ready to deal with the loss of your health and any changes so you deny the problem. It is important to remember that both the person with Fabry's, as well as loved ones, may go through the process of denial. The amount of time that reactions such as denial take are determined by factors such as one's age at onset of symptoms, personality factors, prior exposure to crisis situations, nature and severity of the illness, extent of social support network, and available resources.

Denial is like mental numbness. We are not quite ready to grasp the full implications of what has happened so we dismiss it, rationalize it, minimize it, or question the truth. Some completely deny that they have Fabry's, choosing to struggle through daily routines until they are unable to function any longer. Others may admit they have the disease yet deny limitations, or the seriousness of the illness. You may tell yourself things such as, "So what if I have Fabry's. It's no big deal." "I won't be sick until I'm much older." 'I've felt tired lately, but it's not because of Fabry's." "Now the doctor is telling me it's causing heart problems. Those can't be my test results." Some may deny the disease in their actions as well. For example, they may continue to be active in outside heat even when this causes more pain.

Because Fabry's often has invisible symptoms, it is at times easy to deny. In fact, these invisible symptoms can make it an extremely difficult disease for others to grasp and understand. Overall, denial usually does not last long when you have the disease because sooner or later, the limitations imposed by Fabry's can no longer be ignored. It is not easy to admit to yourself that you are suffering from a chronic illness, but it is an important milestone if you are to end up adapting to new circumstances.

Positive Denial

Although denial has a negative connotation, it is a normal defense mechanism and can play a positive part in coping with Fabry's for both you and your loved ones. It is perfectly normal that you should sometimes find it difficult to admit having a disease, various symptoms, complications, or limitations. Denial provides a respite because for a short time, it permits you to set worries aside and function. It gives you time to deal with things such as a problem or fear, and come to terms with bad news little by little. Denial can be an initially positive coping strategy as long as it does not interfere with proper treatment, self-care, or relationships.

Dysfunctional Denial

Even though denial initially can play a positive part in coping, it will eventually cause problems if it turns into a pervasive condition. When you continue to deny your feelings and the grieving process, you postpone the maturation of your emotions and hinder progress toward a happier future. You may superficially accept the diagnosis, but remain unwilling to learn about Fabry's, or think about the need for adjustments. It is detrimental when your emotional or physical status is denied for too long. It could even end up with you refusing to accept help that is necessary. For example, when someone refuses to use equipment that could improve mobility it could result in being isolated. Coping with Fabry's becomes more difficult if you persist in denying that you are ill and go on stubbornly trying to do everything you were once capable of doing. For example, if you believe that you are a fully productive employee, yet you are not, denial can be detrimental. Denial even can take a dangerously defiant form. For example, someone in denial may ignore his or her doctor's advice that could help keep the disease under control. Denial may cause symptoms to be under-reported, or attributed to some other less serious cause.

Statements like "There's nothing wrong with me" or "The doctor made a mistake" arise from the wish to take control over your life again. However, if this attitude becomes pervasive, it blocks the process of coming to terms with and adapting to the illness. While it is not uncommon to be resistant to limitations, it is far better to define a new more personally adaptive lifestyle. The difficulty can be keeping denial in balance while working towards adaptive responses.

When the person with Fabry's continually engages in dangerous and self-destructive behaviors, perhaps as a result of denial, someone may need to intervene. Speaking with the person in a straightforward and honest way about concerns may be necessary. Pervasive denial can become extremely difficult to watch, yet even a loved one cannot force important changes.

Adjustment

Fabry's is not going to go away regardless of how much anger, sadness, denial, etc. there is. People who have the disease eventually must accept their situation if their life is going to be happy and fulfilling. Each person will adapt or adjust to Fabry's differently and at his or her own pace. Adjustment is a process of a succession of situations requiring specific solutions and not necessarily an orderly process. There will be emotional and physical ups and downs in living with the disease. Feelings that are overpowering today may not be perceived that way tomorrow. Your medical condition is unpredictable and emotions may change with your condition. Not only will you need to learn to deal effectively with emotional responses, it is important to remember that some family members will also find adjustment difficult.

How well you cope with Fabry's will determine, in great part, the quality of your life. Of the three primary factors which measure your ability to cope: your attitude, the social context of your life, and the quality of resources available to you, your attitude becomes the foundation upon which the others build. Undoubtedly, some people adjust better than others to the fact that they have a chronic illness.

Conclusion

Denial and acceptance are normal steps after being diagnosed and as the disease progresses causing more changes in your health and lifestyle. While long-term pervasive denial is likely to be detrimental to physical and emotional health, temporary denial can be a beneficial coping mechanism in moving towards acceptance of Fabry's as a part of your life. When you begin to accept Fabry's, you open yourself up to see possibilities and opportunities in your life, perhaps much different than those you had hoped for or expected. When denial continues, you close yourself off to those possibilities and opportunities. Accepting illness is a process, a personal journey. The process is not easy, does not happen all at once, and it is not something you do once and never again. In addition, it does not mean that you are ever happy about having Fabry's. Acceptance means that you have made room for Fabry's in your life and that it is just one aspect of an otherwise happy and fulfilling life.

We Have The Technology - You Have The Choice

Atul Mehta, Consultant Haematologist Royal Free Hospital

While medical science is not yet quite at the 'Six Million Dollar Man' stage, exciting advances are being made which hold the promise of dramatically improving quality of life and life expectancy for sufferers of lysosomal storage disorders (LSD). As always, these advances have come at a price - and in Europe and the USA, the regulatory authorities have introduced the concept of 'orphan drugs'. Orphan drugs are designed for treatment of conditions where there are only a modest numbers of sufferers; pharmaceutical companies are given incentives which allow them to invest research and development funding with a realistic prospect of making a profit for their shareholders. The recent turmoil in the financial markets illustrates the rough and tumble of the unpredictable world in which these companies survive.

Enzyme replacement therapy (ERT) was first introduced for the treatment of Gaucher's Disease, the commonest LSD, over ten years ago. It is no understatement to say that it has had a dramatic impact on the life of Gaucher's sufferers. It has not only improved and extended life for adults with Gaucher's disease but has led to an explosion in interest, and new money for research, in LSD. Fabry disease is the second commonest of the LSD. There are thought to be perhaps three to five hundred Fabry sufferers in the UK, with perhaps five to ten thousand worldwide. The success of ERT for Gaucher's disease has attracted other pharmaceutical companies into the field and two enzyme preparations underwent trials during the late 1990s. In August 2002, the European regulatory authorities licensed two preparations for enzyme replacement therapy of Fabry disease. These preparations are Replagal (manufactured by TKT Inc. based in Boston USA who have a subsidiary European arm, TKT 5S based in Stockholm, Sweden) and Genzyme Corporation (also based in Boston USA, the makers of Cerezyme, which is the established formulation used in treatment of Gaucher's disease). ERT for Gaucher's disease has never been rigorously evaluated with trials. If you were to talk to a Gaucher's sufferer (we regularly do - we have over 60 patients registered at our centre, nearly all ofwhom are on ERT) she will tell you that such trials are completely unnecessary. The treatment works - period. The mantra of modern healthcare, however, is 'evidenced based medicine' and the gold standard is the randomised, double blind placebo-controlled trial. Both companies recognised that they would not be able to have their products licensed without the rigorous trials - indeed the Food and Drug Administration in the USA is still deliberating and has yet to grant a license to either of these products.

Patients in the UK have had an opportunity to participate in these trials. At the Royal Free Hospital, Dr. Kay McDermott recruited 15 male Fabry sufferers into a randomised placebo controlled trial of Replagal and our centre has gone on to recruit further patients into open label trials and compassionate use studies such that we now have over 40 patients undergoing treatment. Dr. Steven Waldek in Manchester and Dr. Philip Lee and colleagues at University College London, have recruited patients into similar trials of Fabrazyme therapy. The results of both trials have been published in prestigious American medical journals and have been reviewed by experts, healthcare providers and financial gurus worldwide. Both enzyme preparations work and are safe to use. The detailed design and outcome measures of the trials were rather different, but it is clear that treatment with either of the preparations will reduce the blood and tissue level of the storage material that accumulates in Fabry disease.

How is it that two orphan drugs have come to be licensed for such a rare condition?

I can only guess at the answer- I suspect that neither of the two pharmaceutical companies knew precisely which ERT treatments the other was developing. From the perspective of the regulatory authorities, since the two preparations have not been compared in a direct head to head fashion, it is impossible to say which is the 'better' of the two and a sensible compromise was to license both and thus allow patients to benefit from early treatment. The introduction of competition into this specialised area can, to my mind, only offer advantage to sufferers, families and professionals. It has led to a vast increase in research funding and other companies have now come in to the field so that new treatments are already undergoing trials for other LSDs. Ultimately, competition may even lead to a reduction in price. From the investigators' perspective, we were very excited at the prospect of using Replagal, as there are significant differences between it and Cerezyme. Both the Genzyme products, Fabrazyme and Cerezyme are made in mammalian cells (Chinese hamster ovary cells) whose genes have been manipulated to make large amounts of human protein. This protein then undergoes modification within the mammalian cells. Some additional modifications are made to improve the targeting of the enzyme so that it is taken up by affected cells. In contrast, the TKT drug Replagal is made by a new and different technique which uses human cells. The argument is that human cells will modify the human protein in exactly the same way as normal cells do. This should mean more accurate targeting of the therapy to affected tissues. Whereas in Gaucher's disease the affected tissue in adults is largely one cell type (the monocyte/macrophage) in Fabry disease excessive accumulation occurs within a range of cell types within a diverse range of tissues - principally the kidney, heart, skin, blood vessels and nervous system. This is why Replagal is given at a much lower dose than Fabrazyme and has a much shorter infusion time. We need to establish conclusively whether these two different drugs to have different effects.

What does all this mean for patients and carers?

You have patiently waited a long time but all of this is undoubtedly good news. Both treatments have been rigorously tested and are now licensed. As with any new treatment, further testing will be necessary to establish safety and effectiveness. We would urge all Fabry sufferers and their families to be evaluated at one of the treatment centres. By starting treatment early, it may be possible to avoid some of the long term effects of Fabry disease (renal failure, heart failure, stroke, premature death). This applies to both women and men in families in whom there are documented Fabry sufferers.

What does this mean for the centres?

The doctors and their teams at each of the centres have experience in evaluating and treating this rare condition. It is not possible for every district general hospital in the country to have a resident Fabry expert and some travelling on the part of patients will be necessary. The treatment, however, can be delivered to the patient's home and can be administered either within the home or at the local hospital. Each of the centres will need to make provision for shared care arrangements with local providers. The centres will also need to gather, after obtaining appropriate consent, further information on the safety and effectiveness of the treatment. The centres are working with the Department of Health to develop a registry of Fabry sufferers throughout the UK. We are also working with health authorities to encourage equal availability of the treatments throughout the country avoiding 'post code prescribing'. We are establishing a national forum with national written guidelines on assessment and treatment of Fabry sufferers so that all sufferers throughout the country should get the same standard of care regardless of where they attend.

Ultimately, however, you have a choice. There are two products available and they are different. The centres should be able to explain these differences to the sufferer and their family. Each of the centres should be able to prescribe either of these two products (in other words both products should be available on the hospital formularies of each centre). Each of the centres currently depends on support from one, other or both of the pharmaceutical companies and all of us are keen for the Department of Health to provide the support instead.

The future beckons. New enzyme treatments are being developed for other conditions. Competition means that the companies will want to further develop these treatments. Other companies are developing tablet treatments which show promise. Gene therapy may be available within the next five years - although, it has to be said, this is what we have been hearing for the past ten years! The Labour Government is allegedly keen to eliminate post code prescribing and improve healthcare provision for all - although frankly, those of us at the sharp end still feel that they need to put their money where their mouth is. If we in the NHS had a choice between Gordon Brown and Sir Alex Ferguson who do you think we would choose? For the cost of one Rio Ferdinand, we could treat all the Fabry patients in the country.

BioMarin and Genzyme Complete 'Rolling' BLA Filing For Aldurazyme

Novato, CA and Cambridge, MA, July 29, 2002 – BioMarin Pharmaceutical Inc. (Nasdaq and Swiss SWX New Market: BMRN) and Genzyme General (Nasdaq: GENZ) today announced that the companies submitted the final portion of their 'rolling' Biologics License Application (BLA) for Aldurazyme (laronidase) to the U.S. Food and Drug Administration (FDA). The final portion of the BLA includes clinical data from the six-month, placebo-controlled Phase 3 trial of Aldurazyme, six months of data from the ongoing open-label Phase 3 extension study, and three years of data from the Phase 1 trial and extension study. Aldurazyme is an investigational enzyme replacement therapy for patients with Mucopolysaccharidosis I (MPS I), a life-threatening genetic disease for which no specific drug treatments currently exist.

As part of the BLA submission, BioMarin and Genzyme have formally requested Priority Review, which is an FDA procedure generally reserved for products that address an unmet medical need. The companies expect a decision regarding FDA's acceptance of the Aldurazyme BLA and its Priority Review status by the end of September 2002. If the FDA accepts the BLA and grants Priority Review, the companies anticipate a response from the FDA regarding the application to market Aldurazyme in the United States six months from the submission date of the completed application.

On March 1, 2002, BioMarin and Genzyme submitted a Marketing Authorization Application (MAA) to the European Agency for the Evaluation of Medicinal Products (EMEA) for approval to market Aldurazyme in the European Union. The Agency has accepted the MAA and is currently reviewing the application. BioMarin and Genzyme expect a response from the EMEA in the first half of 2003.

1st International Patient Symposium On Fabry Disease Barcelona, 22 - 24 November 2002

In November there is going to be an international meeting for patients with Fabry disease to get together and discuss some of the important medical issues surrounding this disorder.

The event takes place from 22 - 24 November 2002 in Barcelona, and the organisers have offered to pay travel and accommodation expenses for sufferers.

We have already invited Fabry centres throughout the UK to nominate participants, however, if you are a Fabry sufferer and would like to attend please do contact the MPS Society.

The Rare Disorders Alliance And The National Service Framework

Christine Lavery - Director

The Rare Disorders Alliance (RDA) is a network of groups and individuals who support the aims of the alliance which include sharing good practice, promoting the well being of those affected by a rare disorder and to act as a consultative body on all policy issues relating to rare disorders. The European Union's definition of a rare disorder is a condition that affects 5 or less people in every 10,000.

MPS has been involved with the RDA since its conception and still plays an integral part in ensuring that information is shared and issues are raised. The RDA is presently keeping a close eye on the National Service Framework (NSF). There is no NSF for rare disorders and MPS and the RDA are battling to ensure that children and adults with a rare disorder are included in the NSFs. The Steering Committee of which Christine is a member organised a meeting and invited Professor Al Aynsley-Green, Professor David Hall and Professor Bryan Winchester to speak. They offered advice on how to lobby ministers and how to progress the cause. However, sadly neither Professor Aynsley Green nor Professor David Hall felt able to support the call for a separate National Service Frameworks for rare diseases.

It is important that families who have issues that they would like to raise regarding the diagnosis, clinical management, treatment and therapy of their family member who has an MPS or Related Disease pass details to the Society so that in an anonymous form these can be fed back.

Coronary Artery Patency Following Long-Term Successful Engraftment 14 Years After Bone Marrow Transplantation In Hurler Syndrome

Braunlin EA, Rose AG, Hopwood JJ, Candel RD, Krivit W; University of Minnesota Medical School, Minneapolis, MN, USA

Clinician's note: Mucopolysaccharidosis type I (MPS I) results from deficiency of the lysosomal enzyme a-iduronidase. Phenotypic presentations vary between the mild form, MPS I Scheie, the intermediate MPS I Hurler/Scheie, and the severe form, MPS I Hurler. In the latter, deposition of glycosaminoglycans within the myointima of the epicardial coronary arteries causes heart failure, and is responsible for death within the first decade of life.

In this case report, the coronary arteries in a patient with the Hurler syndrome appeared only minimally affected by the deposition of glycosaminoglycans 14 years after successful bone marrow engraftment. This finding adds to the previously reported salutary effects of transplant on large airways, cardiac function, hepatic morphology, and central nervous system function. Enzyme replacement therapy for MPS I is currently at the clinical trial stage. The possibilities and limitations of therapy with either bone marrow transplantation or enzyme-replacement therapy need to be further elucidated in the future.

Taken from Lysosomal Storage Diseases
Vol 2 Number 2, 2002

Significant Medical Breakthrough In Mucolipidoses, Secondary Metabolic Bone Disease

Jenny Noble

In being asked to write an article for your newsletter, I wonder where I should start. There is so much to write about, but I think a little background is needed and then onto treatment for Secondary Metabolic Bone Disease.

Hayden and Sarah have ML3 and were diagnosed in 1987/1988. We went through many years of medical intervention as they progressed through the process of deterioration, but the one major area we could never find an answer to was constant chronic pain.

By age 15 Hayden was in a wheelchair due to cell death in the nervous system and was diagnosed and a paraplegic, he was also in terrible pain. We tried many different kinds of anti-inflammatory drugs with little or no effect.

By age 13 Sarah was finding it very difficult to walk and was losing her mobility very fast. We began to think that perhaps she was heading along the same path as Hayden was, although her pain was much more intense, and once again could not find a drug that would deal with pain.

Being a parent who has never accepted that this was just part of the disease I set out to find an answer. While attending an Australian MPS conference two years ago we asked Dr. Ed Wraith and Prof. David Sillence to have a look at Sarah and give us their expert opinions on what they thought might be happening.

From this meeting it was agreed that Sarah needed a full medical work up, as all her tests did not show spinal cord compression. So after returning home Sarah and I headed back to Sydney where we spent a week going through the process of elimination. By the end of the week it was discovered that Sarah's bones were in a terrible state, and it was no wonder she had so much pain. It was also felt that Hayden's bones would probably be in the same condition.

Prof. Sillence recommended that we trial a drug called Pamidronate which he has used in OI children with amazing results. We were told that if Pamidronate was going to work in these ML3 patients then one would expect to see almost immediate change.

Because Pamidronate had not been used in MPS patients before, we agreed to enter into a trial using Pamidronate and would document the changes as they happened. We also kept a pain diary, which was of no use, as both Hayden and Sarah were pain free two weeks after the first infusion. Also part of the trial, Bone Biopsies were taken before treatment and 12 months after treatment. The biopsies were used firstly as a base line and secondly to check that changes were taking place in their bones. I am pleased to report that the biopsies show that in both young people their bone density has returned to within normal range. Although they still continue to lose bone at the outer surface.

The changes in both young people have been truly incredible. We have seen Sarah go from a wheelchair to a walking frame, to crutches, to walking unaided. She can get down to the floor, wash her own hair, climb stairs, smiles and now enjoys life. Hayden has only just started to walk, and after 5 years in a wheelchair there is a lot of work to do on him. He really has to learn to walk all over again. He is now able to tell us when his feet are hot or cold, and just this week knows where he is putting his feet when walking, he has so much more feeling in his legs, and has gone through 2 winters without chest infections.

Life for these two young people is so much more enjoyable. Pamidronate has stopped the pain, is building bones and I truly believe it is making other changes to their pathology, as Hayden really should not be walking at all.

Both young people have now completed 2 years of treatment and continue to have infusions at a much larger dose in the hope that we can slow down bone loss to the outer surfaces of bone.

Because of the amazing results we have seen in these two, and in some of the ML2 children in Australia, David Sillence wants to carry out an International trial using Pamidronate, so that we have an International protocol in the treatment of secondary Metabolic bone disease in ML, and well documented use of this drug. If there are any families who have ML children we would like to hear from you.

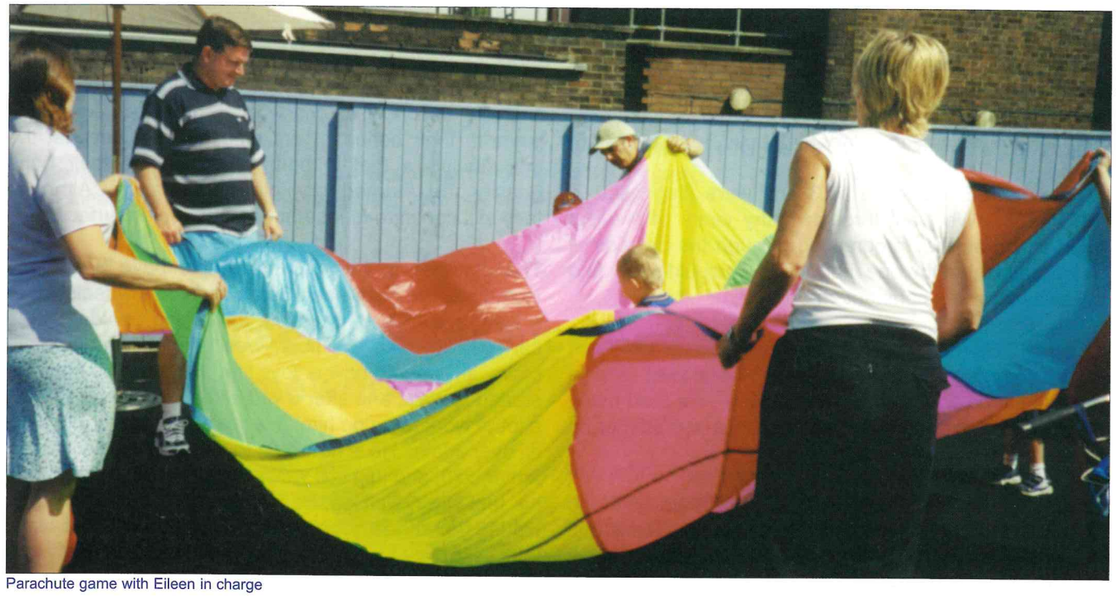
I can be contacted by e-mail jenny.noble@clear.net.nz or by phone +64 3544-5359. Please also look at the LDNZ website www.ldnz.org.nz Our story is to be published there with more detail than I can give you in this newsletter.

Look forward to hearing from you all

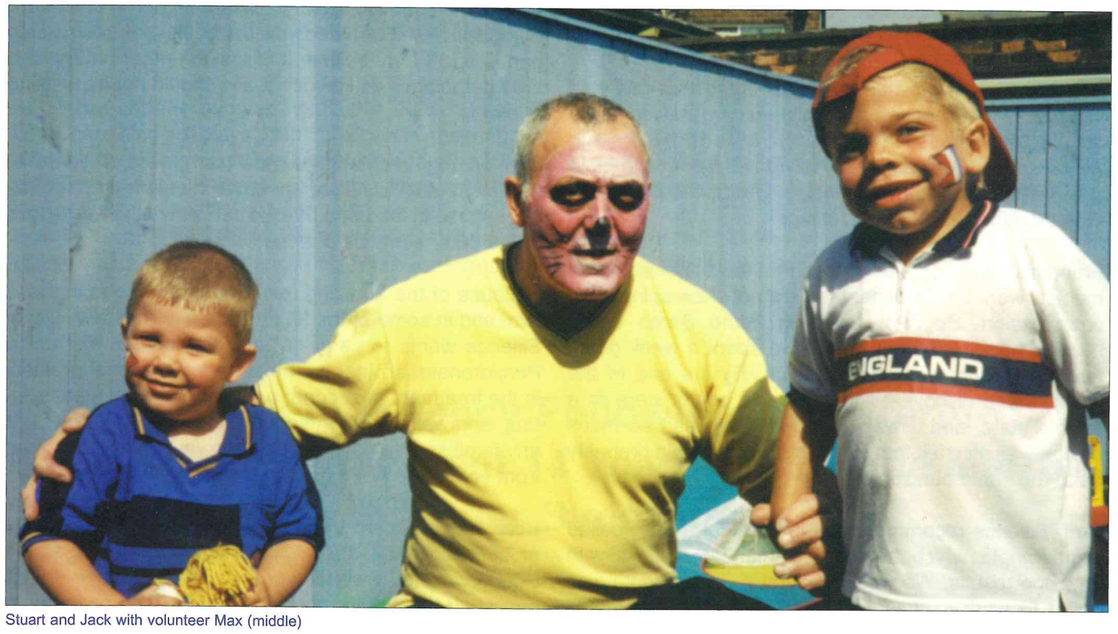
Jenny Noble
Parent of ML
Trustee LDNZ

Kaleidoscope Summer Social

Helen Heard



Parachute game with Eileen in charge



Stuart and Jack with volunteer Max (middle)

For those who came along to the Kaleidoscope in Scunthorpe, on Sunday 28th July, it was one of the hottest and sunniest days of the year. There was fun for all the family with the parachute game, soft play area, sand pit and face painting. The childrens' smiles seemed to match the weather, and the only problem of the whole event was how to make shade so no-one over heated! Some of us secretly hoped the fire station next door would need to do some training and spray their hoses over the fence at us to cool us down, but no such luck!

Skilful planning from Angela Seymour meant that raffle, food, entertainment and decorations were all so well organised that it seemed a seamlessly smooth transition from arrival to departure.

Thanks to Angela and family for all their hard work, to Peter and also to the tireless efforts of Eileen Smale (especially out there in the hot sun flying the parachute) and Max Howlett (with his scary but fabulous face painting), who kept the children (and the parents) busy, happy and entertained.

I hope you enjoy the photos as much as I enjoyed being part of this relaxed and special afternoon.

Family Social Event in Scunthorpe

Angela Seymour angie@seymour.freeserve.co.uk

Sunny Scunny as it is commonly called, really lived up to it's name on the 28th of July, the day we arranged an MPS family get together. But as the temperature soared we realised that there was going to be a greater need for shade in the children's play area. We eventually had three parasols for those who wanted to keep cool in the shade, it got so warm that the parachute that was being used for the children's entertainment was eventually pinned up to give the children playing in the sand pit some respite from the sun.

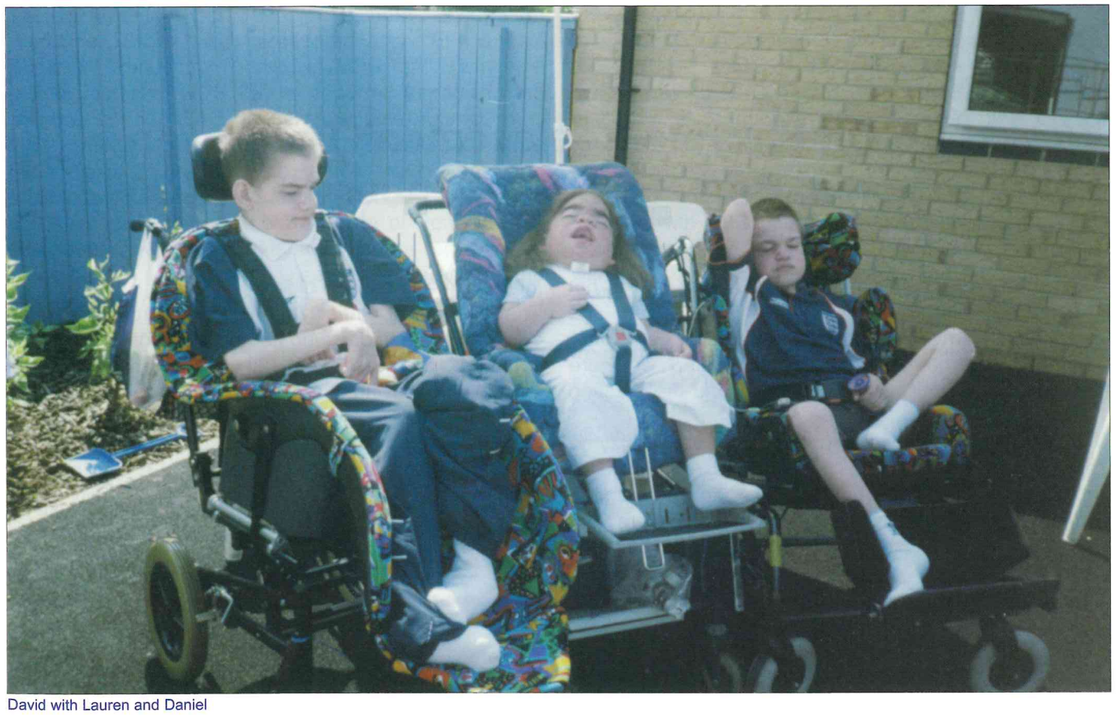
There were about eight families and some friends totalling about fifty people. Helen from head office also attended with her daughter. Before long old friendships were renewed, new friends were made, gossip was caught up on, new ideas were exchanged on the management of MPS, and laughter could be heard from the children, a sure sign that everything was going well. About an hour after arrival we all participated in a superb buffet and some delightful desserts.

We were very fortunate to have our two local MPS volunteers Eileen Smale and Max Howlett to help to entertain the children. Eileen is a dab hand at face painting, but this time the painting wasn't only done on the children, several daft parents also had their faces painted, by other parents, much to the delight of the children!

A raffle was also held, just to make sure that if we went over budget we could make up the difference; I had to rescue a two pound coin from the mouth of a beautiful 10 year Sanfllippo called Natalie whilst I was selling tickets.

Jack and Lewis

I should have known better, having a Sanfllippo son myself, how could I forget how fast their little hands move. She was also very partial to wrapped sweets, so apologies to anyone who opened a Quality Street with teeth marks in it, but Natalie may have got there first. Before we knew it, the time ticked away and it was four thirty. Time for us to say our goodbyes, and exchange phone numbers and addresses. Nobody really wanted to leave, but the children were exhausted from the heat and several had an hour or so to travel. We were all very grateful to the Society for funding the event and hope to be able to attend many more.



David with Lauren and Daniel

6th International COGENT Conference, Modern Management Of Hurler's Syndrome 14th - 15th September 2002

Antonia Crofts

During the weekend of the 14th and 15th September 2002 Christine had been invited to attend this conference and speak on the subject of planning and co-ordinating the long term follow up of post Bone Marrow Transplant patients. I was lucky enough to go too as Conference Co-ordinator for the organisers, COGENT.

Due to a prior event, this conference was being held in Vitznau, Switzerland and meant that Christine and I would need an early flight on the Saturday morning in order to be there for the start of the session. Although we arrived in time for the Zurich flight we did not have as much time to spare as we had planned. On the M25 a certain speaker discovered that she had forgotten her talk for which we had to make a slight detour and return home!

Having arrived in Zurich safely Christine and I met up with several doctors arriving from Manchester who were also taking part in the conference. A taxi took our group on a scenic route across Switzerland to the hotel, located in the picturesque village of Vitznau on Lake Lucerne.

The timetable for the conference was well planned and chaired by Dr Colin Steward from Bristol Children's Hospital. Saturday afternoon was devoted to the general aspects of Hurler disease. After an overview of the disease from Dr Ed Wraith various doctors from Royal Manchester Children's Hospital, the Institute of Child Health in London and the University of Minnesota in the United States gave talks on pertinent issues such as genetics, anaesthesia, and pulmonary complications associated with Hurler Disease. The afternoon session concluded with an update from Dr Wraith on the International Trials of Enzyme Therapy in MPS I.

Sunday morning talks were more specific in nature touching on the neuropsychological effects post Bone Marrow Transplant and the future of transplantation and cellular therapies. Adam and Louise Turner, whose son, Isaac, has previously undergone two BMTs, gave a wonderful talk based on their perspective as parents and the particular problems they have encountered. Unfortunately, the other parent Linda Pack was unable to make the conference and agreed that Christine give her presentation. Christine concluded the conference with a talk focusing on the need for comprehensive psychological follow up for children post BMT.

By the end of the weekend all those attending had felt that the conference had been a very worthwhile success in enabling a number of professionals involved in working with those with MPS I to get together and discuss the modern management of Hurler Disease. We extend our sincere thanks to all who participated and to Dr Colin Steward and Dr Charles Peters for arranging it.

Workshop On Enzyme Replacement Therapy In Fabry Disease, Barcelona September 13 - 14 2002

Angela Ratcliffe

After a very long delay at Heathrow airport due to not one but two planes having trouble Ellie Gunary and myself finally arrived at the hotel in Barcelona at 11pm.
The following morning after an early start we were welcomed in Spanish and Catalonian and the meeting began.
We heard talks on the importance of early enzyme replacement therapy and Fabry disease in children among others and as part of the afternoon's programme 4 doctors presented case reports all of which were very interesting and easy to digest.
That evening at dinner we celebrated Professor Bryan Winchester's birthday who had hoped to get through the evening without any fuss but was serenaded by the Spanish trio playing at dinner and then presented with a cake.
The following day the theme running through most of the talks was Fabry disease in females. We also heard 4 case reports on ERT in Fabry disease and quality of life. Again this was very interesting and informative.

International Conference Offers Ideas Insight For MPS Community

An article from, Courage, the USA MPS Society newsletter

An important part of the International Conference is the International Working group where MPS Societies from around the world submit posters and presentations on the programs they offer in their respective coutries.

Canada strives to meet their members' needs in such a large, unpopulated country by using regional contact families and regional or chapter events, as well as a toll-free telephone number to the Society's office. France addresses the respite needs of their members by organizing annual vacations for families at popular destinations, providing child and medical care so families can be relieved of that responsibility. Germany discussed the tremendous children's activities they provide at their annual conferences. Volunteers take total care of the children leaving the parents free to enjoy and benefit from the conference. Italy uses national TV media, the Internet, conferences and newsletters to increase awareness of MPS, including a National MPS Day. Norway discussed how they use a holiday facility to provide families with a vacation while providing medical care and consultation with MPS specialists. The UK described their comprehensive advocacy program that has benefited more than 1, 000 people including befriending program for members and an annual remembrance day. Our own Unda Shine concluded the session by discussing the process used by the United States to overcome the hurdle of hiring and funding a paid staff.

The scientific meetings began with presentations about animal models and preclinical trials in animal models. Dr. Steven Walkley from Albert Einstein College of Medicine, Bronx, NY, presented the "Efficacy of Substrate Reduction Therapy in MPS IIIA". Using the mouse model of MPS IIIA he reported storage in the neurons of non-GAG (heparan sulfate) compounds, GM2, GM3 gangliosides (fattyacids) and free cholesterol. Treating the mice with an oral pharmacological agent reduced the ganglioside storage. Dr. Mark Haskins from the University of Pennsylvania presented an eloquent keynote lecture on "Animal Models of Lysosomal Diseases" detailing the identification of animal models of lysosomal storage diseases. During the session on prediction of severity of phenotypes from molecular data, Dr. Arnold Reuser from the Netherlands reported that differences in genetic background, specifically the effects of our approximate 35,000 other genes, can affect the clinical findings in MPS if the amount of enzyme produced is close to the critical threshold. Dr P J Meikle from Australia reported positive results from a two-tier screening strategy to identify newborns at increased risk of lysosomal storage diseases, the incidence of which is approximately one in 5,000 births. "The Burden of Illness in MPS I" was presented by Dr. Joseph Muenzer from Chapel Hill, NC. Based on clinical data obtained from the 45 individuals in the phase 3 enzyme replacement clinical trial, it was concluded that MPS I causes numerous medical problems in multiple organ systems and results in significant disability even in individuals with mild to intermediate forms of the disease. Dr. Briony Gliddon from Australia reported a decrease in aggressive behavior and improved learning in MPS IIIA mice treated from birth with intravenous injections of the recombinant enzyme. Using adeno-associated virus medicated gene delivery in MPS III-B mice, Dr. Haiyan Fu from Chapel Hill, NC, has demonstrated long-term and progressive reduction of GAG storage in body tissues. Dr. Joe Clarke from Toronto presented results from "A Pilot Study of the Effect of Glucosamine on the Behaviour of Children with MPS III." He suggests that since glucosamine is a major component of heparan sulfate (HS), treating with D-glucosamine HCi may block the binding of HS. Although there was no effect on hyperactivity, improvements were seen in restless/impulsive behaviours and emotional liability. Drs Wraith, Muenzer and Harmatz presented the Enzyme Replacement clinical trial results for MPS I, II and VI.

An afternoon of sessions for both parents and scientists on advances in clinical management and preventative therapy of MPS and Related Diseases began with Dr Rob Walker from England presenting on anaesthesia for children with MPS. Dr Grace David from Australia showed evidence of increased function and improved quality of life in a preliminary study of individuals with ML II/III treated with cyclic intravenous Pamidronate (bisphosphonate). Dr P Collignon from France reported that pain is common in MPS disorders and can be caused by spasticity, orthopaedic troubles, hydrocephalus and recurrent infections. Dr Ed Wraith from England gave an overview of "Sleep Disturbance in Patients with MPS." One cause of sleep disturbance is obstructive sleep apnea, found frequently in children with MPS I, II and VI, caused by GAG deposits and enlarged tonsils and adenoids. Children with MPS III have two patterns of insomnia: difficulty settling and frequent waking. Management of sleep disturbance includes behavior and environment modifications, respite and a wide variety of medications. A gala banquet was held the last night, and credit was given to the many individuals responsible for this wonderful meeting, including the primary organizers from the U.K. and French MPS Societies. The 8th International Symposium on MPS will be held in 2006.

Benefits Boosted

Two new social security measures to be introduced this month will benefit disabled people and carers.

The earnings of severely disabled people will now be completely ignored in assessments for the Independent Living Fund (ILF). Earnings of partners will also be disregarded.

Maria Eagle, Minister for Disabled People, said severely disabled people could gain as much as £130 a week. There will also be large increases in the amount of savings that are ignored in an ILF assessment and an increase in the upper limit of savings. Eagle said: "All disabled people have the right to live as independently as possible. These measures will give severely disabled people greater financial independence and freedom to choose their own care and support."

April also sees the introduction of a second state pension. The government estimates this will help two million disabled people and two million carers, who will now be able to build up credits for a second state pension as well as a basic state pension.

Book: Fathers' Grief When A Disabled Child Dies

In this study, eight fathers who had suffered the loss of a child with a serious disability were asked about their reactions.

Many of them felt they had experienced a double loss: first, when they knew that their child was disabled, and a second time when the child died. All the fathers believed that their loss was different from that of fathers whose children did not need special care.
Each of them had their particular methods of coping with the problems of living with a disabled child and of coming to terms with the child's death.

Consistent with the literature on gender differences in bereavement, fathers reported greater emotional stoicism and the value of keeping active, rather than talk or social support, as a primary coping strategy. Clinical applications for professionals working with grieving men or with the parents of children with special needs are discussed.

Civil Rights For Disabled People

A draft bill that sets out a comprehensive list of demands for civil rights for disabled people has been produced by a working group within the disability umbrella organisation Rights Now.

The British Council Of Disabled People (BCODP), who played a major part in the drafting of the bill, wants the Disabled People's Rights and Freedoms Bill to replace the Disability Discrimination Act (DDA), which it says is flawed.

The draft bill seeks to outlaw discrimination over a much wider area than the DDA and also says that disabled people should have the right to live independently, rather than in care homes. For more information see

www.bcodp.org.uk

Community Fund

In 2001/2002 the MPS Society was successful in securing the second year of a three year grant totaling £21,176 to fund a regional clinic, an individual advocacy support service and information days in Scotland.

Direct Payments For Carers, 16 and 17 Year Olds And Parents of Children with Disabilities

The Carers and Children's bill came into force in April 2001. This means that local Authorities can now offer Direct Payments to:

-parents of children with disabilities for services for their children
-16 and 17 year old disabled young people in their own right
-carers aged 16 or over for carers services

What are Direct Payments?

Direct Payments are a cash payment given to people by Social Services instead of community care services; Their aim is to give them more choice and control over their lives. It enables people to choose who, when, how and where they get assistance from. The money is not extra income. It must be used to pay for services that meet people's assessed community care needs. It is not extra benefit and it will not be taken into account as income for other benefits.

What can Direct Payments be used for?

Direct Payment users can use this money to pay a member of staff to meet their needs. They are in charge of who that person is, when they will work and what they do. They can use agencies or employ their own members of staff. (However they cannot employ a close family member). The Direct Payments also cover any other costs like training, administration, payroll, insurance etc. People can use their money from direct payments to get assistance to do an activity instead of going to a day centre or to take short breaks if they have been assessed as needing them.

Choice

People can choose whether to have Direct Payments, or have assistance provided by Social Services. You can have a mixture of Direct Payments and assistance through Social Services. For example a person with a disability can employ their own personal assistant for some of the time and still continue to attend a day centre if they wish.

What can Direct Payments not be used for?

Direct Payments cannot be used to buy residential care, health services or local authority services.

How do we get Direct Payments?

Direct Payments are arranged by a social worker. They will only be offered if people meet the eligibility criteria and they are willing and able to manage them with or without assistance. The User/Parent/Carer must remain in overall control about how the Direct Payment is used. Supporters can assist a person to manage their Direct Payments but cannot claim on another's behalf.

The Department of Health wants to make sure that Direct Payments are available to as many people as possible. They have produced an easy guide to Direct Payments which gives you the basic details of what a Direct Payment is and includes some examples of how you might be able to use one.

If you want free print copies of the easy guide to Direct Payments pack, please write to:
Department of Health Publications
PO Box 777
London SE1 6XH
e-mail: doh@prologistics.co.uk

Campaign Continues

The new Work and Pensions Secretary Andrew Smith has rejected calls to extend the £200 winter fuel payment to disabled people.

The move came as the influential Commons' Trade and Industry Select Committee prepared to publish its report into fuel poverty in September.

The committee of MPs took evidence from various groups calling for the payments to be extended. These groups want the payment to be given to severely disabled people of working age on the middle or higher rate of the care component of Disability Living Allowance, or the higher rate of the mobility component of the allowance.

Pensioner households already get the payment. Extending it to disabled people would cost about £340m.

Enabling all pupils

School Standards Minister Baroness Catherine Ashton explains what the government is doing to improve education access for disabled people.

This year marks great progress in education for disabled people. The Special Educational Needs and Disability Act comes into force in September, which will outlaw discrimination against disabled people in education. The Act gives disabled people equal rights of access to education and opportunities like all citizens.

The changes mean that local education authorities (LEAs), schools and post-16 education providers cannot treat disabled pupils, students and adult learners less favourably without justification. They need to make reasonable adjustments to ensure that disabled people are not put at a substantial disadvantage. In practice, this means that schools, colleges and universities need to keep policies under review and consider adjustments to things like classroom organisation, timetabling of classes and teaching methods.

Over time, LEAs and schools will also need to plan to improve access to schools' physical premises, access to the curriculum and written material in alternative formats.

To help make mainstream schools more accessible to children with disabilities and special educational needs, we have provided funding through the Schools Access Initiative (SAI). Over the three-year period 2001-2004, £220m is being invested.

The SAI has funded projects such as installing lifts and stair lifts, improving access to toilets, and making other modifications to help those with sight and hearing problems. Since 1996, more than 6,000 schools have benefited. We will also make £172m available from 2002 to 2004 for similar improvements in further and higher education, as well as LEA-secured education and youth service provision.

The SAI is part of a total grant of £8.5bn to improve buildings for all pupils over the same three-year period. We are currently looking at the future allocation of funding following the boost for education recently announced by the Chancellor.

Special needs have also been considered. One of the amendments we have made relates to the "power to innovate", where the Secretary of State can give schools permission to suspend education legislation for up to three years, if such legislation restricts their ability to raise standards. The Secretary of State must not only consider the interests of every pupil affected by a project; they must also refuse permission for projects where there is likely to be a detrimental effect on the education of children with special needs.

Looking to the future, the working group I set up to look at the role of special schools as part of our inclusion strategy is due to report this December. We have also recently consulted on measures that will enable all young people to fulfil their potential. Our proposals included increasing the learning choices open to young people and encouraging schools and colleges to help individual students progress at the right pace. Ministers are considering the consultation results and will announce the next steps in the autumn. They will take full account of the needs of all young people.

Enabling all pupils and students to reach their potential is a fundamental principle of this government.

John Write, Chief executive, Independent Panel for Special Education Advice

The Minister repeats the assurance she gave the House of Lords on 3 July. She says that no Secretary of State would approve a school or LEA project which involves waiving duties under education law if the project is likely to damage the education of children with special educational needs. This is not only welcome; it is absolutely vital.

For the extension to the Disability Discrimination Act 1995, covering discrimination in schools and colleges, which comes into effect this month, contains an enormous gap: the denial of auxiliary aids and services to children with special educational needs is not deemed in law to constitute discrimination.

So, while schools may no longer leave disabled children out of school photos or school trips, they can be denied the support they need in order to learn, and this will not be defined as discrimination.

The government's explanation for this gap is that children with special educational needs already have a clear legal entitlement to the special provisions their needs call for. Under the 1996 Education Act, LEAs have a duty to identify and assess children who may have special educational needs; to issue, when necessary, statements that specify the extra help children need; and to arrange the provisions set out in the statements.

These duties link to create a child's legal entitlement to additional support. Break one link, and the child's entitlement vaporises. So the protection offered by the new anti-discrimination legislation is as thin as the paper it is printed on. This is why the Minister's assurance is vital. New Labour has created a situation where children with special educational needs can no longer rely on the law to protect their right to learn. From now on, they must rely on the Minister's word.

Disabled Living Exhibition

On the 6th November 2002 the Reebok stadium in Bolton (junction 6 off of the M61) is holding a disabled living exhibition, which is specially for children with a disability. It covers area such as well being, mobility, toys and health.

Tickets are free and are available from disabled living in Manchester (0161 2145959) or you can order them on line at:

disabledliving@aol.com

Digital Hearing Aids

The Government has invested £20 million which means 30 NHS Trusts will receive money to provide digital hearing aids and a further 15 Trusts will be able to apply for government money to help train staff to fit the hearing aids.

In this way almost one-third of people in England will have access to modernized hearing aid services by the end of the year. Twenty NHS trusts already fit the new devices under a pilot scheme launched by the Government at the end of 2000.

The RNID is campaigning for all deaf people to get free digital hearing aids.

Fledglings

Fledglings is a small voluntary organisation who find equipment aids and toys for children with special needs, based on their own analysis and parents recommendation. In their current newsletter they highlight a waterproof cape, special straws which hold the liquid in the straw between sucks and a range of toys. If you can't find something you're looking for they may be able to help. Contact: Fledglings 6 Southfield, Ickleton, Saffron Walden, CBIO ITE
Email: enquiries@fledglings.org.uk Tel 08454581124

New GOSH Website

Children will be able to get health advice on a new website recently launched by Great Ormond Street Children's Hospital (GOSH). The site has been designed by experts at the world-famous hospital and the Institute ofChild Health, but is also based on the experiences of children. It is aimed at children aged five to 16, and will have separate areas for young children and teenagers. The website is at:

www.goshkids.nhs.uk

Higher Education Funding

The Higher Education Funding Council for England has announced a £6.6m fund to improve higher education provision for disabled students. The money is to help institutions who currently have little provision for disabled students and will also help create specialist teaching materials and fund support during work placements.

Holiday Information

Travel information packs to London, Wales and South East England are now available to disabled holiday makers. The charity Holiday Care has produced the guides after research showed that disabled people wanted a 'one-stop' shop when planning holiday travel. The guides give details on the accessibility of transport, accommodation and visitor attractions as well as information on hiring equipment,
Tel: 01293774535 Minicom: 01293776943
www.holidaycare.org.uk

Running the London 10km Road Race for MPS

Clare Titcombe

It all started with a New Year burst of enthusiasm along with a new job, a new gym membership and a new resolution to get fit. I saw the London 10km Road Race advertised at the gym and it seemed like the perfect motivation to ensure that this year's New Year's Resolution lasted longer than the usual two weeks. Besides, July was such a long way away, plenty of time to get myself fit.

Step number two was to find a suitable running mate, my friends unsurprisingly all made polite excuses, my boyfriend feigned a serious knee problem so I decided that my unsuspecting brother would be my chosen victim. Several weeks of persuasion later I had worn him down on the condition that no matter how the run went or how unfit he was I had to wait for him at the finish line so he could beat me over it. (Obviously I agreed to this knowing that I was never likely to be ahead of him in the run but thinking it would give me a great excuse for my athletic failings when he sailed past me at the end of the race).

As July approached my New Year's enthusiasm to get fit had begun to wane somewhat but a dawning realisation grew on me that if I found my 20 minute walk to work tiring how was I ever going to cover 10km running. By the end of June I decided that the time for drastic action had come and I subjected myself to a rigorous training programme of runs along the canals of Reading, with occasional and necessary periods of rest and relaxation (nights out) which every top class athlete knows is essential as part of the build up to top level sporting events!

Training along the canals of Reading was sometimes slightly disconcerting given that I had to run past the local rowing club who all appeared enviably sporty and fit as they cruised tirelessly up and down the river whilst I staggered past wheezing like an old lady. Not only did I have to contend with the rowers but also with the three local tramps on a bench who would urge me to run faster as I tottered along at the speed of a weary snail. However by far the worst spectators on my runs were the small children who would frequently outrun me along the towpath just to prove that they could.

I´m not sure my brother´s training was going too well either, as by a week before the race he proudly announced that he could now run for 25 minutes without stopping to walk. He did not look too pleased to discover that he would probably have to run for over twice as long to cover the full distance.

The race day approached, the tramps no longer jeered quite as loudly and I had not been humiliated by an under five for at least a week, maybe I looked just a little bit like a runner now?

On the morning of the race as I made my way to the start line with my brother, we began to get a bit apprehensive as we spotted what seemed to be an overwhelming number of "serious runners" all doing serious warm up stretches in sophisticated athletics gear. It did occur to me that I had misinterpreted the London 10km Road Race as I desperately looked around for the fun runners. As we found our strategic starting position one row from the back of the field, I was pleased to see that we were suddenly surrounded by fun runners.

The run started well, I was feeling quite energetic as we approached the Houses of Parliament and then made our way along the Embankment towards St Pauls, although the sight of the lead runners approaching the finishing line on the other side of the road, whilst we had barely made it halfway round, was slightly off putting. Nevertheless the run was proving less painful than I had imagined, maybe I was distracted from my pain by the various roadside entertainers on the way or the large group of lads in England shirts who stopped halfway to go and buy some crisps and still caught us up 3 km later!

However by the time we rounded St Paul´s to be greeted by the strains of "When the Saints Go Marching In" we were 7km into the run and no longer in the state of mind for musical appreciation. We reached the Embankment once more and I knew we were nearly home and was determined now that we could run the whole way. My brother however had other ideas and was in the middle of arguing the merits of stopping to walk for a bit, having made it this far, when he saw the finish line looming less than 300 metres ahead. He suddenly set off on an incredible sprint finish undergoing a dramatic transformation into Linford Christie spurred on by this welcome sight. I jogged on in his wake and reached the finish line (about 2 minutes later) only to trip over my brother who was prostrate on the floor vowing never to move again.

Well the race was over, we had finished it in 55 minutes and my brother at least had finished it in style. In fact as we made our way through the finishing funnel to collect our medals still on a high from lack of oxygen to the brain I was actually thinking I had rather enjoyed the whole experience. I turned to my brother "that wasn´t so bad" I said. My brother just looked relieved it was over and then said "well at least now I will never have to run round Amersham sports field again"!

The run was a success, the sponsorship money has been collected, I still maintain that I quite enjoyed myself maybe next stop is the Marathon…now I wonder what my brother would say?

Running the London 10km Road Race for MPS

Clare Titcombe

It all started with a New Year burst of enthusiasm along with a new job, a new gym membership and a new resolution to get fit. I saw the London 10km Road Race advertised at the gym and it seemed like the perfect motivation to ensure that this year's New Year's Resolution lasted longer than the usual two weeks. Besides, July was such a long way away, plenty of time to get myself fit.

Step number two was to find a suitable running mate, my friends unsurprisingly all made polite excuses, my boyfriend feigned a serious knee problem so I decided that my unsuspecting brother would be my chosen victim. Several weeks of persuasion later I had worn him down on the condition that no matter how the run went or how unfit he was I had to wait for him at the finish line so he could beat me over it. (Obviously I agreed to this knowing that I was never likely to be ahead of him in the run but thinking it would give me a great excuse for my athletic failings when he sailed past me at the end of the race).

As July approached my New Year's enthusiasm to get fit had begun to wane somewhat but a dawning realisation grew on me that if I found my 20 minute walk to work tiring how was I ever going to cover 10km running. By the end of June I decided that the time for drastic action had come and I subjected myself to a rigorous training programme of runs along the canals of Reading, with occasional and necessary periods of rest and relaxation (nights out) which every top class athlete knows is essential as part of the build up to top level sporting events!

Training along the canals of Reading was sometimes slightly disconcerting given that I had to run past the local rowing club who all appeared enviably sporty and fit as they cruised tirelessly up and down the river whilst I staggered past wheezing like an old lady. Not only did I have to contend with the rowers but also with the three local tramps on a bench who would urge me to run faster as I tottered along at the speed of a weary snail. However by far the worst spectators on my runs were the small children who would frequently outrun me along the towpath just to prove that they could.

I´m not sure my brother´s training was going too well either, as by a week before the race he proudly announced that he could now run for 25 minutes without stopping to walk. He did not look too pleased to discover that he would probably have to run for over twice as long to cover the full distance.

The race day approached, the tramps no longer jeered quite as loudly and I had not been humiliated by an under five for at least a week, maybe I looked just a little bit like a runner now?

On the morning of the race as I made my way to the start line with my brother, we began to get a bit apprehensive as we spotted what seemed to be an overwhelming number of "serious runners" all doing serious warm up stretches in sophisticated athletics gear. It did occur to me that I had misinterpreted the London 10km Road Race as I desperately looked around for the fun runners. As we found our strategic starting position one row from the back of the field, I was pleased to see that we were suddenly surrounded by fun runners.

The run started well, I was feeling quite energetic as we approached the Houses of Parliament and then made our way along the Embankment towards St Pauls, although the sight of the lead runners approaching the finishing line on the other side of the road, whilst we had barely made it halfway round, was slightly off putting. Nevertheless the run was proving less painful than I had imagined, maybe I was distracted from my pain by the various roadside entertainers on the way or the large group of lads in England shirts who stopped halfway to go and buy some crisps and still caught us up 3 km later!

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Sponsored Swim

Ian Lane

On the 30th August 2002 I did a sponsored swim to raise money for the MPS Society. I started the swim at Barnsley Metrodome at 6.30pm and finished at 8pm after having completed 55 lengths of the swimming pool. I broke my personal record by swimming the greatest number of lengths that I have ever swum for charity. My previous record was 52 lengths but I was very confident that I was going to beat that this time, especially as it was raising money for the MPS Society. I enjoyed the swim very much.

I would like to say a very big thank you to all of my family, friends, neighbours and MPS Society staff for supporting me and the MPS Society and I hope to do another fundraising swim next year.

Sheffield Marathon

Joan & Roy Ingram

£80.00 was raised on behalf of the Ingram's grandson, Matthew Ingram who has Hurlers. Three men who work at the Ingram's factory held a sponsored Beardathon. One the laboratory technicians Dave Keyworth ran the Sheffield Marathon he was sponsored Roxspur Measurement & Control Ltd, they donated £100.00 on behalf of the company and the rest was raised by the Ingram's work friends and relatives.

Above is Dave wearing his medal with the beardathon lads. Altogether £516-60 was was raised in total.

Sheffield Marathon

Kevin & Yvonne Puddy

On Sunday 14th April 2002 the Wells City FC organised a fundraising day to commemorate the 1st anniversary of the death of our daughter Annette. A friendly match between a Wells City XI and a Bristol City Old Boys XI was well supported, resulting in a thoroughly entertaining four all draw. Refreshments were laid on in the Club House after the game, where two other lads from the Wells team joined myself in the sponsored 'Head Shaving' event. The money raised was to be divided equally between the MPS Society and The Little Bridge House Hospice, where we spent a number of happy visits with Annette. Please find enclosed a cheque, which we hope will help to enable other families to benefit and receive the wonderful support that the Society was able to give to us. Time does'nt stand still and life is constantly changing and moving on, but the Society will always remain a part of our lives. Thank you all once again.

Donations and Fundraising

The Society is grateful to the following:

Donations

Vauxhall Motors
Dawn Nelson
3M United Kingdom PLC
Teneza Wall
Zurich Financial Services
KPMG – Birmingham
Rothschild & Sons Ltd
Argos
ILI
The Thorngate Trust
Kelmscot Charitable Trust
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Albert Van den Bergh Charitable Trust
Howes Percival Solicitors
The Robertson Trust
Northgate
Experian
Mr & Mrs Armstrong
Mr & Mrs Selvaranjan
Swale Charity Trust
Mrs M Barralet
London & Scandinavian Metallurgical Co Ltd
Mr & Mrs Whettem
Mr M Briggs
Brackla Junior School – Bridgend

Fundraising

Jean Brooks – Keep Fit Class
Tunstead Pre-School
Hanley St Lukes C of E Primary School
The Snowdonia Challenge
Ian Lane – Sponsored Swim
Steve Robjohns – Marathon Run
Marina and Dave – Car Boot Sale
Charles Richardson – Bike Ride, France
Francesca Oakley – Swim Club Event
Clare and Paul Titcomb – 10K Road Race
Hillside Player's
Ian Lane – Sponsored Swim

Stamps

Miss L Ricketts
Ernie Butler

In Memory

Michael Matthew Armstrong
Shivahram Selvaranjan
Jesica Stuart
Aaron Jennings

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Vice-Chair	Judy Holroyd Bob Devine		
Treasurer	Judith Evans		
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	Alison West	Development Officer	a.west@mpssociety.co.uk

Do let us have your family stories and any helpful hints you would like to share with our newsletter readers. If you have a question that you would like to see answered in a future edition of the newsletter, please do write to us.

To submit information to the newsletter please send materials (preferably via e-mail for text) and mail photos to the address on the left.

The articles in this newsletter 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.

NEWSLETTER DEADLINES

WINTER
17 December 2002

SPRING
31 March 2003

SUMMER
30 June 2003

Autumn
30 September 2003

CONTACT US

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Fax: 01494 434252

OUT OF HOURS HELPLINE: 07712 653258

email: mps@mpssociety.co.uk
Web site: www.mpssociety.co.uk
Fabry: www.fabry.org.uk

Christmas Cards 2002

Sold in aid of
The Society for Mucopolysaccharide Diseases
Registered Charity: 287034

Mucopolysaccharide and Related Diseases cause progressive physical and mental disability usually resulting in death in childhood.

The Society for Mucopolysaccharide Diseases is the only National Registered Charity, providing information, advocacy, and practical help to families of affected children and young adults. At present there is no cure and the monies raised from the sale of these cards will help towards supporting over a 1000 affected families in the UK

ORDER FORM 2002

Card Code	Size (mm)	Description	Pack Size	Cost per Pack	Quantity Ordered	Value
A052/01	100x100	Polar Bear Knitt	10	£2.25		£
A037/01	100x100	Moon & Mistletoe	10	£2.25		£
A032/01	100x100	Christmas Cherub	10	£2.25		£
00/3925	111x111	Santa & hole in Sack	10	£2.50		£
DO66/02	210x110	Santa's Washing Line	10	£3.10		£
BO38/02	125x125	Snowdrop Robin	10	£2.80		£
69338	180x140	Santa's Animals	5	£1.60		£
CO25/02	121x171	We 3 Kings	10	£2.95		£
CO20/02	121x171	A Christmas Wish/ Magic Begins	5 of each	£3.00		£

PAYMENT DETAILS

Postage and Packing	All Card Sizes Approximate	Card Total	£
1-4 Packs £1.20		Postage and Packing	£
5-10 Packs £2.00		Donation	£
11 Packs or more £3.00		Total	£

1. Please fill in your name, address and phone number and indicate your card selection on the order form.
2. All payments should accompany orders. Please make Cheques/P.O. payable to "The MPS Society", or pay by Credit Card.

BLOCK CAPITALS PLEASE

Contact Name:

Address:

Postcode:

Tel No.:

CREDIT CARD DETAILS

Cards Value . . . . . .

Post & Packing . . . . . .

Donation . . . . . .

Total Amount Due . . . . . .

Card No: _ _ _ _ _ / _ _ _ _ _ / _ _ _ _ _ / _ _ _ _ _

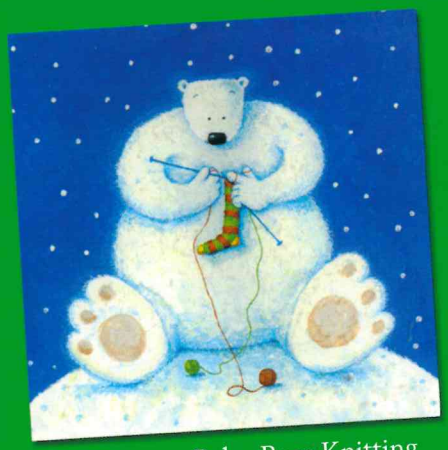
Cardholder Name . . . . . .

Card Type . . . . . Visa/Mastercard

Expiry Date _ _ / _ _

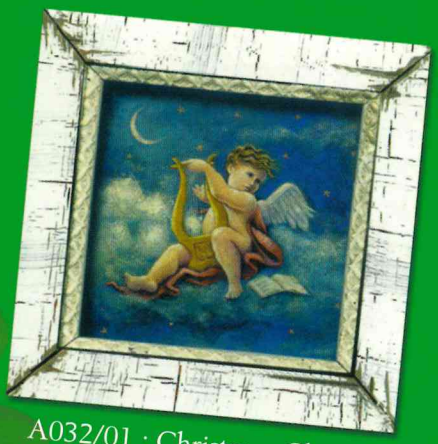
PLEASE RETURN YOUR ORDER FORM TO:
The MPS Society, 46 Woodside Road, Amersham, BUCKS, HP6 6AJ
TELEPHONE: 01494 434156 FAX: 01494 434252

Christmas Cards 2002



A052/01 : Polar Bear Knitting
Size: 100mm x 100mm

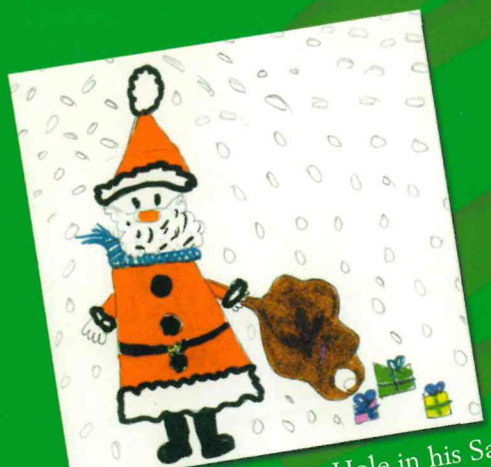
A037/01 : Moon & Mistletoe
Size: 100mm x 100mm



A032/01 : Christmas Cherub
Size: 100mm x 100mm

DO66/02: Santa's Washing Line
Size: 210mm x 110mm

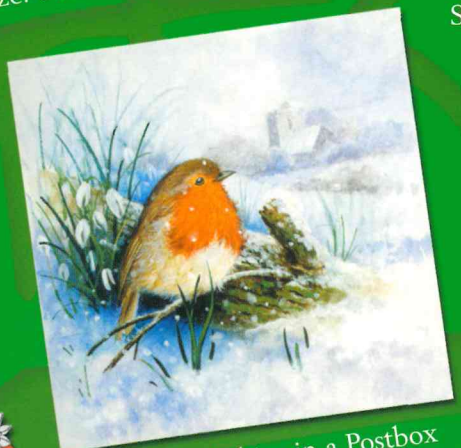
CO25/02: We 3 Kings
Size: 121mm x 171mm



CO/3925 : Santa has a Hole in his Sack
Size: 111mm x 111mm

CO20/02: Magic Begins
Size: 121mm x 171mm

CO20/02: A Christmas Wish
Size: 121mm x 171mm



IC0431 : Robins in a Postbox
120mm x 120mm

69338 : Santa's Animals