Newsletter

Spring 1995

The Society for Mucopolysaccharide Diseases

National Registered Charity No. 287034

The Society for Mucopolysaccharide Diseases

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The MPS Society is a voluntary support group, founded in 1982, which represents over 700 families in the UK with children or adults suffering from Mucopolysaccharide and related diseases. It is a registered charity, entirely supported by voluntary donations and fundraising by members, and run by the members themselves. Its aims are:

- ☐ **To act as a parent support group**
- ☐ **To bring about more public awareness of MPS**
- ☐ **To promote and support research into MPS**

The Society operates a network of Area Families throughout Great Britain and Northern Ireland, who offer support and links to families in their areas. It provides an information service for families and professionals. At the present time it supports two specialist MPS clinics at the Royal Manchester Children's Hospital and at the Hospital for Sick Children, Great Ormond Street, London. The Society also funds three biochemists, one at Manchester Children's Hospital, one at the Christie Hospital, Manchester, and one at the Institute of Child Health, London. It encourages and assists contact and co-operation between parents and professionals and maintains links with sister societies in Europe and throughout the world.

There is at present no cure for MPS diseases, but much can be done to improve the treatment and care of sufferers. The slogan of the Society is:

"Care today, hope tomorrow"

Front cover picture is **Jessica Stuart** of Borehamwood Herts with her new baby sister, **Hollie Anne** born on the 4th of January. Jessica is four and has Hurler disease.

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Deadline for Summer Newsletter

22nd July 1995

DIRECTOR'S REPORT

These last three months have been as challenging as ever in keeping the needs of children and young adults with MPS and their families at the forefront both at a national level through parliamentarians and voluntary organisations and locally through the relevant statutory bodies.

From listening to families it has become very obvious that the means testing carried out to decide the level of contribution from the family before a Disabled Facilities Grant is offered is causing enormous stress both financially and emotionally.

Both Mary and I have, in recent years, since means testing was introduced, supported a number of families in fighting local authorities and appealing against the level of contributions required.

With the limited resources of the Society it is not feasible to continue this piecemeal approach in the long term.

Dafydd Wigley MP for Caernarfon has kindly offered to bring this issue to the attention of relevant Ministers with the support of the Society.

Therefore we need your help. If you are buying your own home and :

a) have received a Disabled Facilities Grant to adapt your home for your MPS child(ren).

b) have been means tested for a Disabled Facilities Grant and not proceeded

c) have been means tested for a Disabled Facilities Grant and are proceeding

Please would you complete the questionnaire and send copies of any relevant correspondence to the MPS office.

We will continue to keep you informed through the Newsletter of any developments.

May I offer a big thank you to all those who took part in our random pilot study 'Genetic Counselling and Carrier Testing for MPS'. The results are printed later on in this Newsletter. If you have not returned your questionnaire we would still like to receive it and if any family not included in the random pilot study would like to participate please do let the office know.

On June the 29th I have been invited by the Genetics Interest Group to present our findings at their National Conference in London.

Following on from this we have been asked to seek the views on carrier testing from siblings aged 10 years and over who have or have had MPS brothers or sisters, and included in this Newsletter is a short questionnaire which we hope parents will feel able to encourage their sons and daughters to complete.

To end on a lighter note **'The Wedding'** went off with out a hitch. Mary looked beautiful and was attended by her daughters, Alexandra and Catherine and Rob's son, Tristan. I am sure you would join me in wishing Rob and Mary every happiness in the years ahead.

Christine Lavery
Director

Births

Congratulations to **Bob and Rhian McKnight** from Lougharne, Dyfed on the birth of their daughter **Rhoswen Amy** born on the 27th of February 1995.

Congratulations to **Peter and Sue Stuart** of Borehamwood, Hertfordshire on the birth of their daughter **Hollie Anne** on the 4th of January 1995.

Congratulations to **Terry and Christine Vigus** on the birth of their son **Damien Frederick**. Damien was born on the 6th January 1995 and the family live in Cornwall.

New Families

Chris and Julie Kembrey from Bristol, whose twin daughters, **Josephine and Francesca** born on the 21st July 1990 have been diagnosed with Sanfilippo Disease.

Bob and Kim Marston-Taylor from Hertfordshire whose 7 year old daughter **Alice** has Sanfilippo Disease.

Gerald and Christine Papworth from Huntingdon whose only son Jeremy has just been diagnosed with Sanfilippo Disease at the age of 21 years.

Rashpal and Sandra Singh from Coventry whose only son, **Daniel** aged 3 years has been diagnosed with Hunter Disease.

Keith and Angela Thompson from Birkenhead whose youngest daughter, **Holly** has been diagnosed with Morquio Disease. Holly is 22 months old.

Zaffer and Rukhsana Mizra from Edinburgh whose 13 year old daughter **Usman**, suffers from (female) Hunter Disease.

Dennis and Connie Yeo from Maidstone whose son, **Matthew** and daughter, **Karen** are both young adults with Mannosidosis.

Deaths

Fredrika (Freddie) Graf from New York in the USA died on the 24th of January 1995. Freddie, who suffered from ML II, and her parents Johan and Suzie won the hearts of all the families when they attended the Society's conference in Northampton in September 1994.

Mark Oliver Harvey from Oxford died on the 29th January 1995 aged 13 years. Mark suffered from Sanfilippo Disease and is remembered along with his brothers, Ian and Stephen who also died from Sanfilippo Disease in 1993 and 1991 respectively.

John Peter Hodgetts from Tamworth, Staffordshire who died from Sanfilippo Disease on the 24th January 1995 aged 14 years. John's parents Geof and Sue, and brother Carl are Area Support Family for the Midlands.

Rebecca Palqez Ruiz from Seville, Spain died on the 8th of January 1995 from Morquio Disease aged 14 years. Christine met Rebecca and her family when she spoke at the Spanish Conference in March 1994.

Darren Tailford from Stocksfield on Tyne, Northumberland died on the 17th of February 1995 aged 16 years. Darren suffered from Hunter Disease.

THANK YOU

Rob and I would like to express our thanks to all the families and friends for their lovely wedding gifts.

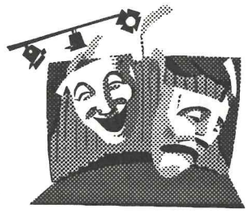
We must say what a great surprise, not to mention odd looks from some guests, when we opened the "tent", "sleeping bags", "rucksack", plus 50 feet of "bright Green rope"!!!

The list of wonderful gifts from you all goes on and on and we would like you all to know how much we appreciate your kindness, thank you all.

The wedding day went well, we only rowed when it came to cutting the cake !

Much Love,

Mary and Rob



1995 Dates for your Diary

21 - 23 April	Austrian MPS Conference
28 - 30 April	Area Family Training Weekend
21 May	Area Family Day at London Zoo (Ron Snack)
28 May - 3 June	Teenage Activity Holiday, Devon
11 June	Area Family Day and Garden Fayre at Nantwich (Sylvia Blackburn)
18 June	'Childhood Wood Working Party'
9 July	Area Family BBQ, Milton Keynes (Ron Snack)
15 July	MPS Trustees Meeting
22 - 29 July	MPS Family Holiday, Primrose Valley, North Yorkshire
10 September	Area Family Day at Cotswold Wildlife Park (Ron Snack)
17 September	Barbara, John Arrowsmith and John Brennan take part in the Great North Run.
20 September	Northern Ireland Regional MPS Conference
22 - 24 September	MPS Annual Conference, Northampton
24 September	MPS Annual General Meeting (at the Conference)
2 December	MPS Giant Tombola at Milton Keynes (Ron Snack)
10 December	Area Christmas Party at Milton Keynes (Ron Snack)
23 December	MPS Flag Day at Milton Keynes (Ron Snack)

Rob and Mary Paget cutting the cake.

The Childhood Wood Planting at Sherwood Pines

24th of February 1995

The Children remembered 1995

Vincent Tucker
Joanne Greenwood
Andrew Robert Kennedy
Simon Keith Meek
Sarah Jane Lowry
Matthew James Hardy
Martin Haigh
Baby Heritage
Gemma Ann Rolinson
Abigail Anna Milward

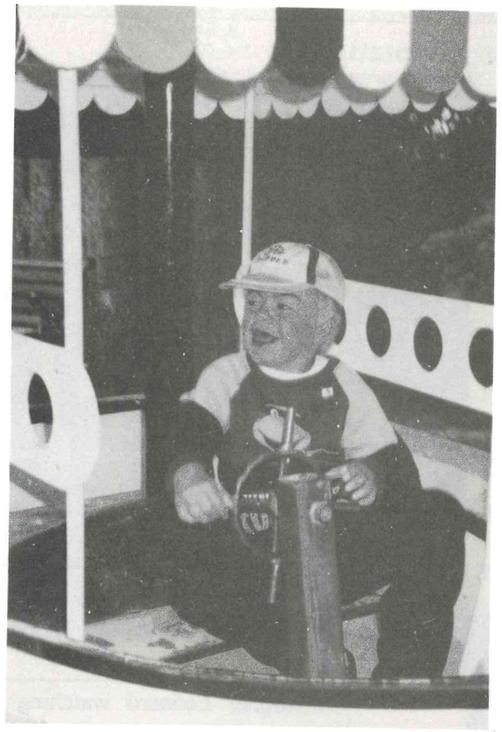
Christopher Leonard watching Daddy, Paul, planting an oak sapling in memory of brother and son, John.

The Children remembered 1995

John Richard Leonard
Amber Le Page
Christopher Paul Shorthouse
Marie Louise Jobson
Lloyd Stewart
Darran Jon Carr
Jamie Mc Donald
John Peter Hodgetts
Darren Tailford

Chairman of Nottinghamshire County Council, Peter Burgess, with Mrs Burgess and Paddy Tipping MP, planting a sapling in remembrance of an MPS child.

West Cornwall MPS Families



Jamie Hicks enjoys being captain of the ship on holiday. Jamie is nine and has Hunter disease.

As Area Family we would like to meet all the MPS families in our area, but because of time and distance it is not always possible. Some of our families live 200 miles away from us and it is very difficult for them to attend family days and Christmas parties, so we try once a year to visit them at home.

All these families live in West Cornwall and they seem cut off from contact with the Society except by telephone, so visiting them is always a pleasure to us and we usually end up being treated to some wonderful Cornish hospitality.

There are four families living in Cornwall, the Hicks family live in Penzance, the Vigus family in Hayle, the Jobson family in Padstow and the Oliver family in Penryn.

It's no good being on a diet if you visit Carol and John Hicks, plenty of cakes and Cornish cream. They need plenty of energy to look after their son Jamie, who has Hunter's and is very active with a wonderful wicked sense of humour. Also they have two other children Curtis and Donna who are equally mischievous.

Emma Vigus who suffers from Sanfilippo is a gorgeous little girl who, although she has had lots of medical problems lately, has responded well. Christine and Terry, her parents, cope with the situation extremely well as they are so far away from Ed Wraith at Manchester.

Sadly, Marie Jobson died last year and although we never met her we had the privilege to hear her talking and singing on tape. Marie, or 'Mimi' as she was known, suffered from Hurler's but this didn't stop her from doing most things a normal nine year old would do. Marie's talent for singing was passed on from Ann, her mother, who has recorded a beautiful song about her which is available on tape. (All proceeds go to MPS.)

Many of you will have seen a picture of Amie Oliver in the Newsletter dressed as a bridesmaid, didn't she look great! Alison and Nigel, Amie's parents have always attended the Christmas parties, whether it's in the South West or South Wales - this means a trip of over 400 miles. They seem to like travelling as they recently took Amie to America.

Although Cornwall is many miles from most of the other MPS families, I'm sure some of you go to Cornwall on your holidays. I'm sure the families in Cornwall would like to meet visiting MPS families and I know from our experience that you would be made very welcome.

Tony and Shirley Eyre

Profile of a Committee Member - Jon Lawrie

Hello I'm Jon Lawrie and I have been married to Cathie for nearly 15 years. We live in Hull and have 3 sons. Stuart aged 10, David aged 7 and Timmy aged 4. Stuart has Sanfilippo Disease (MPS IIIB) and was diagnosed in 1990.

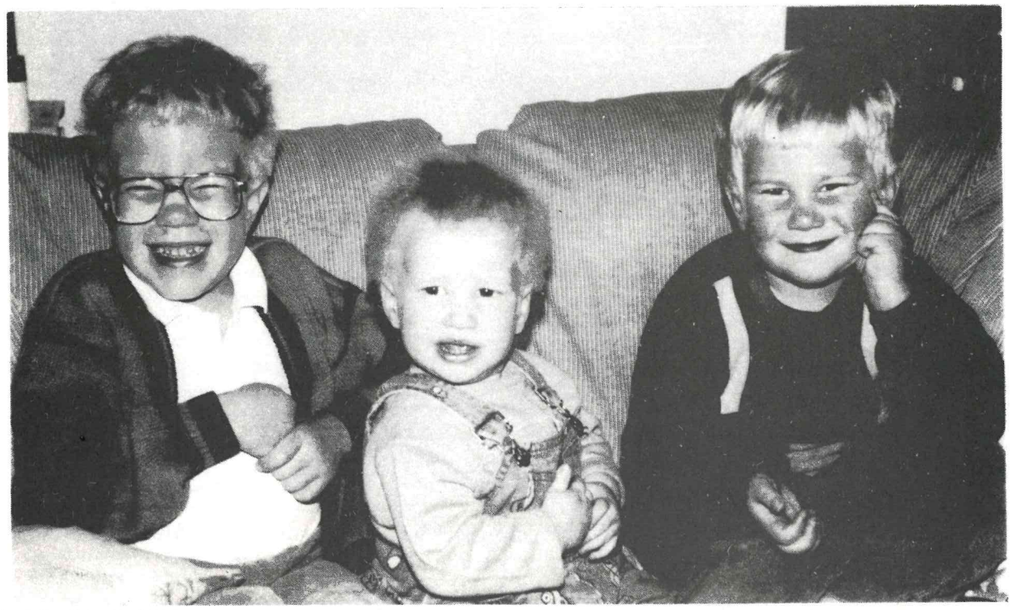
We immediately joined the MPS Society and have benefited greatly from the support and friendship generated by the Society. We have enjoyed family holidays, conferences and area get-togethers and have tried to put something back by way of fundraising and spreading information about MPS.

In May last year I was made redundant and, with more time on my hands, I offered to take a more active part in various groups which help us because of Stuart's condition. This included standing for the MPS committee and becoming Head of Governors at Stuart's school.

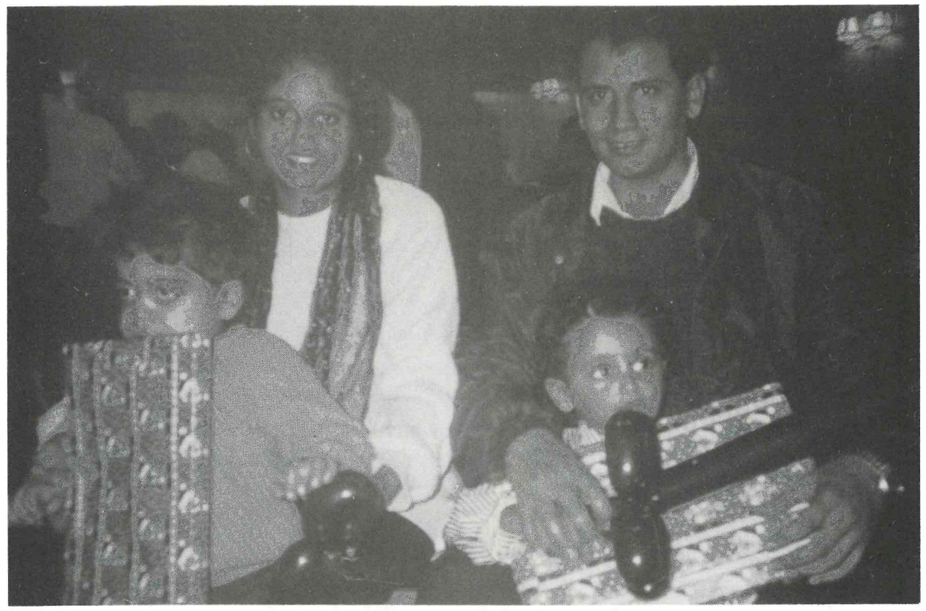
Since January I have been employed as a Manager for the Wakefield branch of Phoenix Landscapes. This (car) and (office) space is counteracted at the weekend by 3 very active boys who keep me fit. (or run me ragged)

Most spare time activities are family based so when an unclaimed moment turns up, I enjoy reading, chess, watching films, listening to music and cooking - preferably when I have the kitchen to myself.

Jon Lawrie
Trustee



Stuart, Timmy and David Lawrie in 1991. Stuart is now aged ten and suffers from Sanfilippo disease.



Mr and Mrs Obad, a new family in Liverpool, from the Yemen, with their sons **Ahmed and Mohammed.** Ahmed suffers from Hunter disease.

Joanne and Myles Broughton from Sheffield hugely enjoying a meeting with a furry friend at the National Holiday Fund trip to Florida in November 1994.

Our special thanks to **Malcolm and Lis Haddow** and the NHF for their continued generosity to the Society in offering holiday places - not just sentimental outings, but a chance for disabled children to grow in independence and confidence and the spirit of adventure. Also a welcome break for parents knowing their children are being cared for by a most reliable and experienced team.

FLORIDA FROLICS - by Joanne Evans - 8 years of age.

I have just come back from a fantastic holiday in Florida with the National Holiday Fund and I am just about to tell you about it. It started off with me and my Mum flying from Glasgow to Gatwick on Air UK. We went on a buggy from the airport to the hotel. (Everyone else had to walk) We were first at the hotel. I made friends with a couple of other children called Toni and Joanne and I met my carer June who was really nice. The actors and actresses of The Bill came to see us and gave us hats badges and autographs.

After saying goodbye to our parents we made friends with one another and then we flew on a Jumbo Jet to Florida. We went to Cypress gardens and my favourite bit was the water ski show. Just as we were about to watch it - American rain started- (rain with thunder and lightening).

On our second day in Florida we went to Seaworld. We saw dolphins, killer whales, sword fish and many other kinds of fish. We saw a fireworks display and the dolphins and whales doing tricks in the water. We also fed dolphins with fish and patted them, they felt wet and rubbery. The third day we went off to MGM Studios. We went on the great Movie ride and behind the scenes tour which was really scary and we all got very wet. We also saw Beauty and the Beast on stage which I enjoyed. I got Mickey Mouse's autograph. The next day we went to the police station, but first we had lunch (hot dogs) then we saw a display of how the police train their dogs. Later we were allowed to sit on the motorbikes and in the police cars; Chris put on handcuffs and couldn't get them off for and hour!

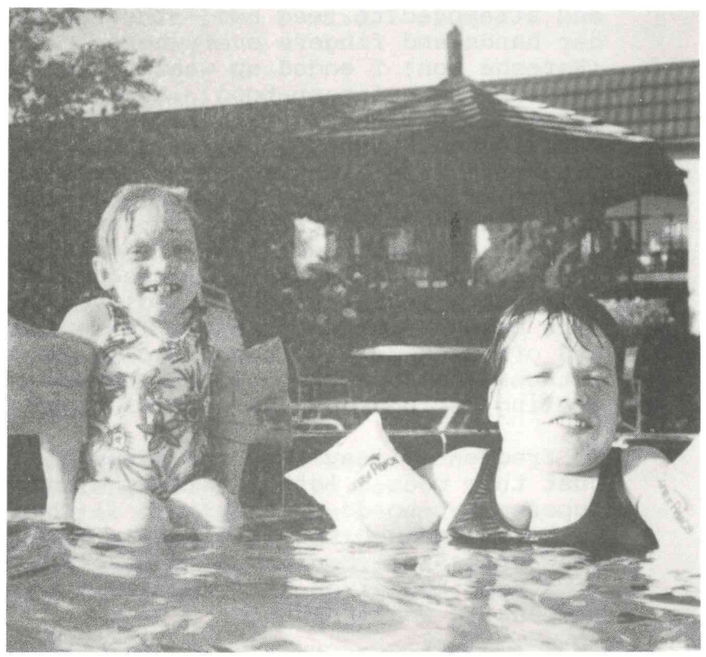
The next day we went to EPCOT where we saw what earth would be like in the future. I got autographs from Daisey Duck, Goofy, Minnie Mouse and Chip and Pluto. I also met Figment who is one of my favourite characters and I bought a Figment soft toy.

When we went to the Kennedy space centre we went into the space garden and saw the model space rockets; I had my photo taken with a spaceman.

Our next trip was to the Magic Kingdom and we went on lots of rides but my favourites were Thunder Mountain and Mr Toad's Wild Ride because they were quite jerky and scary. We had tea in Cinderella's castle and Toni and I spoke to Cinderella, she was wearing her ballgown. We went to Busch Gardens and also Disney Village where we went shopping and bought lots. After returning to the Magic Kingdom we went to see the Blue Knights who are policemen. First we went to look at their motorbikes and later they took us for a ride on the bikes round the county which felt very fast. (All the boys in my class at school are really envious)

The next day we went to Universal Studios and we went on the E.T. and Jaws rides: it was really scary and I'm still having bad dreams but I am glad that I went on it. Later we had tea at the Hard Rock Cafe which was excellent - Toni and I shared an enormous ice cream - the best I've ever had! On the last day of our holiday we had difficulty fitting in all the toys and things we had bought. Later we went swimming and the boys splashed me. After breakfast the following day we all got on the bus to go to the airport; we sang "Ten Green Bottles" (we missed out the "tt" when we sang bottles). Toni and I did magic tricks to amuse ourselves (she certainly fooled me). The flight took nine hours and it seemed a long time. When we arrived in England it was the morning but we wanted to go to sleep. We were all excited to see our Mums and Dads but we were sad that our holiday was over and having to say goodbye to all our friends.

Joanne Evans with friend on a day when they didn't have American rain.



Life Begins at Thirty - when you meet Natasha Macintyre.

Returning to work after having a family I decided to work as a Nurse in the Community. One day in April '94 I recieved a phone call from my Area Manager asking me to visit a family in Bagshot "THE MACINTYRE FAMILY". My Area Manager explained that Natasha, who is 15, has MPS. "Great" I thought, "a nice easy job" (I didn't know much about MPS and Definitely didn't know Natasha!)

Still under my happy delusion I set off to meet Julie and Natasha. I was greeted by Julie, looking frazzled, but still feeling guilty about accepting help. Julie introduced me to Natasha. Two enormous brown eyes weighed me up - so life in the Macintyre household began.

Each morning I go in to bath and dress Natasha, help her with her breakfast and see her off to school. Each evening, about 5.00, I return to help Natasha with her tea, bath and put her P.J.'s on.

For a week Natasha co-operated! Little did I know Natasha was not feeling her best! On the Friday I said Goodbye, not quite understanding why Julie had looked so Frazzled - Ignorance is bliss!!!

I returned on Monday afternoon to be greeted by a new Natasha - bright, breezy, beautiful and UNCO-OPERATIVE!! "Oh" I exclaimed " how wonderful to see Natasha so happy and bright". Natasha just smiled.

Julie gave me Natasha's tea. I sat her in her chair and attempted to feed her - the fun began!! Natasha put her hands and fingers everywhere. After a battle (Natasha won) I ended up wearing her tea. Julie then gave her some finger food. After Natasha had finished her tea I decided to bath her - MISTAKE NO. 2!! I carried Natasha upstairs, laid her down to remove her clothes (or so I thought). Natasha was NOT having this. Up she sat with a shout making me feel so guilty for upsetting her. I tried again, and again, and again, and again - all the time Natasha shouted. Finally, after what seemed like hours I succeeded in bathing Natasha, I took her downstairs with a red face, thinking Julie would fire me. What greeted me was Julie and Kirsten smiling and offering me a soothing cup of coffee - apparently Natasha does do this! I left the Macintyre household feeling as though I had done 12 rounds with Frank Bruno!

I frequently leave wearing Natasha's breakfast or tea. Just this week I left Natasha and went to my local Supermarket to do my shopping. I then went to my Mothers. "Lynne" she said , "you have chocolate all down your dress" - I don't have to explain it wasn't chocolate - Natasha had given me a new (even though offensive) present to wear!

Life since April has been unpredictable. No two days are the same. Natasha and I do battle over baths, food, dressing - in fact you name it Natasha and I battle (in the nicest possible way) and then Natasha throws me and co-operates fully!!

However, what I must add is that Natasha has brought something very special to my life. She suffers so much and complains so little. The whole Macintyre household is something special, and I don't think of Natasha as a job.

Life definitely began at 30! Natasha's account may be different!

NATASHA'S ACCOUNT

One day in April 1994 I woke up feeling that I needed to broaden my horizons - I needed more new friends. My present ones are great, but you know what they say - a change is as good as a rest.

Now I needed a plan of action - in came Mum to get me up "Oh bother just when I needed to think". I decided not to co-operate as I had plans to make. Mum was getting frazzled - I couldn't understand why - I'd only kept her up for a month - who needed sleep!!"Natasha" she said "enough is enough" "Mummy is so very, very tired I am going to get someone in to help". SUCCESS without trying - just what I wanted - a new friend!!

Well, within days Lynne turned up. I wasn't feeling my best so I didn't take much notice, but she looked a pushover! The next day Lynne arrived to help me with my daily ablutions and eating - but like I said I didn't feel well!

The following week I felt great! Lynne arrived Monday teatime. "Oh good now the fun begins". I was hungry, but Mum and Lynne were so impressed with me looking so well I wasn't getting my tea. I chewed my finger - instant response - Mum went to get my tea!

Lynne walked to my chair grinning like a Cheshire Cat (doesn't take much to please this one!). Mum handed Lynne my tea - what was this? I WANTED lemon Sole Goujons - I'd got Pasta - Well I WASN'T eating that!! I tried to wrestle the plate off Lynne - Cor, she did look silly wearing my Pasta - and guess what - I got my Lemon Sole Goujons!

After tea Lynne took me upstairs for a bath. "Oh is there no peace". I am laid gently down to remove my clothes - who does she think she is - I am 15 you know! Anyway, after a disagreement Lynne won (only because I let her). I had my bath. Lynne looked frazzled just like Mum or Morny (my best friend at school) does.

Lynne comes in twice a day and friday evenings to look after me. I frequently let her leave wearing my breakfast or tea (I've been told it is polite to share). Once I even gave her an extra special present! It is good fun having a new friend and I've got Lynne wrapped around my little finger, and I still get to play Mum and Dad up at weekends and night times. It is what is called "Having the best of both worlds"!

Every now and again I lull them all into a false sense of security and just sit quietly, eat my food and let them change me - but you know what they say - "Variety is the Spice of Life!!!!!)

Lynne O'Hara

"Ride For Lives"

One of the pleasures of being Chairman of the Society is being presented with cheques by people who have raised money for MPS. It is not just the receipt of the cheque but of meeting personally the many people who go to tremendous efforts to raise money for the Society.

Earlier this year I was presented with a cheque for £3,700 being the efforts of two local people who rode two 125cc motor cyles around Britain's coastline. The two were **Mark Gutsell** whom I have known for several years and **Phil Scott**, more commonly known as "Haggis" who is the father of Heather who suffers from MLIII. The motor bikes were loaned by **Don Gabriel** of Hamilton's Motorcyles of Streatham, London. Mark and Haggis were supported by **Franklin and Dave** who drove the support vehicle which was loaned by Unigate Dairies.

The journey started from Hastings going in an anti-clockwise route around the coast and I learnt from Mark that throughout the journey they met wonderful people who are now aware of what Mucopolysaccharide Diseases are all about. Mark works for the Child Support Agency and one of the stops was at the CSA in Falkirk in Scotland where over £500 had been collected for Mark to take with him.

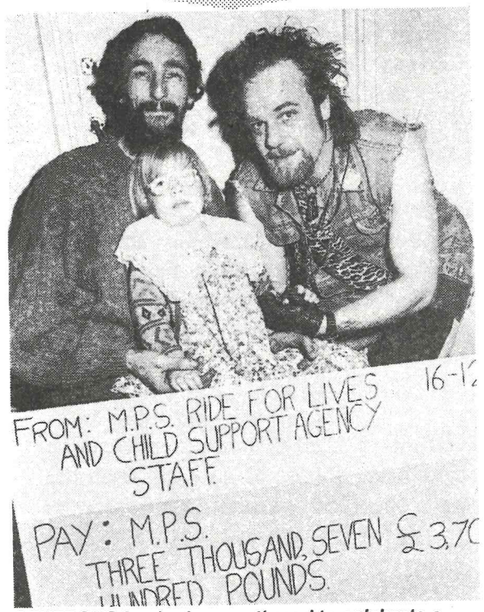
One of the highlights of the trip was a stop in Frome in Somerset where a cheque for £50 was given to Mark and Haggis. The money was raised by children who were either physically or mentally handicapped themselves, one of whom has Sanfilippo - what she wanted most was to ride Mark's bike.

The ride took eleven days to complete and although it wasn't all plain sailing and fun, the riders and back up team did enjoy themselves thoroughly. Mark hopes that "Ride for Lives" will happen again this year and he hopes some time to set a British record by riding all of Britain's coastline, including Northern Ireland on a trike - motorised of course!

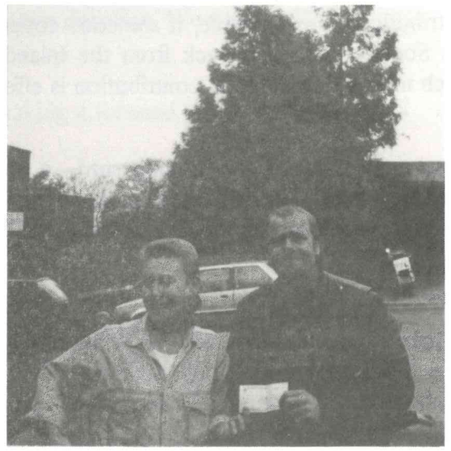
I thanked Mark and Haggis on behalf of all members of the Society for what they had done and I am sure everyone joins me in congratulating them. I should perhaps adds as regards the photos, in particular the presentation one, that Mark is not a "pink rocker" but the function where the cheque was presented was a 50s/60s Night.

Alf G. King

Kind hearts raise £3,700 for kids



GENEROUS fund-raisers gathered to celebrate a good year's work at the Civil Service Club, Harrow Lane, on Saturday night.
The disco and charity night for fund-raisers for the rare children's disease mucopolysaccharide celebrated raising £3,700 this year.
The group, inspired by little Heather Scott, a Hollington girl who suffers from the disease, hope to be able to buy a Christmas present for each of the 25 or so children in the country who have MPS.
Pictured, left to right, Phil 'Haggis' Scott, Heather Scott and Mark Gutsell.



With **Jackie Moore** at Frome, Somerset. Jackie works with severely disabled children and helped them to raise a cheque for £50.

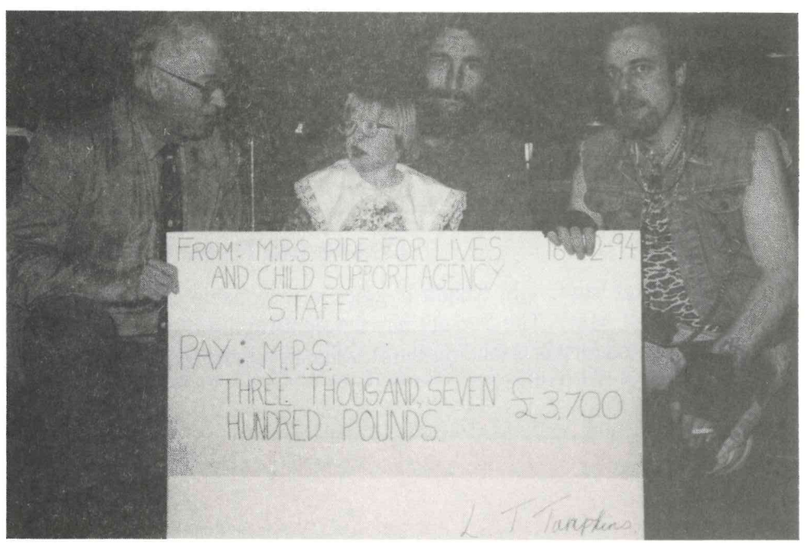
Top right:

Mark with the machines and back up vehicle (Unigate Milk Cart).

Right:

Alf King, Heather Scott, "Haggis" Scott and Mark Gutsell.

The Child Support Agency have decided to adopt MPS as their special charity. We look forward to working with them again in future.



Effective Giving

Although in some areas of the country the economy is apparently improving, in many places it is still in a very depressed state and this means that the funds reaching the MPS Society are diminishing. To enable the Society to continue to move forward we need all the money we can get and one way at no extra cost to anyone is to try to ensure that all personal gifts or donations are as tax effective as is possible and some ways of ensuring this spring immediately to mind.

One way is the common **Deed of Convenant** where a person intends to make the same annual donation to the Society for four consecutive years. If you are liable to Income Tax and make payments under a Deed of Convenant then the benefit to the Society increases by one-third of your contribution. For example, if someone covenants £50 a year for four years she would pay £200. The Society can claim back from the Inland Revenue over the four year period more than £66 which means that the total contribution is effectively £266.

Some people like to give a lump sum - a single payment. If this is £250 or more then it can be given under the **Gift Aid Scheme** which results in the Society being able to claim again from the Inland Revenue an additional one-third of the amount given - the £250 becomes immediately in excess of £333.

For lump sums less than £250 it is possible to use what is termed a **Deposited Covenant.** Here again the money back from the Inland Revenue is one-third of what is given. If for example a donation of £60 is made, £20 can be claimed back from the Revenue spread over four years.

I must emphasise that for the Society to benefit from any of the above schemes the individual who pays the money must be liable to Income Tax. If they are liable then their donation increases with absolutely no extra cost to themselves, the balance being in effect paid by the Inland Revenue. If you are not personally in a position to help the Society in this manner then perhaps you have friends or relatives who might care to do so. If you wish for any further information please do write or telephone me or the Treasurer.

Alf G. King - Chairman

Desperate Need for Buggies!

Six second hand, light weight buggies in reasonable condition urgently needed to be taken to Russia on 11th of May. The Society has the opportunity of free transport of the buggies at this time when Christine Lavery is attending the Russian conference. Contact the MPS Office right away.

FUNDRAISING

It is disappointing but perhaps inevitable given the success of the national Lottery to see a significant drop in Fundraising and donations in the first 5 months of our financial year.

Therefore the Trustees have asked me to highlight this in the Newsletter and ask that as many of you as possible **Think MPS** when fundraising opportunities come along in 1995.

Here in the office we held a very successful collection with the help of volunteers and MPS families at the Amersham Tesco superstore. We had a small single board display with our posters and a thank you notice to every one donating. We were thrilled to raise £555.22.

If you have a fund-raising idea but need help in taking it forward please do let the MPS office know.

Christine Lavery

RAISING FUNDS AT YOUR LOCAL SUPERSTORE COULDN'T BE EASIER.

- **Obtain permission from the local store or head office.**
- **Recruit volunteers to do a 1 hour or 2 hour stint. (maximumof two at any one time).**
- **Once you have a date ask the MPS office for sashes, official collector badges, material for a display board, collection boxes and a paying in slip.**
- **Once the collection is completed use two independent witnesses to count and check the cash and take to the bank.**
- **Send paying in slip to MPS office and a thank you letter and copy of paying in slip can be sent to all volunteers if list of addresses is provided.**

There are a small number of conference places still available for the 13th Annual Conference at the Stakis Hotel Northampton, 22nd to 24th of September 1995. Please apply urgently.

MAGNIFICENT SEVEN CYCLE FROM BRISTOL TO BRADFORD (ON AVON) AND RETURN

The idea for this event stemmed from Ray Purdie, a colleague of mine in Avon County Council's Waste Regulation Branch.

Ray's enthusiasm over the pleasures of a gentle autumn cycle ride along the Kennet and Avon towpath spread to six other colleagues even though three of them had never heard of Bradford-on-Avon, and one of them had never cycled further than the nearest McDonalds!

Further hazards to be negotiated en route were the refreshment stop - generously provided by the staff of Bath's Waste Transfer Station, together with the heavy lunch at the "Cross Guns" at Avoncliff.

However, all the team safely completed the 60 mile round trip, with Granddad Don Tomlinson leading the way home.

A sum of £375 was collected which was a truly magnificent effort, as the whole enterprise was planned and executed in less than a fortnight.

Reporter Peter Morley.

Left to right: **Charlotte Perkins, Zohbair Rehman, Don Tomlinson, Paul Wilkinson, Steve Moran, Ray Purdie, Russ Jones, with Edward and Val Morley** from Bath. Edward is nine and suffers from Sanfilippo disease. Edward's father, Peter Morley works at Avon Waste Regulation but has definitely not been wasting his time in influencing his colleagues to raise funds for the Society.

JULIE AND RUTH'S CHARITY DAY

Julie Burlison and her friend Ruth Furlong organised a charity day at Julie's home in Oldham.

The day started off as a coffee morning but as neighbours, friends and local shops were very generous the girls ended up collecting lots of different items.

The girls raised £600.00 of which £300.00 was donated to Julie's son's school, Gorse Bank School. The school donated £42.80 to the MPS Society instead of sending Christmas cards.

The day was a great success and everyone enjoyed themselves.

Ruth Furlong and Julie Burlison at their charity day.

Mrs Emma Andrews and her daughter Linda presented a cheque for £692.05 donated by the Park Social Club in Knaresborough to Pauline Mahon, Honorary Treasurer and Area Family Representative.

Mrs Andrews two great-grandchildren, David and Jamie Andrews, who both suffer from Hunter Disease live with their parents Peter and Julia Andrews at Thurmaston in Leicestershire.

Pauline and Sean Mahon would like to thank Mrs Andrews and her three daughters the 'Andrews Sisters' for a most enjoyable evening at the Park Social Club.

Pauline Mahon
23rd January 1995

Mrs Emma Andrews from Knaresborough, with her daughter Linda, presenting a cheque for £695.00 to Pauline Mahon at the Park Social Club in December 1994. Her grandsons David and Jamie Andrews, who live at Thurmaston in Leicestershire, suffer from Hunter disease.

Derek Denham writes: (*See next page*)

Could you please place the enclosed essay in the next MPS magazine. It was written by one of my friends and won first prize in a writing competition at his school.

I would also like to say what a pleasure it was to meet Dr Wraith. It made a world of difference to meet someone who knew what I was talking about. Thanks to Alan Byrne for arranging the clinic and to Ed for his time.

Derek Denham.

38 Glenmuir Rd. Ayr, Scotland

"A FOUR FOOT WONDER"
By Sandy Baird

Raising over three thousand, four hundred pounds for charity is an achievement on its own, but it is even more of an achievement when you are physically disabled. Local resident, Derek Denham, has done exactly that. He has been the brains behind numerous fund-raising events. Two thousand, four hundred pounds of his money was donated to Children in Need, where Derek handed it in personally at the BBC studios in Glasgow. Other charities which have received donations from Derek include: Comic Relief and The Society for Mucopolysaccharide Diseases.

Derek suffers from one of the Mucopolysaccharide Diseases (Hunter's Syndrome), which results in him being four feet tall, is asthmatic, has stiffjoints and has hearing and sight difficulties. Derek wasn't born deaf, it wasn't until he was six years old that it was discovered that he needed hearing aids. Even though Derek has these difficulties, it does not stop him from participating in many sports, such as table tennis, badminton and pool. On an adventure holiday he also tried horse riding, archery, canoeing, sailing and orienteering. In 1991, Derek was Junior Pairs Champion at Prestwick Indoor Bowling Club, it was his first ever season playing indoor bowling. Derek also enjoys watching football, and is a keen Rangers supporter.

Computing and home videoing are two more of Derek's favourite pastimes. He has filmed football games and parties with his camcorder, and performs a variety of jobs on his Amiga 600. Derek has also met a few celebrities in his time, Hazel Irvine, Paul Coia and Ally McCoist, to name a few.

Derek has also got some strong views on the treatment of disabled people. He says, "Just because we are unable to do some things, we are not incapable." He also feels that disabled people are given too much sympathy from those around them.

I admire Derek very much. I cannot imagine what it would be like being unable to participate as well as an able bodied person and being unable to hear. He has raised a lot of money for charity, which is more than most able bodied people are capable of. I also think that it is amazing that he is still able to do some sports and have some fun at the same time, knowing that he is physically disabled. I really think he is a FOUR FOOT WONDER!.

This article was written by Derek Denham's friend and it won first prize in a writing competition at Sandy's school.

Doughnuts, Coffee and "T".

On a sunny Saturday last July, Jenny and Andy Hardy held their annual coffee and doughnut morning at their home in Haddenham. Andy is a dancer with the Towersey Morris Men who have supported the Society for some years - Matthew Hardy was an honorary member until he died last April. One of the dancers owns "the T Shop" in West Wycombe which sells Ford Model T cars. He very kindly offered a ride in one of his cars as a raffle prize and nine year old Sarah Maker was lucky enough to win the trip - much to the envy of her dad and other fathers also. Here is the report of Sarah's ride, written by herself.

"In the summer I won the raffle at an MPS charity fete. A vintage car arrived on my birthday to take me and three friends for a ride around Stokenchurch. The car was a 1920's Model T Ford and it had an open top. It could not go very fast and people waved at us as we were driven around Stokenchurch. I enjoyed my prize very much."

Andy and Jenny's next coffee and doughnut morning will possibly be their last. It will be held on Saturday July 15th. and the Towersey Morris Men will be there - will you?

Andy and Jenny Hardy
48 Churchway,
Haddenham, Bucks.
Phone 01844 291773

Sarah Maker with her prize Model T Ford.

Information - For Sale -Wanted

For Sale:

Ortho-Kinetics Reha-Buggy (large size).

Excellent condition.

Please contact:

Bill or Sylvia Blackburn

01270 626809 (evenings)

Wanted New Owner

For **Atari 520 Computer**, 2 joysticks and one mouse, 12inch Fexan colour monitor, 40 games, word processing programme etc.

Proceeds will be donated to the MPS Society. Buyer to collect or pay for postage.

Contact:

Sue and Dan Butler

Spriggs Holly House,

Spriggs Holly Lane

Chinnor Hill, Oxon OX9 4BY

0149 483185

Dave the Disk Doctor:

For a small fee the Disk Doctor will recover corrupted files from floppy disks. They deal with Amstrad PCW (CPM & Locoscript) PCs and MACs. All the proceeds go to charities. Don't forget to mention MPS when you contact them. That way we may be put on their list of charities. Phone 01892 835974.

I can recommend the service as I received my disk back within the week with all the files recovered and some useful information.

David Briggs.

TRAVEL INSURANCE FOR CHILDREN WITH SERIOUS ILLNESS AND THEIR FAMILIES

Our Way Travel Ltd has sent us details of a new policy designed for children up to and including the age of 19 years who are medically and physically disadvantaged.

They highlight that within the last couple of years all other insurers have put an exclusion to conditions of a serious, terminal and HIV related nature in their General Conditions. The Our Way Travel Ltd policy has been planned is to allow children affected by such conditions to obtain insurance and thus travel with their family.

For further details and a proposal form contact Our Way Travel Ltd, Foxbury House, Foxbury Road, Bromley BR1 4DG. Tel: 0181 313 3652.

printed by courtesy of 'Contact a Family'.

NATIONAL LOTTERY DRAW

Every Saturday when the lottery is drawn a charity or good cause is selected to benefit. If you have a chance, please nominate the MPS Society when the National Lottery Draw comes to your area.

RESULTS OF PARENT SURVEY 1995

GENETIC COUNSELLING AND CARRIER TESTING FOR MUCOPOLYSACCHARIDE DISEASES

* * * * *

Sample of 100 families who have one or more children affected by MPS.
Questionnaire sent out on 4th February 1995.
Response rate at 22nd February 1995 - 44%.

GENETIC COUNSELLING

A **Which MPS Disease affects your family?**

Hurler/Hurler Scheie/Scheie	11
*Hunter	19
Sanfilippo	13
Morquio	1

*Pattern of inheritance for all diseases is autosomal recessive except Hunter which is x-linked.

B **Did you as parents receive Genetic Counselling?**

Yes	25
No	18
'of a sort'	1

Of these 'Yes' responses, only in 11 families were geneticist or genetic counsellors the providers of this information.

C **Who provided information on the pattern of inheritance?**

Consultant Paediatrician	25
Genetic Counsellor	7
Geneticist	6
GP	0
Nurse	0
MPS Society	5

CARRIER TESTING

D **Ages of unaffected children who have been tested.**

This ranged from pre-natal to 21 years

2 - *Pre- natal*	**1** - *5 years*	**2** - *13 years*
3 - *Birth*	**1** - *7 years*	**2** - *16 years*
2 - *2 years*	**1** - *9 years*	**1** - *17 years*
1 - *3 years*	**2** - *10 years*	**1** - *21 years*
3 - *4 years*		

(i) **17 families *have had* carrier testing for their unaffected children**
(ii) **20 families *have not* had carrier testing.**
(iii) **6 families *didn't know* it may be available.**
(iv) **1 did not answer question..**

Note: 11 out of the 17 families whose daughters have been tested were at risk of Hunter Disease.
Of the 6 families where no testing has taken place in Hunter families, the daughters are all under 11 years of age.

IF CARRIER TESTED

E **Which Professional gave you the result?**

Consultant Paediatrician	- 14
Geneticist	- 2
Awaiting results(from 1994)	- 1
Didn't answer question	- 1

How were these results given?

In writing	- 3
In person	- 10
By telephone	- 4

If your child(ren) have been carrier tested, do they know the result of the test?

YES	- 12
NO	- 6

If 'YES'
How was your child told of the result?

By parents	- 10
By professional	- 2

With the benefit of your experience would you have made a different decision for your child, regarding carrier testing?

YES	- 0
NO	- 18

If your child(ren) have NOT been carrier tested. What age would you consider appropriate? (*some Yes's included as families have had one child tested and not others)*

Under five years	- 5
5 years - 8 years	- 2
9 years - 12 years	- 2
13 years onwards	- 4
13 years onwards if my child agrees	- 19
Do not agree with carrier testing	- 1

* * * * * * * * * * *

MPS Survey of Newsletter Readers

Nearly 700 one page questionnaires about the newsletter were sent out to families in January 1994. By April 58 had been returned. Here is a summary of the findings.

How many people read the newletter?

A. (1) Nobody, it goes in the bin - 0
(2) One person - 8
(3) Two to four people - 34
(4) Four to eight people - 13
(5) More than eight people - 3

B From the selection of - 'Very Good - Good - Indifferent - Poor - Very Poor' , how do you rate the newsletter?

Very Good = 33
Good = 25

Regular items were listed and people were asked to rank them in order of preference.

C The regular items were a very mixed bag but it seemed clear that most people enjoy the variety of regular items.

37 replies noted Letters, features and MPS activities, MPS families, Fundraising etc. amongst their top three preferences.

17 replies gave medical information as their top priority.

4 replies gave no preference. "I read through the newsletter from start to finish.".

One comment was that more medical information would be welcome.

D ***Suggestions for what people would like to see more of were as follows:-***

Pen Pal Section
More Medical Information
Photos of Area Families
For Sale Section
Information on Equipment. i.e. seating wheelchairs etc.
Most people suggested more photos and stories about families and children on holidays.

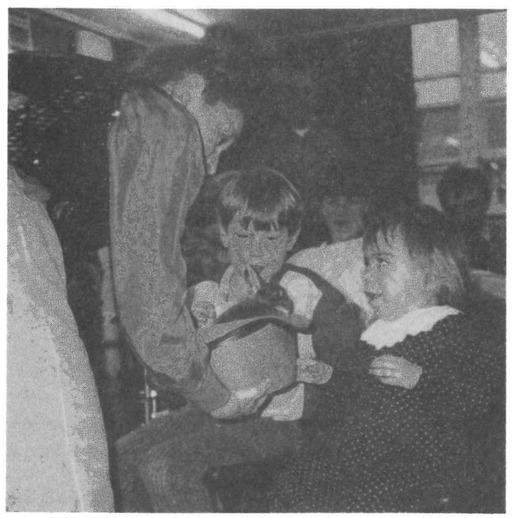
A "Doc Spot " - Information from Ed Wraith or Dr Vellodi - was suggested by one person.

E ***What do you dislike about the newsletter?***

The only comments in this section was that the photographs could be a bit clearer and that one reader was not too happy when the content of their article was changed.

The General Survey of the Newsletter was very good. We are grateful to those who returned the questionnaire and views expressed will be considered in the production of future editions.

There is just space here to squeeze in a picture of **Karen Agget** from Bridgend, Mid Glamorgan, with her daughter Carissa,aged four who suffers from Hurler disease and her son.



THE PORTAGE SYSTEM

Portage is a home teaching system for pre-school children with special needs.

History of Association

In the early 1970's a Portage home-teaching service was set up in Wisconsin, USA for pre - school children with special educational neeeds It was introduced to Britain in 1976 through a series of workshops, known as The Wessex Portage Workshops.

In 1983 a service for the community was founded and in 1986 the Department of Education and Science made available Education Support Grants.

How the system works?

Portage believes parents are the experts as far as their own children are concerned, a worker will work with both the child and the adult for one hour per week.

Parents are actively involved in the development of the child's abilities.

It is precision teaching that is breaking the tasks down into small manageable stages.

An activity chart is drawn up by the worker and parent together and on this is recorded what the worker and parent wants the child to achieve that week, the objects used and how much progress the child makes daily. At the next visit the child progress is assessed and improvements noted, the parents are encouraged to keep the charts but obviously it is not always possible to keep these records depending upon the child, the parents and the 'situations'. As the worker is in the home environment every week sometimes just the contact can be as valuable as the teaching.

Portage has its own check lists which are based on the following:

Socialisation, Self Help, Cognitive development, Stimulation, Language development.

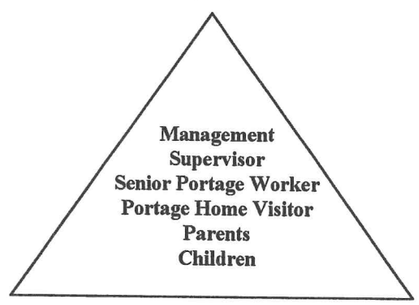
Meetings

There are regular meetings of the Portage workers to discuss the development of the children in their care and offer help and support to each other.

In Summary the Portage Approach is based on:

Weekly visits. Written activity charts with long term goals. Teaching and recording carried out by parents. Weekly supervision of the home visits.

Portage Pyramid



Archives of Disease in Childhood 1995; **72**: 263–267

PERSONAL PRACTICE

The mucopolysaccharidoses: a clinical review and guide to management

J E Wraith

The mucopolysaccharidoses are a group of inherited metabolic disorders caused by a deficiency of specific lysosomal enzymes. The enzyme deficiency results in interference with cellular function because of excess accumulation within the cells of partially degraded glycosaminoglycans (GAGS), which are also excreted to excess in the urine of affected patients.

Although the mucopolysaccharidoses were first described clinically by Hunter in 1917, their biochemical basis was not fully elucidated until the 1950s and 1960s. More recently the molecular biology of many of the subtypes has been described.[1]

The ubiquitous nature of GAGS within the connective tissue of the body results in a wide range of clinical effects. The type of GAG stored depends on the specific enzyme deficiency and classification of the disorders is now based upon these deficiencies, rather than clinical features. The table outlines present day classification with details of the accumulating compounds and enzyme deficiencies, as well as the eponym used for each condition. Excellent accounts of the biochemical basis of the mucopolysaccharidoses are available and this area will not be considered further.

In keeping with many other multisystem disorders affected patients are often under the care of many different clinicians and coordination of care can be difficult. A special clinic for children with mucopolysaccharidoses has been established in the Willink Biochemical Genetics Unit and this review outlines important management issues that occur with each particular subtype. The information is based on the clinical details of 225 patients who have attended this clinic from the UK and abroad. A breakdown of the patients has been appended to the table.

Willink Biochemical Genetics Unit, Royal Manchester Children's Hospital, Pendlebury, Manchester M27 4HA

Correspondence to:
Dr Wraith.

Classification of mucopolysaccharidoses and patients attending clinic

Type	*Eponym*	*Stored material**	*Enzyme deficiency*	*No of patients*
MPS IH	Hurler	DS, HS	Iduronidase	57
MPS IS	Scheie	DS, HS	Iduronidase	
MPS I H/S	Hurler-Scheie compound	DS, HS	Iduronidase	
MPS II	Hunter	DS, HS	Iduronate sulphate sulphatase	46
MPS III A	Sanfilippo	HS	Heparan-*N*-sulphatase	62
MPS III B		HS	*N*-acetylglucosaminidase	13
MPS III C		HS	Acetyl-CoA-glucosaminidase acetyltransferase	4
MPS III D		HS	*N*-acetylglucosamine-6-sulphatase	0
MPS IV A	Morquio	KS	Galactosamine-6-sulphatase	38
MPS IV B		KS	β-Galactosidase	0
MPS VI	Maroteaux-Lamy	DS	*N*-acetylgalactosamine-4-sulphatase	5
MPS VII	Sly	CS, DS, HS	β-Glucuronidase	0
Total				225

*DS=dermatan sulphate, HS=heparan sulphate, KS=keratan sulphate, CH=chondroitin sulphate.

General considerations

A multidisciplinary approach to management is necessary. Paediatric subspecialties, such as cardiology, anaesthesia, orthopaedics, otorhinolaryngology, ophthalmology, and neurosurgery, as well as many paramedical groups: for example physiotherapy, occupational therapy, audiology, speech therapy and psychology, will all have an important input. Diagnosis and evaluation is best undertaken in a major centre. The creation of our special clinic has allowed colleagues in the above related specialties to become highly experienced in the management of complications associated with the mucopolysaccharidoses. After assessment and counselling the subsequent care of the children can often be shared with the local paediatrician.

Regular assessments

As with many chronic progressive disorders the child has to be seen frequently enough to allow recognition of potential problems, leaving the parents free to enjoy the periods of better health. An annual assessment at the mucopolysaccharidoses clinic has proved valuable. Enough time is set aside to allow parents to express concerns and anxieties often not expressed in subspecialty clinics. Often a short stay in hospital can incorporate reviews by a number of subspecialists and in this way the progress of the disease can be monitored and complications recognised and treated early.

Anaesthesia

A number of reviews have highlighted the difficulties associated with this group of disorders.[2–4] A careful preoperative and perioperative management plan is required if disaster is to be averted. Intubation is often extremely difficult, although this has been helped by the use of fibreoptic laryngoscopy and bronchoscopy. In difficult cases the laryngeal mask airway has proved to be a useful additional aid. Anaesthesia must be undertaken only by anaesthetists expert in maintaining paediatric airways and in centres with adequate intensive care facilities. In the severely affected patient operations should only be performed for life threatening complications or in an effort to greatly improve the child's quality of life. Minor or cosmetic procedures should not be undertaken or should be performed under local anaesthesia.

Eponyms, heterogeneity, and phenotype

The mucopolysaccharidoses are most commonly known by their eponomous titles, as outlined in the table. However, as our understanding has increased it has become increasingly clear that these are a gross simplification. Most commonly the disorders are divided into 'mild' and 'severe' variants. For example, in MPS I the severe variant is known as Hurler's syndrome and the mild variant Scheie's syndrome. Anything apparently not conforming to this division is called Hurler/Scheie syndrome by most paediatricians. It is now apparent, like most genetic disorders, that there is a continuous spectrum of phenotype from the very severe to the most mildly affected. Genotype analysis, where known, in this group of disorders has confirmed that many different mutations are responsible for these phenotypic differences. It is preferable now to speak in terms of enzyme deficiency rather than eponym, but as the latter are so well established it will take time for this to achieve universal acceptance.

Presenting features

Patients with a mucopolysaccharidosis usually present in one of three ways: (i) as a dysmorphic syndrome, for example MPS I, MPS II, MPS VII; (ii) with severe behavioural disturbance and dementia, for example MPS III; and (iii) with evidence of a severe bone dysplasia, moderate dysmorphism, and normal intelligence, for example MPS IV, MPS VI.

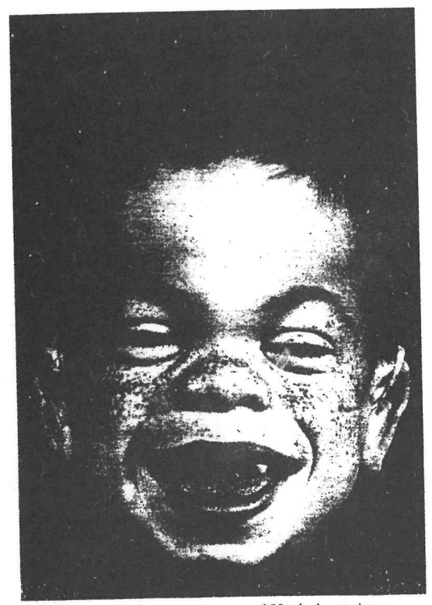


Figure 1 Typical facial features of Hurler's syndrome.

Particular problems associated with each enzyme deficiency

MUCOPOLYSACCHARIDOSIS TYPE I, IDURONIDASE DEFICIENCY (MPS IH, MPS IS, MPS IH/S, HURLER'S SYNDROME, SCHEIE'S SYNDROME AND OTHER VARIANTS)

Fifty seven patients with iduronidase deficiency have been assessed in the unit and the full clinical spectrum ranging from death in early infancy due to cardiomyopathy to near normal adult patients has been observed. The commonest phenotype is that of 'classical' Hurler's syndrome and a number of these patients have been found to be homozygous for a recently described common mutation in the iduronidase gene.[5] The characteristic dysmorphism associated with this disorder is familiar to all paediatricians (fig 1).

In the severely affected patient the clinical course is dominated by airway problems and frequent upper and lower respiratory infections are common. Diagnosis should be followed by immediate referral to the ear, nose, and throat clinic as all affected children require treatment for upper airways obstruction and middle ear disease. Obstructive sleep apnoea is an almost universal finding. Initially this can be helped by tonsillectomy and adenoidectomy, but many patients require nocturnal oxygen treatment later in the course of the illness.

Cardiac disease is very common and presentation and early death from cardiomyopathy is a recognised complication of this disease.[6] In other patients asymmetrical ventricular septal hypertrophy is a frequent finding early in the disorder and is often followed by variable thickening of the mitral and aortic valves. Coronary insufficiency is known to occur and sudden death from arrythmia has been assumed in some patients.

Despite these potentially severe physical problems early intellectual development is usually normal and affected patients do not have severe learning difficulties until much later in the course of the disease. The majority of children attain some social skills and many are capable of starting normal nursery schools. Mobility may be limited by joint stiffness which is progressive, but usually not painful, and by the protuberant abdomen due to hepatosplenomegaly. The latter is also probably responsible for the high incidence of umbilical and inguinal hernias seen in these patients.

Hydrocephalus requiring shunt insertion was noted in six patients. If ignored the head can reach enormous proportions and make nursing of the child difficult in the latter stages of the illness.

Corneal clouding can be very variable. Glaucoma is often quoted as a complication of MPS I, but was seen in only one patient in this series. Sudden blindness occurred in one patient with no apparent cause, electroretinograms were normal, but visual evoked

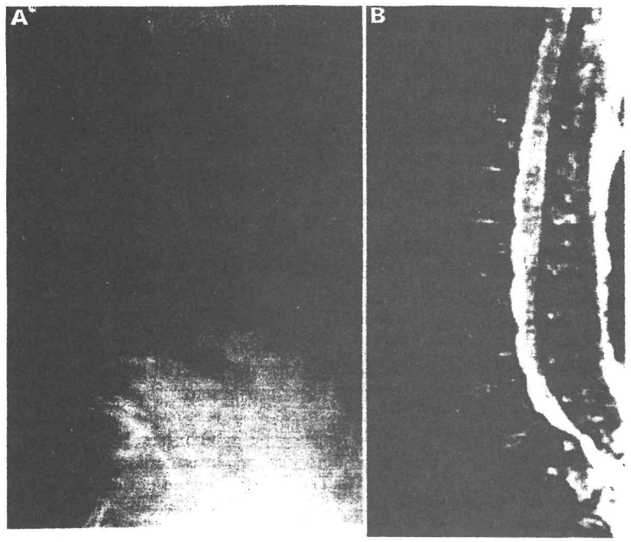


Figure 2A Radiograph of the lumbar-sacral spine in a patient with MPS IS showing the typical lumbar-sacral spondylolisthesis. (B) Magnetic resonance image of the same patient showing narrowing and compression of the spinal canal at the level of the spondylolisthesis.

responses absent. Magnetic resonance imaging of the visual pathways was normal.

Death usually occurs in the first decade of life and is most often due to cardiorespiratory failure.

In the adult patient with iduronidase deficiency, the dysmorphic features are often very mild and there are different priorities with regard to management. The clinical picture is usually dominated by the orthopaedic complications of the disease. Poor hand function, partly due to chronic carpal tunnel syndrome is extremely common.[7] Progressive joint stiffness and severe back pain are usual. Radiography of the lower lumbar spine has shown a very high incidence of spondylolisthesis in these patients and in some this has been associated with spinal cord compression (fig 1A and 2B). As the patient gets older corneal clouding may interfere with vision and corneal grafting has been performed on two patients. In these circumstances it is essential to confirm that the loss of vision is not due to retinal disease.

In older patients mitral and/or aortic incompetence are common.

The oldest patient seen in the clinic is currently 48 years of age: she shares the same genetic mutations as Scheie's original patient.[8]

MUCOPOLYSACCHARIDOSIS TYPE II, IDURONATE SULPHATE SULPHATASE DEFICIENCY (MPS TYPE II, HUNTER'S SYNDROME)

Forty six patients with this variant have been reviewed. The vast majority of patients have been severely affected (35), but again a spectrum of clinical effect can be observed. Severely affected patients share many of the features of classic Hurler's syndrome, but tend to be milder and have less severe skeletal involvement. The classic nodular rash said to be pathognomic of this disorder is rare and has been observed in only two patients. Once again ear, nose, and throat and airway problems dominate the clinical course. In addition most patients are prone to severe bouts of diarrhoea, the cause of which remains obscure. In addition we have observed other unusual manifestations of gastrointestinal dysfunction not previously appreciated, such as, spontaneous perforation of the stomach and intestinal pseudo-obstruction.

Progressive neurodegeneration results in a vegetative existence for most patients from early teenage years with death around the age of 15–16 years.

In mildly affected patients cervical myelopathy due to dural hyperplasia and thickening of the ligamentum flavum is probably inevitable. In patients with declining exercise tolerance or upper motor neurone signs in the limbs, magnetic resonance imaging of the craniocervical junction should be performed. At the same time the airway can be imaged and an assessment made of the inevitable anaesthetic problems in this group. No operative procedure should be performed on a mildly affected MPS II patient until it has been established whether or not cervical cord compression is present. The manipulation involved in a difficult anaesthetic can lead to sudden cord compromise and result in quadriplegia.

It is interesting to note that even in mildly affected patients with normal intellect that imaging of the central nervous system is often grossly abnormal (fig 3).

The genetics of this mucopolysaccharidosis differ from the others as the disorder is inherited as an X linked recessive. Only a minority of affected boys have a demonstrable deletion in their iduronate sulphatase gene (20%), the

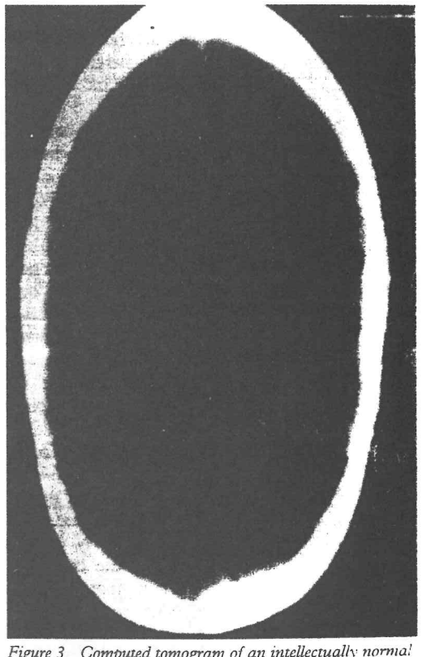


Figure 3 Computed tomogram of an intellectually normal man aged 24 years with MPS II. Note the ventricular dilatation and 'cyst-like structures' in the cerebral matter.

majority of defects being various point mutations.[9]

MUCOPOLYSACCHARIDOSIS TYPE III (MPS IIIA, IIIB, IIIC, IIID, SANFILIPPO'S SYNDROME)

In this disorder four enzyme defects are known to cause an identical clinical phenotype. This is the commonest of the mucopolysaccharidoses in the UK and its management has been reviewed recently.[10] Seventy nine patients with MPS III have been assessed. Again, heterogeneity produces a variable clinical phenotype and a spectrum of effect is noted. Some patients are very mildly affected and it may be that this disorder is underdiagnosed. Mild dysmorphic features are combined with a very severe behavioural disturbance and the disorder is extremely difficult to manage.

The molecular basis of the commonest variant of MPS III is yet to be established.

MUCOPOLYSACCHARIDOSIS TYPE IV (MPS IVA, MPS IVB, MORQUIO'S SYNDROME)

No patients with MPS IVB have been seen in the unit; this appears to be a rare variant in the UK. Thirty eight patients with MPS IVA have been seen and assessed. Affected patients are of normal intelligence and the clinical picture is dominated by a severe bone dysplasia. Skeletal radiographs are often abnormal from birth, but diagnosis is usually established in the first year of life after parents notice the obvious spinal deformity associated with this disease.

Motor milestones are achieved at the normal times, but the gait becomes increasingly abnormal due to the development of severe genu valgum secondary to ligamentous laxity; this, combined with the increasing sternal protrusion and severe pes planus, leads to a very characteristic posture. In the severe form of the disease height prognosis is extremely poor and few patients will be taller than 100 cm as adults.

The major complication of this mucopolysaccharidosis is the development of neurological abnormality secondary to cervical myelopathy (also occurs in MPS I but is less common). This may be of sudden onset resulting in death after a simple fall as has occurred in two of our patients and a number of their siblings, not seen in the unit, or it may occur insidiously with a lack of exercise tolerance predating frank neurological abnormality. This complication must be looked for with vigour and we currently perform magnetic resonance imaging of the craniocervical junction on an annual basis in patients with MPS IV. It is our practice to perform prophylactic occipitocervical fusion in patients with instability. We do not wait for the onset of neurological abnormality. The instability is due to a combination of a hypoplastic, poorly ossified dens and ligamentous laxity in the cervical spine. What has become clear is that successful fusion in the upper cervical region is often followed by progressive instability further down the cervical spine and multiple fusions may be required in some patients. This complication is most commonly seen in patients who remain independently mobile after the procedure. In patients who are chair bound before surgery, sitting posture may lead to subluxation of vertebrae lower down the spinal column, for example in the mid-thoracic region.

Affected patients are not dysmorphic, but do have an increased incidence of ear, nose, and throat disease and in addition have characteristic teeth which are prone to dental caries. Fine corneal opacity is usual, but never as severe as patients with MPS I. Aortic incompetence is a usual finding in the adult patient. Life expectancy should be good if cervical complications are avoided. Some patients, however, develop progressive cardiorespiratory disease secondary to their restricted chest movement.

Again, there is a very wide spectrum of clinical phenotype which has been broadly divided into severe, intermediate, and mild MPS IV. The mildly affected patient can be of normal adult height and in our patients hip stiffness and pain appears to be the commonest mode of presentation.

As adults patients with MPS IV often develop aches and pains in various joints. We assume that this is 'wear and tear' accelerated by the abnormal joint posture. The symptoms respond variably to analgesia and non-steroidal anti-inflammatory agents.

It has been proposed that the prognosis for girls with this disorder is better than for boys and this was attributed to their 'less active lifestyle'.[11] In our patients we can detect no difference in the severity of the disease between the sexes and the incidence of cervical cord problems is the same in both groups.

MUCOPOLYSACCHARIDOSIS TYPE VI, *N*-ACETYLGALACTOSAMINE-4-SULPHATASE DEFICIENCY (MPS VI, MAROTEAUX-LAMY SYNDROME)

This mucopolysaccharidosis appears to be very rare in the UK. Only five patients have attended for assessment and follow up. Again this is a heterogeneous disorder and is usually considered to the 'mild' because of the retention of normal IQ by affected patients. The disorder should not, however, be considered benign. In addition to relatively easily treated early complications such as ear, nose, and throat disease or carpal tunnel syndrome there are some more serious potential problems. Presentation with endocardial fibroelastosis[12] or cardiomyopathy[13] has been reported, and in addition cervical myelopathy is known to occur.[14] The most serious complication, as there is no effective treatment, is the development of progressive, diffuse airway narrowing leading to cor pulmonale and death in late teens or early twenties.

In theory, MPS VI is the one mucopolysaccharidosis most suitable to 'curative' treatment by enzyme replacement, as the central nervous system is not affected. There is also an excellent feline animal model which makes assessment of such treatment appropriate before human trials.

MUCOPOLYSACCHARIDOSIS TYPE VII, β-GLUCURONIDASE DEFICIENCY (MPS VII, SLY'S DISEASE)

This disorder is extremely rare and no affected patients have been seen. Indeed, in our laboratory where we receive annually over 1000 blood samples for lysosomal enzyme assay and over 500 urines for mucopolysaccharide analysis we have encountered this disorder on only two occasions in the last 10 years. The phenotype of affected patients is again very variable and ranges from presentation with hydrops fetalis to a relatively mild Hurler phenotype presenting in adult life.

OTHER DISORDERS

While not the purpose of this review it should be noted that there are a number of storage disorders which produce a phenotype not dissimilar to the mucopolysaccharidoses. In addition to the patients outlined above we have also seen and assessed 20 patients with mucolipidosis II and III, eight patients with mannosidosis (5α and 3β-mannosidase deficient), and one with sialic acid storage disease. All presenting initially as possible mucopolysaccharidosis patients.

Discussion

The mucopolysaccharidoses are a group of devastating disorders and parents need considerable support in dealing with an affected child. The mucopolysaccharidoses society* has offered many parents and affected individuals considerable help as well as raising significant sums of money for research.

A sound understanding of the phenotypic possibilities within each subgroup allows for a logical approach to management which is inevitably multidisciplinary. The paediatrician should play a lead part in this process and coordinate services for affected patients. The disorders should no longer be regarded as 'untreatable' as quality of life for many individuals can be greatly improved after relatively simple procedures.

Finally, it is important to remember that these are genetic disorders. First trimester prenatal diagnosis is possible for all of the subtypes.

Thanks to Professor John Hopwood and colleagues, department of chemical pathology, Adelaide Children's Hospital, for molecular analyses on many patients.

*The Society for Mucopolysaccharide Diseases, 55 Hill Avenue, Amersham, Bucks HP6 5BX.

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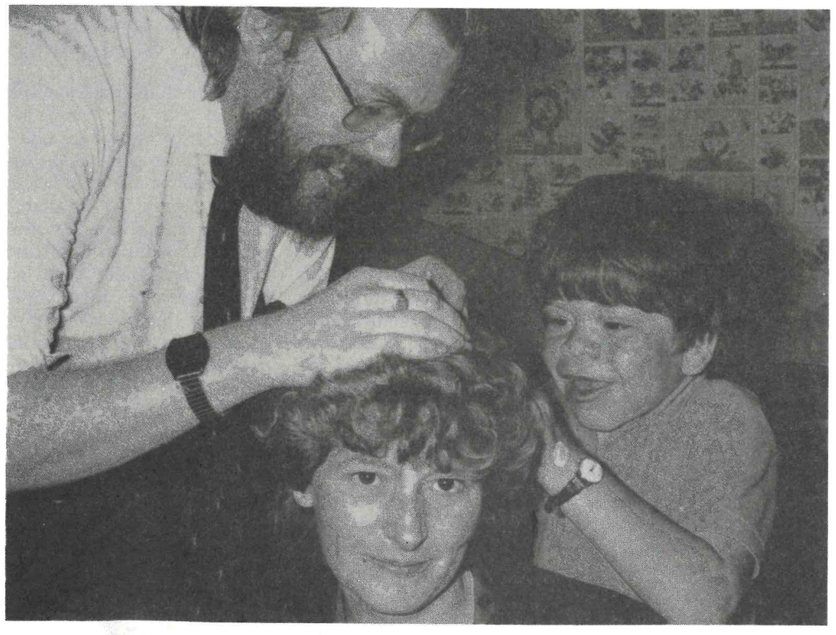


"Carrier Testing" Dr Ed Wraith is helped by Colin Arrowsmith to take a hair root sample from Barbara Arrowsmith.

OFFICE HOURS

"This is the Society for Mucopolysaccharide Diseases....the office is now closed ....."

How many times have you phoned the office, only to hear our answerphone? In order to help you phone when someone will be there to take your call, we are detailing below the times when the two offices are usually open and who you are likely to speak to.

Please bear in mind that sometimes Christine and Mary work away from the office.

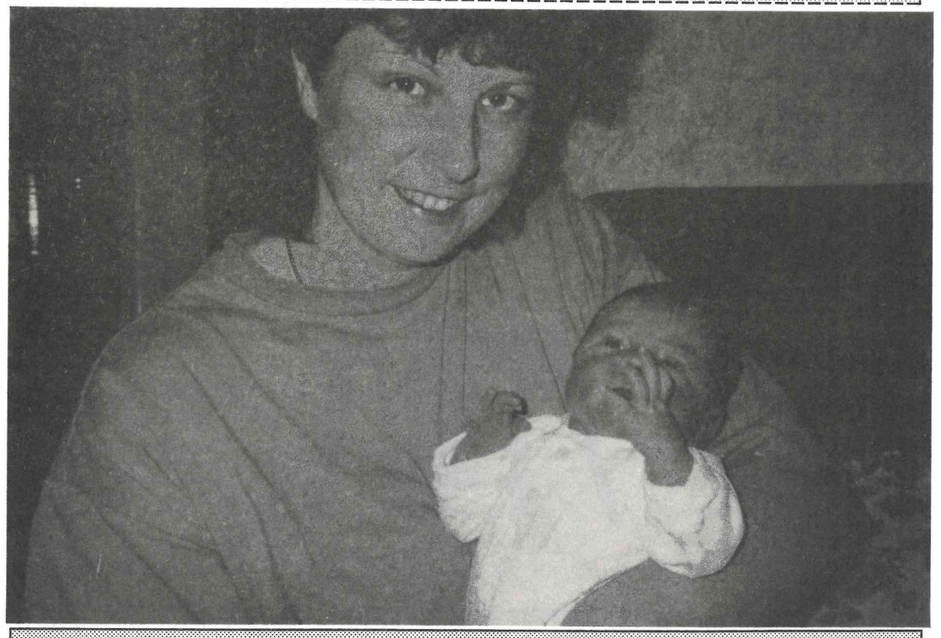
In case of an urgent enquiry out of hours you may try Christine and Mary at home where you may, if they are not available, leave a message.

MPS office hours:

9.00am - 5.00pm	**Mon, Tues, Wed, Thurs**	**Joan**
9.00am - 3.00pm	**Mon, Wed, Fri**	**Sheila**
9.00am - 3.00pm	**Tues, Thurs**	**Sue**

Northern MPS office hours:

10.00am - 3.00pm	**Mon, Tues, Wed, Thurs**	**Pam**



Births

Congratulations to Bob and Rhian McKnight from Lougharne, Dyfed on the birth of their daughter Rhoswen Amy born on the 27th of February 1995.

The Association for Children with Life-Threatening or Terminal Conditions and their Families (ACT) is concerned that some children are not being referred to regional centres for specialist treatment.

Every child is entitled to the best current treatment and expertise in the management of their particular condition. Access to such specialist care and information can be of such tremendous value to children and parents, and in the case of MPS can improve prognosis or the quality of life for our children. Some local practitioners are reluctant to refer families when long distances are involved, but our two regional specialists in London and Manchester and their teams are willing to undertake shared care with local professionals. ACT and MPS would like to hear from anyone who has experienced difficulties in obtaining a referral to either specialist centre of expertise. The information will be held in the strictest confidence and will only be used as statistical evidence without reference to particular individual families or professionals involved with them.

Stella Elston
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copy to : MPS Society
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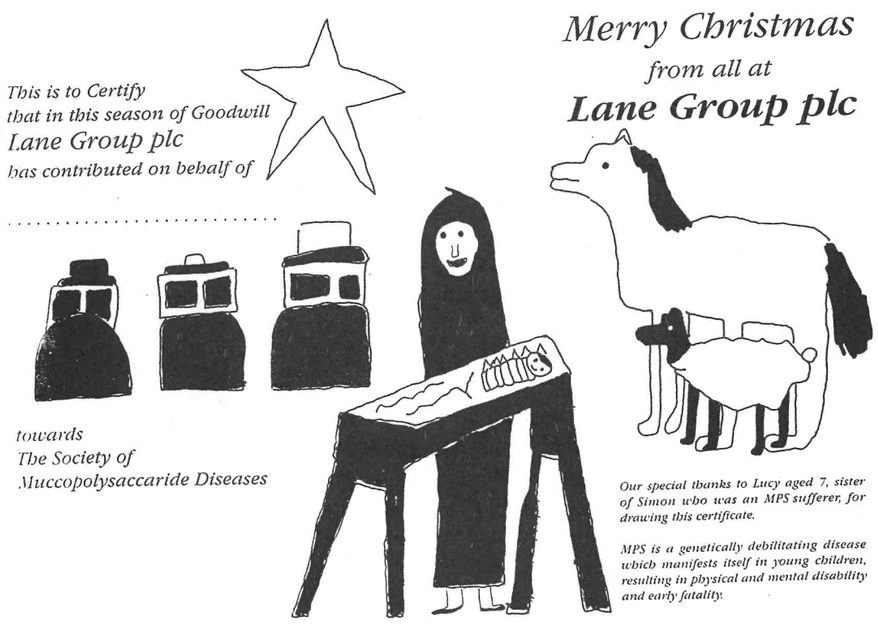

If you work for a company that sends out small gifts to clients at Christmas why not persuade them to send out charity donation certificates, like the one above instead? This simple idea raised £1,500.

The Society is grateful to the following who held Fund-raising Events

St Mary and St Paul CE Primary School	South Harting	Collection at school play
Pauline Mahon	Sheffield	Raffle
Mervyn Insley	Billingham	Race Night
Ron Thompson	Darlington	Raffle
Peter and Valerie Morley	Bath	Bike Ride
Tracey and Lindsey Hawkins	Wokingham	Xmas pub collection
UK Truck Stops	Midlands	Staff Xmas parties
St Paul's School	Shepton Mallet	School concert
Chesham High School	Chesham	Charity Day
Andy and Jenny Hardy	Haddenham	Soup morning
Haddenham P. CC	Haddenham	Golf
John and Martine Brennan	Lancaster	various fundraising
Julie Burlison	Oldham	Coffee morning
Metropolitan Police	London	Golfing tournament
Lane Group	Bristol	in lieu of Xmas gifts
Karen Weedall	Runcorn	Selling Webb Ivory
Laurie Davis	Saffron Walden	sponsored maths worksheets
Catherine Rush	Tunbridge Wells	Sale of tapes
9th Supply RLC	Hullavington	Regimental dinner
R 'N' M Trust	Chesham	Dancing competition
Mike, Ann and Sarah Kilvert	Nantoer	Bingo evening, stall

The Society is grateful to those who have made donations

Treowen School	Larkrise School	'Freemasons' Grand Charity
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Mr and Mrs Greenwood	Mrs S Pascall	Ann and Mervyn Canton
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Natalie Pidden's Grandma	Dr A Galbraith	Sue Balmforth
Mr and Mrs Johnson	St Michael's Playgroup	

DONATIONS IN MEMORY

The Society is grateful to the friends and relatives of:

John Leonard
Christelle Voumard
Mark Harvey
Gethin Robins
Mr Frain
Darren Tailford
John Hodgetts
Lloyd Stewart
P J North

SPONSORED EVENTS AND APPEALS

The Society wishes to thank all those who supported:

Edward Nowell Appeal
Tesco Collection, Amersham

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Mr Peter Maver
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Lothian and Borders Police Communications Department.

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Danetre School, Daventry

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