Newsletter

The Society for Mucopolysaccharide Diseases

National Registered Charity No. 287034

Spring 2003

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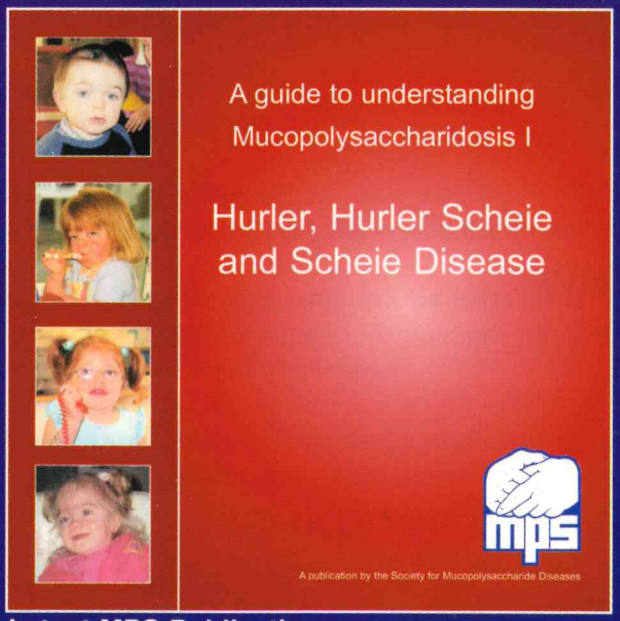

Latest MPS Publications

Register now for your 10K Road Race Place

Members' News

Help Design an MPS Tea Towel for the MPS 21st Birthday

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SOCIETY NEWS

'CARE TODAY, HOPE TOMORROW'

What is the Society for Mucopolysaccharide Diseases ?

The MPS Society is a voluntary support group founded in 1982, which represents from throughout the UK over 1000 children and adults suffering from Mucopolysaccharide and Related Lysosomal Diseases, their families, carers and professionals. It is a registered charity entirely supported by voluntary donations and fundraising. It is managed by the members themselves and its aims are as follows:-

- **To act as a Support Network for those affected by MPS diseases and related diseases**
- **To bring about more public awareness of MPS and related diseases**
- **To promote and support research into MPS and related diseases**

How does the MPS Society meet these Aims?

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

Telephone Helpline
Includes out of hours listening service

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

Support to Young People and Adults with MPS
Empowering individuals to gain independent living skills, healthcare support, further education, mobility and accessing their local community

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

National and International Conferences
Held annually and offering families the opportunity to learn from professionals and each other

Sibling Workshops
Specialist activities for siblings who live with or have lived with a brother or sister suffering from MPS or a related disease

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

Quarterly Newsletter
Containing information on disease management, research and members' news

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

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

Chairman's Report

Barry Wilson

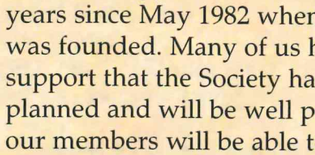
Hello and welcome, particularly to our new members. It has been almost a year since I last reported to you. As you might expect there has been much going on from our Amersham office during that time. As usual the office staff have been as busy as ever, coping with all the changes that legislation has demanded. Some changes in personnel have occurred, and you will have read all about these in the past newsletters. Throughout these changes, which have enlarged the advocacy support team, the Society continues to provide a first class advocacy support service to its members on a needs led basis. Don't forget there is always somebody there whom you can talk to.

This coming year is an occasion to celebrate. **The Society is coming of age**. It is 21 years since May 1982 when things really started to happen and the MPS Society was founded. Many of us have been grateful of the help, advice, information and support that the Society has given us over that time. A number of events are planned and will be well publicised in the coming newsletters and I hope that all our members will be able to participate in at least one of these events.

On the 10the May 2002 the Society's Annual General Meeting will be held at the Alton Towers Hotel at 9.30am. All member families attending can purchase up to two adult tickets and a ticket for each of their children under 18 years for an unbeatable price of £20. A booking form is enclosed and ticks can be collected at the AGM. We need at least 40 members present at the AGM and look forward to seeing as many of you as possible on 10 May.

Our forthcoming conference in Northampton, over the weekend commencing Friday 20the June 2003, is already virtually a sell out! Those of you that have attended in previous years will know just how valuable and enjoyable these conference weekends can be. So, if any families or, indeed, professionals are thinking of attending, please contact the Society as soon as possible.

Another event to take place on the 13the July 2003 will be the 'Teddy Bear's Picnic' in the Childhood Wood. I would like to welcome as many of our bereaved members as possible so that we may all reflect on and celebrate our childrens' lives. We hope that many, if not all of you bring along your teddy bears, each with their own personal story. After lunch the Forestry Enterprise Wardens will provide activities for the children and this will be followed by a memorial balloon release.



Barry Wilson
Chairman

Annual General Meeting of the Society for Mucopolysaccharide Diseases will be held at 9.30am on Saturday 10the May 2003 at the Alton Towers Hotel, Staffordshire.

Advocacy Support Diary For 2003

April

East Anglia Clinic:- Friday 4the April

Cardiff Clinic:- Wednesday 30the April

May

Bristol Clinic:- Thursday 1st May

AGM:- Saturday 10the May

Scottish Clinic:- Thursday 15th May

Scottish Conference:- Friday 16th May

Northern Ireland Clinic:- Wednesday 21st May

June

House of Commons tea party:- Wednesday 4th June

National Conference – Northampton:- 20th – 22nd June

Birmingham Clinic:- Friday 27th June

July

Remembrance day & Teddy Bears Picnic in the Childhood Wood:- Sunday 13th July

October

Jeans for Genes:- Friday 3rd October

Tree Planting- Childhood Wood:- Friday 24th October

November

Cardiff Clinic:- Wednesday 5th November

Bristol Clinic:- Thursday 6th November

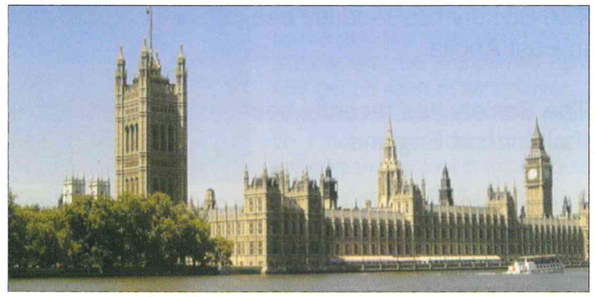
Northern Ireland Clinic:- Wednesday 12th November

Northern Ireland Conference:- Thursday 13th November

UK wide information days will take place throughout the year.

The advocacy support team remain committed to establishing a regional clinic in the South East of England.

10th May - The Alton Towers Hotel is hosting the AGM



4th June - House of Commons tea party

24th October - Tree Planting - Childhood Wood

3rd October - Jeans for Genes Day

New Members

Mrs Cessford has recently contacted the Society. Her son Colin has Fabry Disease. The family live in Scotland.

The Society has recently been contacted by Mrs Dickinson who has Fabry Disease. She lives in the Home Counties.

The Society has been recently contacted by Steve Horrocks who has Fabry disease. He lives in the North West.

Carl and Clare's son Landon has recently been diagnosed with Sanfilippo disease. Landon is five years old. The family live in the Midlands.

The Society has recently been contacted by Carole Lunnon who has Fabry disease. She lives in East Anglia.

The Society has recently been contacted by Ian Roebuck who has Fabry disease. He live in the North of England.

The Society has recently been contacted by Darren Swayne who has Fabry disease. Darren live in the North of England.

Deaths

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

Jeremy Papworth who suffered from Sanfilippo disease 02.02.74 - 15.03.03

Emma McLellan who suffered from Sanfilippo disease 08.05.91 - 13.02.03

MY SISTER

I never want to be apart
I've loved you with all my heart
On the days I'm feeling sad
You are the one who makes me glad
You've always been a sister who cared
You've always been a friend who shared

Goodbye

Written by Issy Gee

Befriending Link

The Advocacy Support Team had an enquiry from a family with a 13 year old son with Aspartylglycosaminuria (AGU). They would like to speak to others in a similar situation and asked if the MPS Society could help. Although this family is based in the UK, we would welcome responses from across the world. So if you have a child who has AGU and would like to share your experience with another family please contact Alison at the MPS Office.

Living With Morquio Disease

Adika Batool

Adika and sister in their shared bedroom

My Adaptation - Adika Batool

At the moment I am waiting for my new bedroom and bathroom to be built which has been promised to me for a very long time by social services. I was always told it would be next month next month and then I was told it would be next year. The room is taking so long to be built and it makes me very mad having to wait all this time, as I need my own room. My sister and I share a room and we sometimes fight and get on each other's nerves. I can't wait to have my new room so I can have some privacy. I have been told that it will be built soon. I really hope it is.

The good news is I also have a new electric wheelchair, which is bright yellow and black which was given to me by Whizz Kids.

Adika goes to Tunisia

It all began on the 19the January 2003 at Manchester Airport. It was a very long journey to Tunisia and I was very frightened about being on the plane. My mum hugged me and because my ears were hurting my mum told me to pretend I was sucking on a sweet. The food on the plane was disgusting and I did not like it.
The people at the airport were very helpful and kind.

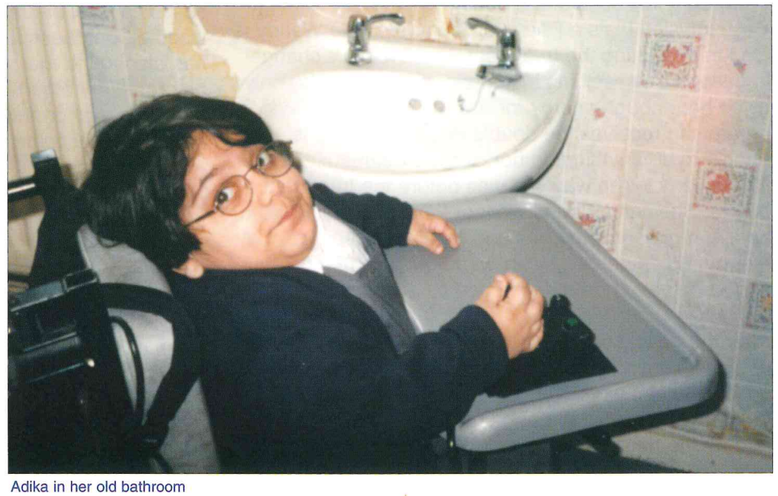
When we arrived in Tunisia it was very cold and dark. The hotel we stayed at was very welcoming and our accommodation was on the ground floor. I made lots of friends. One in particular was called Mohammed and he was very kind and caring and helped me around the shops. My friends and I used to go out a lot and come back laughing. My mum used to ask what we were all laughing about. One night at the hotel we watched some cabaret dancers and they were very funny as they were men dressed up as women. One was very tall and one was very small.

We went out quite a lot on holiday to theme parks, on Safaris and to the dessert. My mum went on a camel and I went out in carriage pulled by a donkey.

I was very upset when I had to leave and lots of tears were shed. I still keep in touch with the friends I made in Tunisia.

My School - Adika Batool

I really like the school I am currently at as I have lots of friends who look after me. I hope to move up to secondary school with them this year but I may have to go to another school if my appeal is refused. I do not want to go to another school, as I will miss my friends. Some children can be very nasty and ask why I am so small, why do I need a wheelchair and what is wrong with me. Sometimes I get very upset and my friends stick up for me and look after me when I cry. They tell me to take no notice but it hurts sometimes. This is why I do not want to go to another school as I will be on my own and I will not have my friends there to say everything will be alright and make me laugh.



Adika in her old bathroom

A Personal experience of Fabrazyme

Wendy Douglas

Wendy is 26 years old. She was first diagnosed as having Fabry Disease when she was 8. At this time she suffered limb pains in her joints and especially in her fingers and toes. Wendy found that whenever she had a sore throat or slight infection this would make her feel more run down and trigger the pain. Throughout her school life Wendy could never join in any exercise as this would also trigger severe pain episodes.

Wendy's health as an adult has not been good. She needed painkillers regularly for the joint pain and, besides a constant background pain, often suffered severe pain crises brought on by exertion, damp weather or extreme heat. She had no energy at all, feeling constantly shattered, and would often lie in bed all day as the effort to get up and get dressed, let alone go anywhere, was too great. It was impossible for Wendy to work and she had little in the way of a social life as she couldn't keep up with her friends.

In addition to this, Wendy began to suffer constant backache and increasing episodes of kidney infection. She also began to suffer palpitations and was put on beta-blockers for an irregular heartbeat.

2 years ago Wendy was admitted to the hospital for an MRI scan after 3 occasions on which she lost all feeling in her left leg. This was attributed to her Fabry Disease and Wendy was told that her heart was being affected as well as her kidneys. These episodes of loss of feeling, she was told, were like mini-strokes.

Wendy was asked to start the trial for Fabrazyme at Hope Hospital in Manchester but, at this time, she was pregnant and so refused. Last year she was asked again and in August 2002 she had her first infusion.

Wendy has now had 14 infusions and alternates between a low dose and a double dose every fortnight. Each infusion lasts a little over 2 hours. While Wendy is glad to be getting the treatment, she finds this change in dosage an unsettling experience. The low dose only gives her enough energy for a week and in the days before she receives the double dosage, Wendy is unable to do anything. In her own words, her life stops. She reverts to the way she was before the treatment began. She cannot go to work or look after her 3 and a half year old son, who has and requires a great deal of energy. Wendy doesn't want to go out with friends and wishes to be at home and in bed by 10 o'clock.

After the double dose however, Wendy reports a feeling of overwhelming energy which lasts through to the next dose. She can work 20 hours a week at Thomas Cook easily and still have enough energy to play with her son and do the housework as well. Should friends ask her out for a meal in the evening she knows that she can go out and enjoy herself without feeling shattered halfway through the night or suffering the next day.

Wendy has found that the palms of her hands sweat now where she could never sweat before and the background joint pain has not worsened. Severe pain crises can still be triggered by exertion and extremes in weather, but these are not as regular as before. Wendy has also found that where she never used to get spots before, every other week she now gets them and she has had to go on the Pill to help regulate her periods which have become erratic since starting treatment in August.

Wendy is very happy about being on Fabrazyme. She is very grateful to be getting ERT as, while she knows it isn't a cure, it will prevent the disease from getting any worse. Wendy has found that one of the benefits of ERT is that the infusion can be done at home, which is much more relaxing than having to go to a hospital, and she is currently learning how to administer it herself. Wendy would recommend ERT to anyone and they only have to look at how it has changed her life to see why.

Equipment Ideas And Exchanges

Camper Van Adaptation

I am just replying to the query about camper van adaptation, in the winter 2002 newsletter.

We bought a camper van last year, 2002, this was a Volkswagen, the newer type, not the old type, often used by surfer's now! We thought this would be ideal, as during the summer months, we go camping most weekends, taking Georgia with us, as she loves it.

The camper van was intended to sleep 2 adults and 2 children, however, as my husband is 6ft 4 and Georgia is big for her age (Sanfilippo, age 7), it was extremely uncomfortable for the 3 of us to sleep in. Eventually, we resorted to Georgia sleeping in the camper van, using up the double bed space, and me & my husband sleeping in our tent, with the opening attached to the camper van, so nobody could get in either way! This wasn't at all practical, and totally defeated the object of having a camper van in the first place!

I daresay, if we had spent more money and bought a bigger camper van, we wouldn't have had this problem, but we didn't fancy buying a really big one. Since then, we have sold the camper van and bought a trailer tent, which has more space. We will be trying that for the first time this summer! Sorry that I didn't have more constructive advice, but I just thought I'd let you know of our experience anyway.

Louise Lewis

Multi-Sensory Equipment

Multi-sensory equipment that would like to go to a new home. The equipment consists of a fibre optic curtain, bubble tube, projector and slides, rope lights, illuminated drawing board with fluorescent markers, and fibre optic lamp. For more information please contact Alison at the MPS Office.

Wash Basin

If you find it hard to use a normal wash basin, then check out the Astor Banneman adjustable height wash basin. It lets wheelchair users get under the basin without bashing wheelchair joysticks, and the basin is sunk into a surrounding surface, which offers support if your arms are weak. The height of the sink can be controlled by an electronic handset, and there is an automatic safety cut-out, which means your legs won't get trapped underneath. The sink also contains touch operated taps and optional infra-red touch sensors. Tel. 01242 820820

Indestructible Dummy

Just replying to the query, in the winter 2002 newsletter, about finding an indestructible dummy!

Well, Georgia (sanfilippo, age 7) still loves to have her dummy at bedtime. I've been through hundreds, I'm sure, as she constantly bites and chews on them! However, I must say that the best one, and it's still going strong, was bought from our local Spar shop. They have them hanging up on a cardboard sheet called 'Galpharm' 123 Baby Soothers, by all the baby products and they cost approximately 59p! so, it's a real bargain.

This is the only one I've found that can withstand the treatment it gets from Georgia! Hope this helps!

Louise Lewis

Quality Bibs

Quality Bibs for mealtimes (and in between) that are effective, dignified and long lasting. Made in cotton towelling with a supple waterproof backing that does not crack or split.

Designed by a carer, for carers & wearers. For more information tel. 01834 814 652.

New Home Wanted For Manger Electric Wheelchair

Sarah's Manger chair was bought in 1998 and she has had it serviced every six months. The base is fine but it will need a new cushion. Sarah sayes she would not necessarily require any money for it (it would help!) but be please to see it go to a good home. Contact: sarah@longconsult.fsworld.co.uk

New Home Wanted For Small Arm Chair

Has fully washable cover and suitable for a young person with Morquio's. Sarah says it is a lovely chair which supports your back and doesn't look like a "special" chair. It is adult looking.
Contact: sarah@longconsult.fsworld.co.uk

Birmingham Clinic - 24th January 2003

Sophie Denham

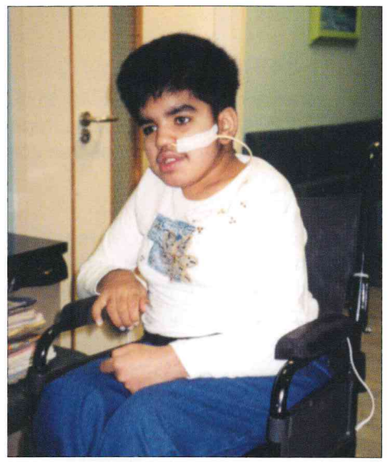
On the 24th January, the Birmingham MPS clinic took place at the Birmingham Children's Hospital. The clinic as ever was very busy but ran relatively smoothly.

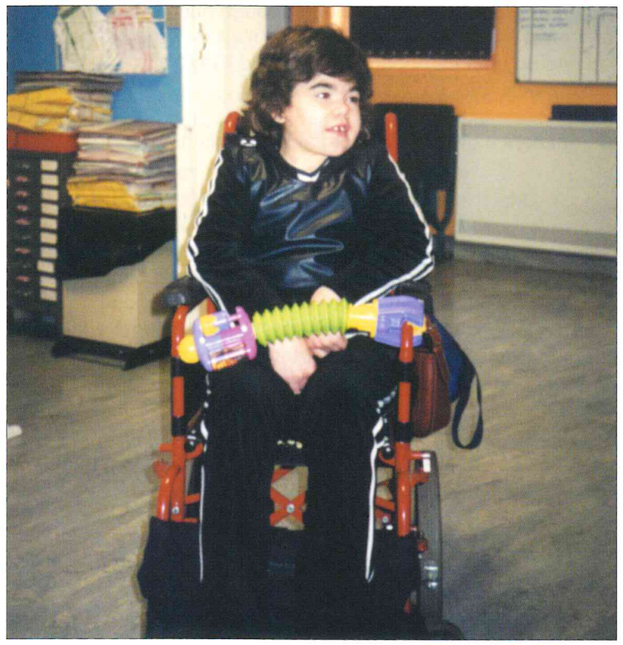
This clinic covers those individuals who live within the West Midlands area. At this clinic we were able to see thirteen individuals and their families.

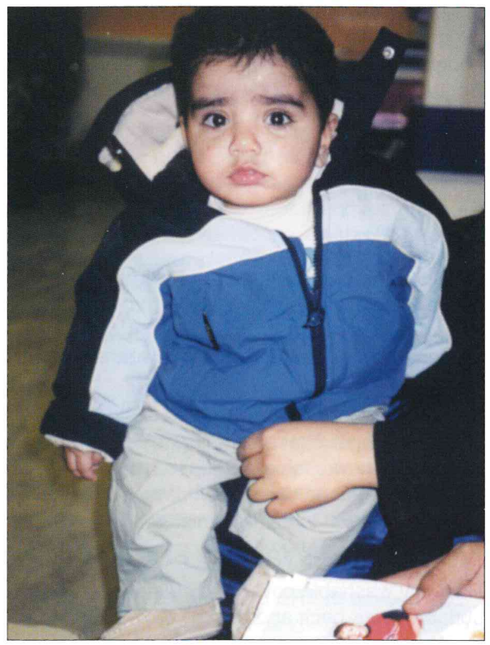
This clinic not only enables the families to meet Dr Chakrapani and Dr Wraith but also gives the Advocacy Support Team the opportunity to meet with new families as well as old.

We appreciate that the waiting area afforded to us is not ideal at the clinic and we are liasing with the hospital in looking at an additional room being made available. The additional room would be used to give families the opportunity to speak with the Advocacy Support Team in confidence.

Our thanks go once again to Dr Wraith, Dr Chakrapani and nurse specialist Joy Wright for helping to make this clinic a success. We would especially like to thank Joy for organising the interpreters that were needed for some families who attended the clinic.

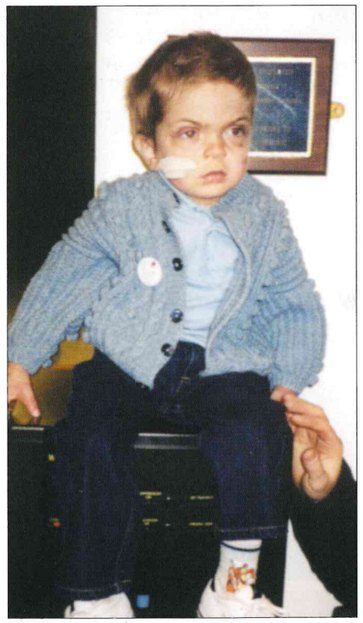


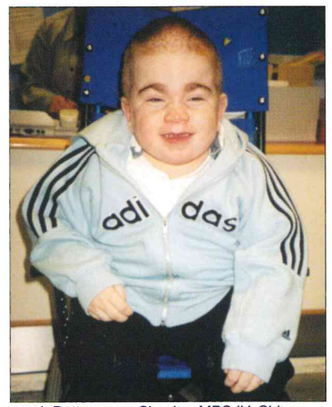




Clockwise from top left: Shabana - MPS IV, Joy, Jebram - MPS IV, Ansan - MPS III, Fahim - MPS III, Faye - MPS III

Birmingham Clinic





Top row from left: Parvash - MPS III, Sumaira - MPS IV. Middle row: Nathan - MPS IH BMT, Dr Ed Wraith & Dr Chakrapani. Bottom row: Shazia - MPS IV, Shinyar - MPS III, Jack - MPS II.

Newcastle Clinic - 4th February

Sophie Denham

On a very cold and wintery day, back in February the Newcastle clinic took place at the Royal Victoria Hospital in Newcastle.

The Advocacy Support Team arrived at Newcastle bright and early on Tuesday morning and made our way to the hospital. After meeting with the doctors we were shown to the waiting area where we could meet with the families. The waiting area was very brightly decorated, had lots of toys and was playing the film toy story, which was a huge, hit with many children and adults!

The clinic went very well considering the bad weather conditions and only a few people were unable to make their appointments.

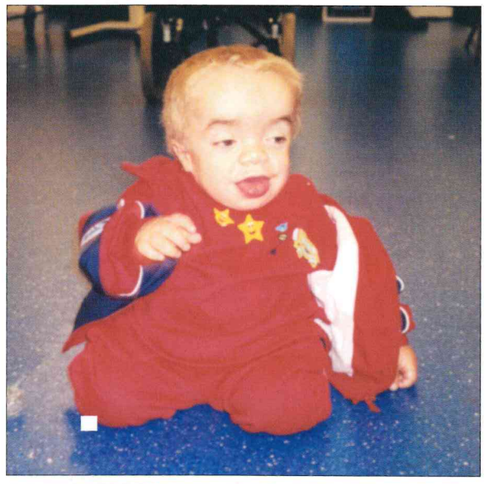
Your feedback on the clinic is very important to us and we very much appreciate all your views about the clinic and its location.

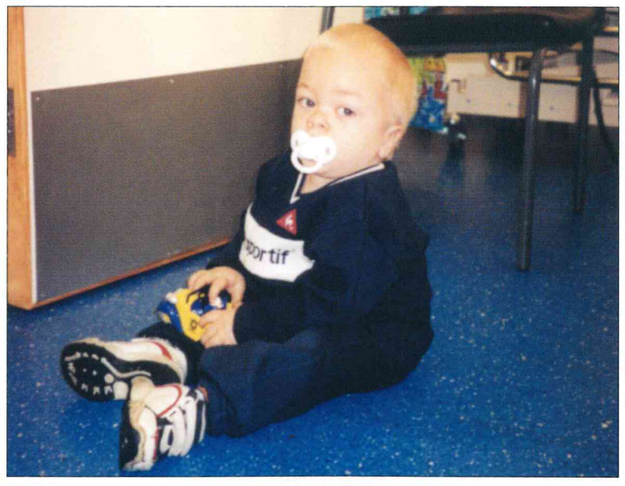
This is the first clinic that Dr Rylance has run and we look forward to liasing and working with him in the future.

Our thanks once again go to Dr Rylance, Dr Leech, Dr Wraith and all the staff at the Royal Victoria hospital who made this clinic possible.

MPS I ERT Trial for Under 5's

The MPS I Under 5's Clinical Trial started in December 2002 at the Royal Manchester Children's Hospital. Five British children are participating as part of a worldwide study. All the children on this trial are receiving the enzyme Aldurazyme as weekly infusions.





Clockwise from top left: Miles - MPS IH, Reece - MPS IH, Lisa Marie - MPS 1H\S, Sophie - MPS IH

ERT MPS I Extension Study

Following two years of commuting weekly to Manchester nearly all of the eleven clinical trial patients are now receiving Enzyme Replacement Therapy at regional centres closer to home. Fond farewells have been said to the Willink Genetics Unit and in particular the nursing staff team lead by Jean Mercer.

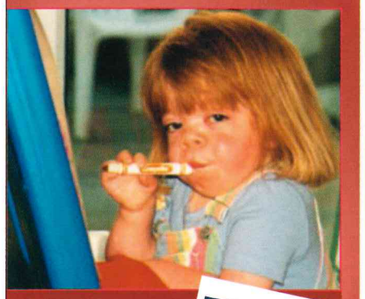
Clockwise from top left: Samantha - MPS IHS, Jenifer - MPS IHS

MPS Publications

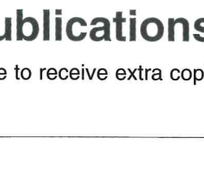
If you would like to receive extra copies of the MPS I Newsheet free of charge, please contact the MPS Office.

MPS1 News

Jessica's Story



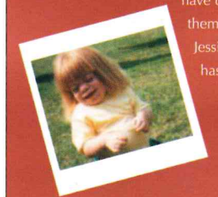
Jessica



Jessica has Hurler Disease. For nine years the family have lived with the knowledge that their ten-year-old daughter is dying.

Over the years Jessica has slowly lost her sight and her hearing and can only now utter the word "Daddy". On some days she is in so much pain Jessica has to be given morphine and will sleep on a bean bag in the living room. Her head is very heavy and Jessica's family believe she has lots of headaches. Sadly there is no way Jessica can show where she is hurting.

This is a very different Jessica from the bubbly enigmatic toddler who charmed everybody singing nursery rhymes and dancing to Robbie Williams. Sadly, as the Mucopolysaccharides have deposited themselves throughout Jessica's body her life has ebbed away.



A description of MPS 1:

"MPS I is a life-threatening genetic disease caused by a deficiency of the enzyme α-L-iduronidase. This deficiency leads to the accumulation of complex carbohydrates in the lysosomes of cells, bringing about the progressive dysfunction of cellular, tissue and organ systems. Resulting symptoms can include impaired cardiac and pulmonary function, delayed physical development, skeletal and joint deformities, reduced endurance, and in some cases, delayed mental function. A majority of patients die before adulthood from complications of the disease."

Mucopolysaccharidosis type I (MPS I) is caused by an inherited deficiency of the lysosomal enzyme α-L-iduronidase. The disorder is recessive, affects males and females equally, and results in an inability to degrade certain glycosaminoglycans (GAGs) - namely dermatan sulphate and heparan sulphate. These glycosaminoglycans, which are important constituents of the extracellular matrix, joint fluid and connective tissue throughout the body, progressively accumulate in the lysosome, ultimately causing cell, tissue and organ dysfunction by largely unknown pathophysiological mechanisms.

The abnormal storage of GAG leads to a very wide range of clinical disease that historically has been divided into three clinical syndromes. Hurler syndrome is used to describe patients at the severe end of a clinical spectrum, whilst patients at the opposite end of the disease spectrum have Scheie disease. The intermediate phenotype is usually referred to as Hurler-Scheie syndrome. This classification, however, is recognised as being over-simplistic and does not accurately reflect the variability seen within this enzyme deficiency.

Symptoms in Hurler Syndrome

Clinically, the disorder is progressive and associated with symptoms and signs in most organs. Patients with Hurler syndrome are normal at birth and usually present in the first year of life with obstruction of the upper airways, often with a persistent nasal discharge (rhinorrhoea). There may be a past medical history of groin (inguinal) and umbilical herniae and many parents notice a prominence of the lower lumbar spine (gibbus). Developmental progress is usually normal at this stage but over the next few months the patient's physical appearance changes. Underdevelopment of the mid-face leads to a characteristic facial appearance often labelled as "coarse". The head is often large and "boat-shaped" (scaphocephalic), the nasal bridge is flat and the lips and tongue are large. The breathing becomes more obstructed and noisy and the patients usually have middle ear effusions secondary to eustachian tube dysfunction. Abdominal examination will usually reveal enlargement of the liver and spleen. The diagnosis is usually made between the ages of six to eighteen months.

Cardiac muscle involvement (cardiomyopathy) may be a prominent early feature but in most patients, combined muscle and valve disease are detected initially on echocardiography and do not produce symptoms until later in the course of the illness. By the age of two years the corneas are usually visibly cloudy and the child's developmental progress slows. Skeletal involvement, known as dysostosis multiplex, leads to short stature, large joint stiffness and claw hand deformity.

As the disease progresses with age other complications may become apparent. Some patients develop glaucoma whilst others (approximately 60%) will develop communicating hydrocephalus requiring shunting. By the age of three years mean DQ (developmental quotient, the childhood equivalent of IQ) is usually <70.

With age, cardio-respiratory involvement becomes more life threatening. Severe cardiac ischaemia or cardiac arrhythmias, secondary to coronary artery disease, commonly occur in the later stages of the disease. Respiratory disease at this stage may include severe obstructive sleep apnoea and a combined obstructive and restrictive respiratory failure develops. Death usually occurs in the first decade of life.

Dr. J.E Wraith, Director, *Willink Biochemical Genetics Unit, Royal Manchester Children's Hospital.*

MPS Publications

A guide to understanding Mucopolysaccharidosis I

Hurler, Hurler Scheie and Scheie Disease

A Guide to Understanding MPS I

This booklet is designed for adults affective by MPS I, parents of children with MPS I and professionals. It gives a detailed Overview of the way classic Hurler and the attenuated forms manifest themselves, and options for clinical management and treatment.

Available from the MPS Office price £2.00.

Funded with an educational grant from Genzyme.

The First International Fabry Meeting

This meeting that took place in Barcelona 22-24 NOvember 2002 was organised by the German Fabry patient association. Fundind for 15 sufferers of Fabry disease was made available to the MPS Society.

This meeting offered a unique opportunity for Fabry patients from Europe and the USA to meet together to share experiences and learn of up to date advances in the management and treatment of Fabry disease.

Extra copies of the published proceedings are available free of charge from the MPS Office.

Barcelona

Highlights from Barcelona, 22–24 November 2002

First International Fabry Patient Meeting

Enzyme replacement therapy in Fabry disease

Fabry disease has been treated using a range of drugs for the many different symptoms of the condition, such as pain and intestinal problems. Management has also involved dietary and lifestyle changes. As the condition progresses, renal dialysis and kidney transplantation are often necessary. Now we have a new treatment – enzyme replacement therapy (ERT) – that is aimed at replacing the missing enzyme (α-galactosidase A) and correcting the underlying cause of the disease. Chaired by Dr Miguel-Angel Barba Romero (Albacete, Spain) and Dr José Ballarin (Barcelona, Spain), one of the main sessions of the Barcelona meeting covered some of the important issues relating to this new treatment.

Editorial

As President of Morbus Fabry Selbsthilfegruppe e.V., the support group for patients with Fabry disease in Germany, and myself a Fabry disease patient, I am very proud to have been a participant and organizer of the First International Fabry Patient Meeting held recently in Barcelona. This newsletter reports the highlights of what, for patients with Fabry disease, was a valuable experience and a truly historic event.

The significance of the Barcelona meeting is that we managed to gather together the largest group of patients ever to meet in one place – over 170 men, women and children with Fabry disease from 18 countries. We heard distinguished clinicians and scientists from around the world give clear and informative presentations that helped us to better understand our disorder and the treatment options available. Just as importantly, we got to know one another and were able to exchange our individual views and experiences.

I sincerely believe that meetings such as this, involving both healthcare providers and patients, will help to ensure that, in future, anybody with Fabry disease will receive optimal and timely treatment. For me, it took 44 years to be correctly diagnosed. During this time, I was treated with many different drugs for numerous other disorders that doctors told me I had. For example, for over 20 years I took cortisone, which I didn't need, prescribed for a non-specific rheumatological disorder that I didn't have. The introduction of enzyme replacement therapy and an increased awareness of Fabry disease within the medical profession should reduce such misdiagnosis and delayed treatment.

I hope that, like me, you left Barcelona with a greater understanding of Fabry disease, with renewed hope for the future, and with the feeling that you are not alone.

Dietmar Basalla

Dietmar Basalla

Dr Miguel-A. Barba Romero (left) and Dr José Ballarin (right)

Measuring treatment effects

Short-term beneficial effects of ERT have been published in the medical press, said Dr Kay MacDermot (Oxford, UK). Patients have reported a reduction in pain and have been able to decrease their pain medication. Kidney function is also stabilized, and beneficial effects have been recorded in the heart. In addition, blood flow in the brain appears to be normalized.

Although these positive effects of ERT are encouraging, we need to demonstrate that treatment works in the long term. This will be done by looking objectively at the effects of ERT on such things as life-expectancy, long-term kidney and heart function and the number of premature strokes. Importantly, we also need to know whether ERT improves the quality of patients' lives. Only in this way will we really convince governments and healthcare providers about the need for ERT, suggested Dr MacDermot. This is where FOS – the Fabry Outcome Survey – is so important. Established by European physicians, in collaboration with TKT Europe-5S, FOS will be absolutely invaluable for determining the long-term effects of treatment in a large number of patients.

Delegates at the Barcelona meeting – the largest ever meeting for patients with Fabry disease.

Americans still wait for treatment

The US Food and Drug Administration (FDA) has not yet licensed either Replagal™ or Fabrazyme™ for use in the USA. In view of this, Jack Johnson of the US patient support group distributed a petition for delegates to sign during the meeting. This will be sent to the FDA demanding that ERT be made available to US patients.

Two products available for ERT

Dr Ravi Ishwar Thadhani

"The good news is that there are two ERT products licensed in Europe", said Dr Ravi Thadhani (Boston, USA), "Replagal™, made by TKT, and Fabrazyme™, made by Genzyme." The two products have identical core protein structures. However, they differ in the sugar and phosphate molecules that are attached to the core structure, which are thought to be important for enzyme activity. The molecular differences are due to the fact that Replagal™ is made in human cells, whereas Fabrazyme™ is made in Chinese hamster ovary cells. There are also differences in dosing and infusion times, and reported clinical effects and side-effects, explained Dr Thadhani.

Because of these differences, Dr Thadhani encouraged delegates to ask their physicians to explain the advantages and disadvantages of each therapy. It is only by knowing the differences between the two products that patients can make an informed choice about which one they should use, he concluded.

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Childhood Wood News

Come and join us for a day of reflection and celebration in the Childhood Wood on Saturday 13 July 2003.

It is 10 years since the Childhood Wood was created to remember those who have lost their lives to MPS or a related disease

THE SOCIETY FOR MUCOPOLYSACCHARIDE DISEASES

ANNUAL PLANTING OF THE CHILDHOOD WOOD

FRIDAY 25TH OCTOBER 2002

CHILDREN AND ADULTS REMEMBERED

Thomas Thacker	Shivahram Selvaranjan
Steven Grandidge	Jessica Stuart
Jade Robinson	Rozeena Ismail
Daniel Hughes	Eleanor Gee
Jake Corcoran	Kim Eggleton
Aaron Martin (Jennings)	Charles Green
Michael Armstrong	Sarah Lowry

Rare Disorders in Europe

Christine Lavery - Director

EPPOSI - the European Platform for Patients Associations, Science and Industry was established in 1998 as an interface between the different specialities with the aim to debate issues of common interest and influence policy decisions. Each themed workshop aspires to bring together the academics working on research with the project managers, pharmaceutical companies and venture capitalists who might between them see potential and a way to develop and fund the development of new therapies to everyone's advantage.

In October 2002 EPPOSI held its third workshop in Rome to examine rare diseases therapy development. It brought together over 100 delegates to discuss how the Orphan Drug Regulation has been of benefit to date; the process and to emphasise the need for accessibility to funders and companies; the regulatory process involved in bringing the drug to the patient. I felt enormously privileged to be invited to speak on the development of the MPS disease registry and how it has contributed to the development of new and future therapies. The occasion was all the more poignant as the meeting was held in the Italian Senate. You will see from the photo below that as we queued for a considerable time whilst our access and securities issues were overcome, one of the speakers decided to give his talk in the cobbled street outside the Senate building.

Hope For Alice

Elaine Kornbau

While in Ukraine with the Peace Corps, I tutored an amazing little girl named Alice. Born with a degenerative disease called Morquio Syndrome Type A, Alice stands about 3 feet tall, wears thick glasses and has extreme difficulty walking, writing or standing for any length of time. Because there are no laws dictating that public buildings be handicapped-accessible, Alice is confined to the tiny 3-room apartment she shares with her parents and is home schooled. In January of 2002, I was asked to tag along with my friends Lena and Bill to one of Alice's lessons. Upon meeting Alice, I was struck by her intelligence, grace, wit and exuberance for life- the kind one rarely sees outside the realm of terminally ill children. Her joy at receiving a new visitor was almost overwhelming, and knew I'd be making many more trips to see her during my time in Chernigov. As our lessons increased, so did our fondness for each other, and Bill, Lena and I began researching Alice's illness and possible treatments.

Doctors have recommended that Alice have a spinal fusion surgery to stabilize her vertebrae and protect her spinal cord. Such procedures are not available in Ukraine, so Alice will have to come to America for the surgery. My friend Bill recently contacted me with good news; he and his wife, Lyuda, have secured a commitment from the Scottish Rite Hospital in Dallas, Texas to donate the surgery and have also arranged for Alice to stay at the Ronald McDonald House for the 4 months she's expected to be in the states.

We've cleared a huge hurdle in arranging for the hospital to donate Alice's surgery, but now we're left with the task of raising $5,000 to pay for the rest of her expenses. Alice and her mother will need money for visa fees, passport application fees, plane tickets, meals, and the ten-dollar per day donation that the Ronald McDonald House asks for. Bill has set up a special account for donations to Alice's fund, and we welcome any donation you are capable of making. Any money we receive will go directly to Alice. The surgery she's slated for will actually be the first of many, so if we exceed our goal of $5,000, we will hold the money to fund future treatments for Alice.

As returned Peace Corps volunteers, we feel it is our duty to continue to help our former community of Chernigov, and through our personal contact with Alice, we would like to see the life of this special little girl transformed by the kindness of others. Please share this information with anyone you know who might also be interested in donating.

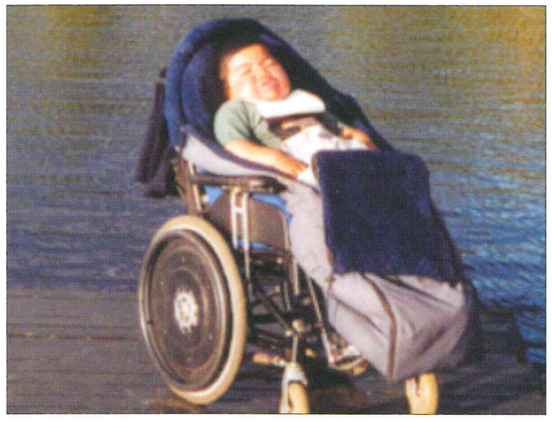
Thank you a million times over!

Editors note:
If anyone in the UK feel they want to make a donation to directly help Elaine and her friends raise these funds, please send a cheque made out to MPS (Alice). We will then pass these donations on. At the time of going to print Elaine had reached the halfway mark and has only $2,500 to go. Well done Elaine.

Einar's sprung wheelchair

Oddrun Grønvik - Oslo

Our son Einar (MPS II), now 16, has had his own wheelchair since 1994 and walked his last steps in March 1996. The first chair was a proper high wheelchair with big hind wheels, not to heavy and easy to manouver. You could set the angle between back and seat, and you could tilt the whole seat backwards - not bad at all! But as time went on, Einar sat less well in it. He was not able to keep his body stable when we wheeled him around, and would slump sideways or even start sliding out of the chair (despite his belt). Sometimes he did it on purpose, to show us that he was fed up with sitting in it, but more often it was a sign of tiredness.



Bumping hurts

Einar's sister Helga used to try all his tools and copy all his movements, "to find out what it feels like", and it was she who told us that it hurt to drive over cobbles and on stony tracks, up and down steps and over pavements. Indoors, carpet edges, low thresholds and wires on the floor would give an unpleasant bump. Even when we - my husband and I - used all our strength to lift the wheels over hindrances and bring the chair down gently, there were too many jolts for a strong and fit girl. And she could soften the bump by holding on to the armrests and lifting her bottom off the chair seat, or bend and stretch to loosen up her back. Einar couldn't do any of those things. If it hurt for her, it must be a lot more painful for him.

In addition, handling the unsprung chair was heavy. Even quite small stairs into shops became major hindrances when I pulled Einar and the wheelchair backwards in and up, trying to do it without bumps and jolts for him, and taking the strain on my arms and shoulders. And doing my best, I would still be giving him a very rough ride.

Why not springs?

Looking round, I saw that all longterm lying and seating tools for human beings rested on springs - from mattresses and baby buggies to every variety of car seat. Try offering a taxi driver an unsprung car seat for him to sit in all day, and see what you get! We take sprung seating completely for granted, and wouldn't dream of accepting anything else. An unsprung bus seat makes you think of backpacking in Nepal or Zambia - the thought is enough to make one stiff!

Yet this is the condition most often offered to the physically helpless, and especially to those who cannot manage their own wheelchairs, and have to be wheeled by others. After all, most of the really helpless live in institutions (huge floors, no thresholds) and do not get out much, so their carers won't notice how heavy and difficult a stiff wheelchair is to handle in the streets. The users themselves are often unable to complain, or their complaints are too indistinct to be understood.
But we didn't know that then. We just saw that if we were to take Einar round with us so that he could be part of our family life, he needed a better chair - both a form-fitted shell to sit in, and springs to make transport safe and comfortable for him and easier for his helpers.

The form-fitted shell was not a problem. Experts arrived and did a 3D scan of Einars body, supported in a position recommended by his school physiotherapist. A plaster cast from the scan was made and adjusted through a couple of fittings. Finally two detachable shells were provided, one a near body fit and one slightly more spacious for winter and outdoor use (room for clothes and driving bag).

No proven need

The springs were a major problem. The local ergotherapist had never heard of sprung wheelchairs, but very helpfully tackled the Norwegian social services equipment registry. No go - they had never approved a prototype for a sprung wheelchair. The approval procedure would be expensive and take time. No manufacturer had ever approached them with a project idea even. When asked whether they would support such a project, they told us that the need was not proved. There was simply no demand.

Our ergotherapist now realised that she had got onto untrodden ground, and got keen. She approached the Norwegian manufacturers' association, and individual manufacturers of wheelchairs in Norway and Sweden. They too raised their eyebrows and explained to her and us (in words of one syllable) that there was no such thing as a sprung wheelchair, because no-one needed it.

This reply really got to us, and I made up my mind that Einar was to have his sprung wheelchair in record time even if we had to fund the development of one ourselves. Einar's father trained as an engineer once upon a time and he too got annoyed by the arrogance we were met with. He started sketching possible constructions and I cast around for strategies of war.

Try television

One of Einar's uncles suggested that we should try to interest a television consumer programme that is sent immediately after the evening news, and therefore gets seen by a lot of Norwegians. I emailed them, they liked the idea, and one cold and slushy November day in 1998, we received journalists and television crew first in our home. Then we braved the elements and went down to the centre of town - cobbled streets, tram line tracks and all - and I took Einar through our "Saturday round" while they filmed.

The result was terrific. Until I saw the film, I had had no idea how I struggled to bring Einar with me, and I had never actually seen his face when he bumped down a step, bracing himself to cope with the pain, struggling to keep himself up. No-one could doubt that Einar was uncomfortable in that chair - nor fail to see that all I did, was ordinary, everyday stuff. We were not asking for luxury, just the possibility to cope.

The television people then went on to interview all the people we had spoken to, and I am happy to say that neither the wheelchair industry nor the bureaucrats backed down. They both insisted that the need for sprung wheelchairs was non-existent. The whole thing was cross-cut and put together to excellent effect. and seen by a lot of people two weeks before Christmas 1998.

Better service

The next day the chairman of the manufacturer's association phoned our ergotherapist. "We didn't show up as very service-minded yesterday. I want to put that right." He had found a manufacturer who was willing to develop a prototype, and the association would bear the cost in case the prototype was not approved. Two days later a team of engineers arrived at our house and spent the day crawling about on the ground, studying the motion of the old wheelchair with and without Einar in it on all sorts of surface, making sketches, taking photographs and discussing ways and means with Einar's father. There were several tests to be carried out, and a lot of paperwork for the manufacturers and for us, but four months later the first of two new wheelchairs was in place, and a new life of comfort and mobility started for Einar.

Tourist in Paris

The chair is, incidentally, made so that it is easy to take apart, and it fits into the back of for instance a five door Renault Espace.

How do we know? In June 2002, Einar was one of many MPS children attending the international symposium in Paris. Parts of the children's programme was not open to him for insurance reasons - fair enough. But Einar and his father could book a taxi through the hotel, drive into central Paris, and be tourists on their own - thanks to the wheelchair! That was how Einar made it up the Eiffel tower, saw Paris from the Seine, and got around on the Left Bank and Ile de la Cité. Cobbles and gutters have to be crossed with care, but they don't block progress. We even braved the Metro to see Paris by night (and met a lot of enormously helpful Parisians on the way).

At the moment, Einar is waiting for new wheelchairs - sprung, of course. The old ones have been through almost four years of hard wear. We have been informed that three different prototypes now have been developed for us to chose between. And this time, we don't have to prove the need!

First MPS Clinic At Frambu in Norway

Oddrun Grønvik - Oslo

10.-14.February this year, the MPS family group in Norway had a highly successful gathering at the Frambu foundation, outside Oslo, with Dr. Ed Wraith and Christine Lavery as guests and chief contributors. A bit further down, you'll get some glimpses from this ver content-packed week - but first of all I want to say a little about the Norwegian MPS family group, our relationship with the Society for Mucopolysaccharide Diseases, and about Frambu, the venue of our gathering.

The Norwegian MPS family group is small. It has roughly 35 families on its address list, including those whose children have passed away. Not so surprising perhaps - the whole of Norway's population equals that of greater Manchester, roughly 4.5 million. But distances are bigger - families from Northern Norway have a three hour flight to get down to the South.

We do not have families with all the MPS diagnoses. MPS I (Hurler) is the most common form, after that MPS II (Hunter). We have very few cases of MPS III (San Filippo), and even fewer with MPS IV (Morquio) or MPS VI (Marotaux-Lamy).

The English MPS Society has been a tower of strength for our families for more than twelve years, and many families have brought their children to see Dr Ed Wraith over the years. Ed Wraith and Christine Lavery have visited Norway to give seminars for Norwegian parents and professionals several times before. These arrangements have always been very successful, both on the day and in the sense that they have led to general improvement in the support of Norwegian MPS families.

The Frambu foundation, south of Oslo, also has to be introduced, as it is a type of organisation that has few parallells. It started almost sixty years ago as a camp school for disadvantaged children, moved on to health camps and habilitation training for first polio, then cerebral paresis, and has over the last fifteen years

developed into one of three national centres with tertiary level responsibility for a group of (at present) 35 rare diagnosis groups, including all progressive neurological disorders in children. The MPS disorders are included under this umbrella. In addition Frambu looks after undiagnosed cases involving mental retardation. The approach is multidisciplinary, and includes medicine, psycho-social issues, special needs education and all sorts of practical advice on equipment, rules and regulations etc. Lately, Frambu has started a project, Pro-Unik, to improve its care for the most severe diagnosis groups, and again, the MPS disorders is one of those, so our principal partner at Fambu is the Pro-Unik team.

Frambu has a complex of modern buildings situated in the forest land twenty minutes drive south of Oslo, with a couple of lakes and a good deal of wild life around it.. In the summer, they still run summer camps for handicapped children. The rest of the year is devided between information courses for diagnosis groups, specialised topic seminars, outreach efforts and producing popular information about the various illnesses. All of this is dealt with in close cooperation with the various user groups. Frambu also tries to keep abreast with new developments in and outside the country on behalf of the diagnosis groups, from the Ministry of Social affairs, EU, etc.

Since The MPS family group is too small to run a proper full scale organisation, we count ourselves lucky to be included among the diagnosis groups which have the Frambu foundation as their home from home.

The February MPS gathering was organised as an information stay for families, lasting from Monday until Friday. The programme was as follows:
Monday - Tuesday In the course of these two days each family was offered three individual consultations, i.e.

- MPS clinic with Dr Wraith,
- consultation with Christine Lavery,
- introductory interview with Frambu staff

In addition, the children were introduced to the Frambu observation school, and settled into their weekly programme.

Wednesday: One day seminar for parents and professionals (education, primary health care, care home staff etc), with Ed Wraith and Christine Lavery as lecturers. Lectures were translated into Norwegian.

Thursday: two parallell sessions.
1. The families were offered seminars by Frambu staff on two topics: talking to children (the affected and siblings) about illness, and an update on rules and regulations, benefits etc.
2. One day seminar at Rikshospitalet, our largest tertiary hospital and the only one that does BMT. Ed and Christine were again chief contributors. This seminar was specifically designed to attract medical specialists, who are normally hard to get hold of if the topic is multidisciplinary. This time there was no translation. At the end of the day, Ed and Christine went home (we hope not in a state of total exhaustion). Friday was parent contribution day. We had asked some of the families to prepare short talks on given topics from their own (recent) experience, and to focus on what they themselves had done, and how they (looking back) thought and felt about it. There was also a final summing up, individually and jointly. Parents were offered consultations with the teaching staff in the observation school, and an individual meeting with Frambu staff to go through the list of what had been arranged for each family in terms of future follow-up. Since the families stay at Frambu for the whole week, there is also a social programme in the evenings. In February, this had to be mostly indoors.

How did it go?

The turn-out was the best we have had. It isn't easy for families to take a whole week off work, school etc, but the programme was obviously attractive, and so this time Frambu was fully booked, and we had several families who have never come before. Sadly, ther eweresom last minute cancellations due to illness. The Wednesday seminar likewise had a full house, with more than 100 people attending from all over Norway - Frambu has had a couple of MPS seminars, but never had this attendance before.

The Thursday seminar at Rikshospitalet was a new venture, and again successful beyond expectation - the specialists came, stayed, and took part.

What made it successful?

Well, first things first:
Content: The medical research update, and especially the possibility of Enzyme Replacement Therapy, is very exciting. Several families and many professionals came to hear about that, and came out reassured that this treatment is going to come and will be on offer in Norway to those who can benefit from it. Dr. Arvid Heiberg, head of the Genetic disorders in children unit at Rikshospitalet, has already set the Norwegian apparatus in motion to have ERT treatment funded by our public health service. ERT is already being used for other diagnoses in Norway, so the principal hurdle is crossed.

Most of the Norwegian MPS I children have had a BMT, and they and their families have shared the experience that BMT doesn't take the condition away, but it does change it. Our oldest member with a BMT is now 21 years old - the youngest one was unable to come as he is still in hospital, recovering from it. So many families wanted to know about follow-up treatment, complications etc, and to get a state of the art summary - which they got.

The possibility to talk to Christine about anything that mattered, also brought up new issues. In particular, the near adults with MPS mentioned that of clothes for young people who don't fit into standard sizes, and friends outside the family network. We hope to be able to do something there. Some families chose to bring along an interpreter, but most went on their own.

Many Norwegian families have wanted to come to meetings and conferences organised by the MPS Society in England, but feel shy about their English, anxious about practical difficulties of travelling with an MPS child etc. Some families overcame that shyness during our MPS week and determined to travel, others are thinking about it. But contact with the UK MPS society will definitely become more important for our members after this gathering.

Organisation: There were some features to this MPS gathering that were new to both the Norwegian MPS group and to Frambu, the most important one being that Frambu had never offered a clinic with a foreign medical specialist before. This opportunity for the families to meet Ed and bring their own doctor with them, was new, and a great success. One of the Norwegian pediatricians, who was responsible for two families who couldn't come, also booked himself in for his absent families!

When my son Einar first was diagnosed, 13 years ago, communicating with the doctors was a real challenge, and to get them to talk to each other across specialities and nationalities very hard indeed. It seemed as if the most important thing in the world was never to betray ignorance, therefore they never asked questions. I don't claim that this attitude has vanished entirely, but at least within child habilitation the mood is different, and the opportunity to learn from a more experienced colleague very much valued. So we had lots of medical staff at Frambu that week, and they were not afraid to raise issues that had puzzled them, or to ask questions, and grateful for the very informative answers they got.

The seminar at Rikshospitalet was a new venture as well, and we did not expect miracles. It is notoriously hard to get decent attendance at multidisciplinary seminars in an institution which has specialisation as its guiding light. But the attendance was good and active, and it gave us a platform to work from on a very important issue, which is registration. A chief aim for the Norwegian MPS family group is to get a central register of all MPS families. At present we have several different labs doing diagnostic tests and prenatal tests, diagnostic procedure is carried out at regional hospitals, so the medical information is uncoordinated. The family group has an address list, but not everyone comes to us. Frambu is not allowed to keep address lists, or offer information stays to families whose children have died. If the laboratories could coordinate their information, we would in fact have the registry we need.

The point of the exercise: The aim of this MPS gathering was, as usual, fourfold:

Improved information to parents and professionals
network building between families and professionals

Feedback to specialists, the MPS family group and to Frambu, resulting in general competence build-up

Isolate issues that need central follow up

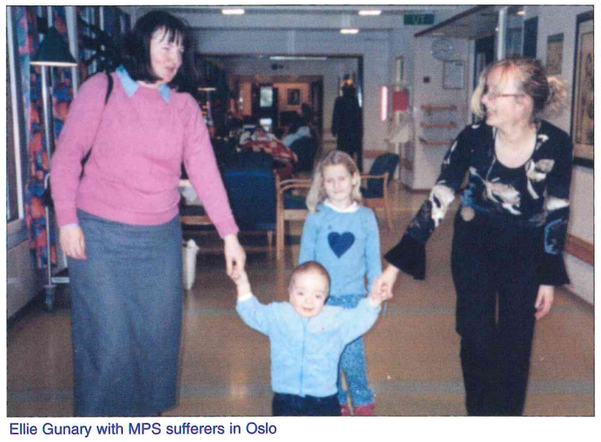
We, the organisers, think that those four aims were reached. In particular, Frambu gained a lot of valuable knowledge and experience which probably can be applied to other diagnosis groups, making it possible for them to sharpen their profile as professional facilitators in a difficult and very composite field.

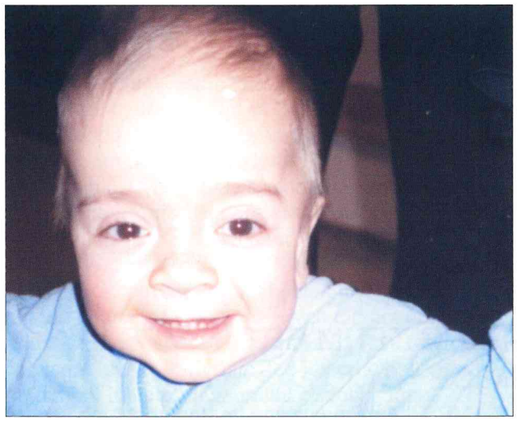
Whom to thank

Ed and Christine were the star atractions, not only because of what they know and pass on, but also because of who they are. What has been mentioned in particular by many who met them here, is their professional and personal generosity, which makes contact easy across language barriers. They both have a talent for making the difficult sound simple, without underestimating their audiences. We think that this professionalism and generosity set the mood for the whole week, and was essential in achieving our very successful MPS gathering. Thank you!

Oddrun Grønvik
Contact person
Norwegian MPS family group

PS. If you want to know more about Frambu, try http://www.frambu.no/main.asp





Ellie Gunary with MPS sufferers in Oslo

INTERNATIONAL EVENTS

MPS I

The committee for propriety medicinal products at the EMEA gave a positive opinion of AldurazymeTM in Feb 2003. The normal procedure is that following a positive opinion it takes a period 3-4 months before full approval is granted. We are optimistic that we are able to give you the very exciting news about Aldurazyme in the next newsletter

MPS II

It is hoped that the phase III enzyme replacement clinical trial for MPS II will start in May 2003. If anybody would like to know more about this clinical trial please contact Dr wraith or the MPS Society.

Sanfilippo - Research Update

Presented by Briony Gliddon at the 2002 Canberra Conference. Briony is a Sam Lister Ph.D scholar at the Dept Of Paediatrics, University of Adelaide Lysosomal Diseases Research Unit, Chemical Pathology, Women's and Children's Hospital.

Mucpolysaccharidosis Type III (MPS III, Sanfilippo Syndrome)

- Lysosomal Storage Disorder (LSD)
- Absence in a lysosomal enzyme needed to break down heparan sulphate
- 4 subtypes ofMPS III (A,B,C and D)
- inherited disorder, characterised by severe CNS degeneration
- frequent and severe temper tantrums
- hyperactivity
- aggresslon
- severe mental retardation

Therapy for MPS III Patients

- no specific therapy
- Bone Marrow Transplantation

Therapies under investigation

- Enzyme Replacement Therapy (ERT)
- Gene Therapy
- Stem Cell Therapy
- Substrate deprivation

MPS III Animal model

- MPS III
  A naturally occurring mouse
- 2 canine models
  -New Zealand Huntaway dog
  -Wire-haired Daschund
- MPS III B
  -emu
  -knock out mouse
  -cow
- MPS III
  -goat

AIM

To evaluate whether enzyme that can enter the brain is effective in preventing/reversing the pathology observed.

LONG TERM ERT IN MPS III A MICE

- MPS IIIA mice enzyme treated from birth
- MPS IIIA mice enzyme treated from 6 weeks
- MPS IIIA mice untreated
- Normal mice enzyme treated from birth
- Normal mice untreated

*injected 1 mg/kg of sulphamidase enzyme weekly into the mice
*Ist injection in the superficial temporal vein
*remaining injections in the tail vein
*duration of study -20 weeks

ERT RESULTS

Behavioural Improvements

- From weaning (3 weeks of age) 5-6 mice were caged together
- Untreated Male MPS IIIA mice and those treated from 6 weeks of age had to be separated 10 weeks into the trial.
- Male MPS IIIA mice treated from birth had to be separated 18 weeks into the trial
- Normal male mice did not have to be separated during the 20 week study

SUMMARY

- Long term ERT was very unsuccessful in the MPS IIIA mice, only if initiated early in life
- Enzyme that can enter the brain is effective in reversing pathology. Storage will return if enzyme is not regularly supplied to the CNS

FUTURE WORK

- Optimise enzyme dose rate in MPS IIIA mice to get maximum efficacy
- Investigate ways of overcoming the Blood Brain Barrier (BBB)
- Investigate ERT in a larger animal model -the MPS IIIA dog

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Enzyme Replacement Tharapy for MPS IV-A

Biomarin Pharmaceutical Inc. has an active programme aimed at developing enzyme replacement therapy for MPSIV-A. At this point there is no timeline for a human clinical trial. Studies in MPSVI and VII animal models suggest that, if given early, the enzyme can potentially change the outcome of bone and cartilage disease.

Work to produce adequate quantities of enzyme for clinical use is still in progress; at this time, there is no clinical trial planned.

The BioMarin Website is www.biomarinpharm.com

BioMarin Presents Positive Phase II Data On Aryplase For MPS VI

Aryplase(TM) Well Tolerated and Associated with Functional Improvements BioMarin to Initiate a Pivotal Phase III Trial in 2003

NOVATO, Calif., Mar 14, 2003 /PRNewswire-FirstCall via COMTEX/ -- BioMarin Pharmaceutical Inc. (Nasdaq and Swiss SWX New Market: BMRN) announced positive results from its Phase II open-label study of Aryplase, recombinant human arylsulfatase B (rhASB), an investigational enzyme replacement therapy for the treatment of mucopolysaccharidosis VI (MPS VI). MPS VI (also known as Maroteaux-Lamy Syndrome) is a debilitating, life-threatening genetic disease, for which no drug therapies are currently available, that is caused by a deficiency of the enzyme arylsulfatase B.

Dr. Paul Harmatz, M.D. of Children's Hospital Research Center of Oakland, California will present the results today at the American College of Medical Genetics 9the Annual Clinical Genetics Meeting in San Diego, California. The open-label, 10 patient, clinical study of Aryplase was conducted at two sites, one in the United States, and one in Australia. The study evaluated clinical and biochemical measures of safety and efficacy in male and female subjects between the ages of 6 and 22, for a duration of 24 weeks. During the study, subjects received weekly 1.0 mg/kg infusions of Aryplase, a dose selected based on results from both a Phase I study, and the feline MPS VI model.

Study Results

Results from this Phase II study indicate that Aryplase is well tolerated and is associated with improvements in several clinical endpoints. On average, subjects experienced improvements in endurance as measured by the distance walked at the 6 and 12 minute time points of a 12-minute walk test, and the number of stairs climbed in 3 minutes. Functional improvements were also observed in joint pain and stiffness, and in shoulder flexion, extension, and rotation in subjects who exhibited less than 90 degrees shoulder flexion at baseline. Results are provided in more detail below:

The average improvement at the 6-minute time point in the walk test was 64 meters over baseline distances, which ranged from 19 to 247 meters. On average subjects demonstrated a 62% improvement in distance walked at 6 minutes.
The average improvement at 12 minutes in the walk test was 155 meters over baseline distances, which ranged from 33 to 475 meters. On average subjects demonstrated a 98% improvement in distance walked after 12 minutes.
The average improvement in the number of stairs climbed in 3 minutes was 48 stairs over the baseline number, which ranged from 20 to 92 stairs. This increase in the number of stairs climbed represents an average improvement of 110% over baseline.
Subjects also experienced improvement in pain and joint stiffness based on a modified child health assessment questionnaire (CHAQ) in which subjects were asked to score pain levels on a scale from 0 to 100. Joint pain and stiffness scores were reduced on average by 57% and 54% compared to baseline, respectively.
Small improvements in both passive and active measurements of shoulder range of motion for flexion, extension and lateral rotation were observed, with the largest gain achieved in the three subjects who exhibited less than 90 degrees of active shoulder flexion at baseline.
In addition to the clinical improvements observed, study participants also demonstrated an average decrease in urinary glycosaminoglycan (GAG) excretion of 71% in 24 weeks, indicating Aryplase reduced carbohydrate storage in MPS VI subjects. As part of the Phase II trial, several other exploratory endpoints were evaluated, but on average did not indicate meaningful changes in the 24- week study period. These include pulmonary function as measured by forced vital capacity, a pinch and grip strength test, physical activity, oxygenation level during sleep, expanded time to get up and go test, and a set of tasks reflecting quality of life. Improvement in these clinical endpoints may be observed with continued infusions over a longer period of time.

Results of the study demonstrated that Aryplase was generally well tolerated. Out of 240 infusions over the 24-week period, 5 adverse events were reported during infusion and an additional 8 during the day of infusion, which were most commonly abdominal pain, febrile reactions, and pruritis. Separately, there were 7 serious adverse events, 6 unrelated to drug, and 1 possibly related to the drug. Consistent with infusion of protein drugs, subjects developed antibodies to Aryplase during the course of the study, which did not correlate to reduction in urinary GAG excretion. Mildly reduced complement levels were detected in some patients, which did not have a measurable clinical impact.

A Genetic Condition In The Family

Contact a Family

When you or a member of your family are first told that your child might have a genetic condition you may feel shocked, very upset, or just numb. Perhaps this news confirms what you have suspected for some time. Your doctor may have suggested genetic counselling to you. Many people are not sure what genetic counselling involves, however, or what to expect from a genetics appointment. Furthermore, many people may want to know how a condition arose in the family and whose side of the family it came from. These unanswered questions may cause tensions within a family and generate feelings of anxiety.

What are genes and how do they cause genetic conditions?

If you were to look at your skin under a microscope, then what you would see would be millions of tissue cells. In fact, every part of our body is made up of cells, rather like a house is made up of bricks. In the centre, or nucleus, of most cells in our body are thread-like structures known as chromosomes. Usually, there are 23 pairs of chromosomes (46 in total) in each cell.

Chromosomes carry genes. We can think of genes as 'strung' along chromosomes, in much the same way as beads are strung along a necklace. Genes are the instructions about how to make a new baby from a sperm and an egg -the blueprint from which the body is constructed. Genes contain all the biological information needed for us to grow and develop and remain healthy from the moment of conception to the day we die. Genes give us our physical characteristics including our eye colour, our ability to run fast as well as our susceptibility to disease.

Genes are made up of DNA (Deoxyribonucleic Acid). This is the code or language of the body's instruction manual. If there is a change or spelling mistake in this instruction manual, then the body is not able to function properly because it has not received the correct instructions in order to do so. The features of a genetic condition occur, or may occur, therefore, when there is a spelling mistake or change in a person's genetic material.

The functioning of our bodies requires that many thousands of genes work together. Changes or spelling mistakes in different genes has resulted in many different genetic conditions. Some genetic conditions, such as cystic fibrosis or Huntington's disease, are caused by changes in single genes. Other conditions, such as spina bifida, are caused by changes in a number of different genes. Chromosomal conditions are due to changes in the number or structure of chromosomes, an example of a condition caused by a change in the number of chromosomes is Down syndrome.

Does the condition run in the family?

Although it is often said that a condition 'runs in the family', not all genetic conditions are inherited or passed on in families. Some genetic conditions occur sporadically. This means that usually other members of the family are not, or will not be, affected by this condition.

Many genetic conditions, however, are inherited or passed on in families. There are a number of ways in which genetic conditions may be inherited. In some families, a condition is inherited when both parents pass on a change in a single gene to their child. In other families, a condition is inherited when a change in a single gene is passed on by only one parent. In yet other families, the likelihood of a genetic condition arising in a child depends upon whether the change in a single gene is passed on by the mother or father.

There are many genetic conditions where scientists have not been able to identify the specific gene changes which cause the features of an individual's condition. In these cases, an estimate of the likelihaood that the genetic condition will be passed on or inherited is given to families.

What is genetic counselling?

Genetic counselling involves giving information to individuals and families about genetic conditions and the way these conditions are inherited. Clinical geneticists (doctors) and genetic counsellors (who may have a nursing background) provide information about the likelihood of a genetic condition happening in a family and about the medical management of a condition. Individuals are supported in the choices they face and are helped to make the decisions which are best for them. Genetic counselling helps individuals to deal with the psychosocial issues arising in their situation.

Who is offered genetic counselling?

Your GP or hospital doctor may have suggested genetic counselling to you or alternatively you may have sought genetic counselling for yourself. Some of the reasons for this might include:

1) You or your partner already have a baby or child who has a physical problem or delay in development. The diagnosis may be uncertain and either you or your doctors are wondering if there may be a genetic cause for your child's problems.

2) You and your partner have lost a baby during pregnancy or infancy.

3) You are concerned about a condition which may be genetic and you would like further information.

4)You or your partner have, or carry, a condition which might be passed on to your children.
5)There is a known genetic condition in your family or your partner's family.

6) You and your partner are close blood relatives.

7) There is a strong history of cancer in the family.

What happens at a genetics appointment?

You and your partner, and possibly other members of your family, will be seen by a clinical geneticist or genetic counsellor or both. You will spend time discussing your concerns and will be asked for information about your personal and family medical histories. A family tree will be drawn up which can give valuable information. Sometimes you will be asked for medical details about other people in your family, if this is relevant. However, relatives will never be approached without your permission. If appropriate, your child or other members of the family will be offered a physical examination. Blood tests and other tests may be discussed and arranged.

If your concern is about a child in the family, he or she will usually have a detailed physical examination and the doctor may ask if photographs can be taken for the child's medical record. This helps the staff to recall the child accurately, without having to rely on memory.

When there is a genetic condition within the family this will be explained to you. The clinical geneticist or genetic counsellor will discuss ways of coping with the condition and will give information about the available medical and social support. During this time, you will be encouraged to ask questions. You might have questions about the chance of either inheriting or passing on a genetic condition. You may want to know what tests are available to confirm a diagnosis or if there are tests that can be offered during a pregnancy.

Sometimes it is not possible to make an exact diagnosis, because of the current level of scientific knowledge. However, the doctors may be able to say whether they feel the condition is genetic or not, even if the diagnosis is not certain.

Often people who attend a genetics appointment will leave feeling reassured. Others will face difficult decisions such as whether or not to have a particular test or what to tell their children or other relatives about a genetic condition. Relationships within the family may be brought into sharp focus at this time. Feelings of guilt may arise when a child is diagnosed with a genetic condition or if parents have taken the painful decision to terminate a much wanted pregnancy. Healthy members of a family may feel guilty if they have not inherited a condition. Blame may occur where only one member of a couple carries a genetic risk. In addition, couples aware of an increased genetic risk to their children must decide whether or not this knowledge will affect their plans for a family. Family support can be very important for individuals coping with the impact of a genetic disorder.
If you would like to know more about genetic counselling or about a genetics centre near to you, see the useful contacts section at the end of this factsheet.

Is genetic counselling the right choice?

You may like to consider the following points before committing yourself to a genetics appointment.

1) Why have you been referred for genetic counselling?

2) Could you meet or talk with a professional, such as a genetic counsellor, to discuss any concerns you may have about your appointment? If not, is there someone else you could talk with (Antenatal Results and Choices, Contact a Family)?

3) What are your reasons for attending an appointment? What are your reasons for not attending an appointment?

4) What do you hope to achieve form your appointment?

5) What questions are you going to ask the geneticist or genetic counsellor?

6) How will you deal with the information you are given? Who are you going to tell afterwards?

The Genetic Interest Group (GIG) has suggested some 'helpful hints' prior to attending a genetics appointment in a leaflet entitled *'Has your child a genetic disorder? What do you need to know from whom?'* These include:

1) Take a pen and paper with you to write down the answers.

2) You could take a tape recorder with you if you want to record the meeting.

3) Take a friend; they will remember more and can write down information.

4)Don't be afraid to ask what you really want to know.

It is common practice for the genetic staff to send you a letter after the appointment, summarising the main points of discussion. A copy may be sent to your GP with your consent. You could ask at the start of the appointment if this is to be done, as this may enable you to listen carefully without having to take notes.

Common beliefs about genetic counselling

The following statements reflect some commonly held views about genetic counselling:

At the appointment I can find out if a condition is genetic by having a blood test done to look at my

genes.

People often think that there is a test for every condition. This is not true. Genetic tests are available but for only a certain number of conditions. If a test is available, then the doctor or counsellor will spend time discussing the reason for the test, its accuracy and the implications of the result. The extent of counselling and the issues to be addressed will depend upon a person's situation and the type of test being offered. Individuals are required to give informed consent before a blood test can be done.

The doctor or nurse is going to tell me that I shouldn't have any more children.

Genetic counselling aims to be non-directive and non-judgemental and this means that doctors and counsellors refrain from telling individuals what they should or should not do. By discussing all the options available in a given situation, doctors and counsellors enable individuals to make their own decisions and offer support for the decisions individuals make.

Genetic counselling is the same as psychological counselling.

Often individuals are reluctant to attend a genetics appointment because they do not consider themselves to have a 'psychological' problem. Genetic counselling differs, however, from psychological counselling. Genetic counselling is a process of communication between doctor or counsellor and individual or family in which the medical and psychosocial issues associated with the occurrence or risk of occurrence of a genetic disorder in a family are discussed.

The Internet

Many people use the Internet to search for genetic information. However, not all information available on the Internet is relevant to each individual or family situation, and furthermore not all the information will be reliable. Importantly, therefore:

1) Any information you obtain from the Internet should be discussed with your doctor or counsellor.

2)The website should be from a reputable institution such as a hospital, university or government body. Furthermore, information about a condition should be authored and dated.

Useful contacts

Antenatal Results and Choices (ARC)
73 Charlotte Street, London W1T 4PN
Tel. (020) 7631 0285
Helpline Tel. (020) 7631 0208
Admin e-mail: arcsatfa@aol.com
Web Site: http://www.arc-uk.org
Information and support to parents at the time of antenatal testing and when fetal abnormality is detected.

Genetic Interest Group (GIG)
Unit 4D, Leroy House, 436 Essex Road
London N1 3QP
Tel. (020) 77043141
e-mail: mail@gig.org.uk
Web Site: http://www.gig.org.uk
A national alliance of organisations which promotes awareness and understanding of genetic disorders.

***Unique* -Rare Chromosome Disorder Support Group**
P.O. Box 2189, Caterham CR3 5GN
e-mail: info@rarechromo.org
Web Site: http://www.rarechromo.org
Information and support to families of children with any rare chromosome disorder.

Additionally, the Contact a Family Web Site http://www.cafamily.org.uk has a Glossary of Genetics terms, information on Patterns of Inheritance and addresses of Regional Genetics Centres. Our freephone Helpline 0808 808 3555 can also give information and support.

Disabled Children's Mobility Centres

The Department of Health has awarded a two year grant to Whizz-Kidz to establish six specialist mobility centres for disabled children and young people throughout England.

In 1986 the McColl Report recommended that children requiring wheelchairs and prosthetics should be assessed in designated clinics offering a child-friendly environment along with appropriate equipment and expertise. This is currently available through Disablement Service Centres around the country, and now, thanks to the grant to Whizz-Kidz, can be extended.

The Whizz-Kidz centres will specialise in the trial and provision of paediatric mobility equipment. They will also be on hand to offer impartial advice to families. training workshops and support to professionals. Contact: Whizz-Kidz, Tel. (020) 72336600

The Housing Needs Of Disabled Children: The Evidence

Joseph Rowntree Foundation

This study provides, for the first time, data at a national level on the housing needs of disabled children and their families, and their experiences of addressing these needs. The researchers - Bryony Beresford and Christine Oldman from the University of York - surveyed almost 3,000 parents of severely disabled children, and interviewed over 100 housing, social care and occupational therapy practitioners. They found:

1) Families with a disabled child experience far greater problems with their housing than families with non-disabled children. Nine out of ten families reported at least one difficulty with their housing, with many reporting multiple problems.

2) Difficulties with housing can be experienced by any family with a disabled child: not just where there is physical impairment.

3) Many families would prefer to deal with their housing problems by moving rather than adapting their current home.

4) Inside the home, the most frequently reported problem was the lack of space: space for play, for privacy or 'time out' , for equipment use and storage, and for carrying out therapies. Other common problems related to house condition arid access.

5) Over a third of families found the location of their home to be a problem, either because it was an unsafe place for the child or because of difficulties with neighbours.

6) Only a minority of families had received assistance from statutory agencies in order to address their housing needs. Typically, at a local level no single agency or department assumes lead responsibility for meeting the housing needs of disabled children. In addition, the lack of strategic information collection hampers improvements in service provision and delivery.

Background

Until the late 1990s little was known about the housing needs of disabled children, especially where those needs differed from the stereotypical issue of physical 'access'. Research published since then has drawn attention to the central role housing plays in the lives of disabled children and their families and, likewise, the inadequacies of current policy and practice in ensuring that families' housing needs are met. What has been lacking is the robust quantitative data needed to drive and inform change. This research provides such evidence.

What makes a home unsuitable?

Housing condition

Four out of ten families in the survey said their homes were cold, damp, and/or in poor repair. Poor housing conditions were reported more by families living in local authority housing compared with owner-occupiers and housing association tenants. Families with a disabled child are more likely to be living in poor housing conditions than families with similar incomes but non-disabled children. For example, 14 per cent of families in the current survey reported inadequate heating facilities compared with eight per cent of families participating in the 1999 Poverty and Social Exclusion (PSE) Survey.

Housing problem areas

In addition to poor housing conditions, there are a number of ways in which a home can be unsuitable for families with a disabled child. These 'problem areas' are located both inside and outside the family home. Table 1 shows the proportion of families reporting difficulties with each housing problem area.

Just under nine out of ten families were living in homes with at least one problem area, with one in four stating that there were six or more problem areas in their homes. Some of these problem areas may be experienced by any family with dependent children, but

Table 1: Proportion of families reporting difficulties with each problem area

Housing problem area	% of families reporting problem
Lack of family space (space to play, space apart from other family members)	55
'Functional rooms' (kitchen, toilet, bathroom) difficult to use due to size	42
Only one toilet and/or bathroom	41
Lack of space for storage equipment	38
Location	38
Access around, and in and out of, the home	33
Lack of downstairs toilet and bathing facilities	33
Housing condition	27
Lack of space to use equipment and carry out therapies	21
Inadequate facilities to meet carer needs re: lifting, toileting and bathing	21
Child's safety inside the home compromised	3

comparisons with equivalent general population datasets show they are experienced by a greater proportion of families with a disabled child.

Space
This was the most frequently reported problem and was experienced by families regardless of the nature of the child's impairment. Families needed more space for: play, use and storage of equipment, and 'space' or time out from each other.

More than one in two families reported a lack of space. Lack of space can be a problem for any family, yet comparisons with data from the 1999 Poverty and Social Exclusion (PSE) Survey suggest that families with a disabled child are much more likely to have problems with a lack of space in their homes (33 per cent PSE families, more than SO per cent families in the current survey).

Factors affecting housing need

Income
While families on the lowest incomes reported more difficulties with their housing, middle-income families also experienced considerable housing problems.

Tenure
Unlike families with non-disabled children, families with a disabled child are more likely to rent their home. Yet families renting from their local authority or a private landlord reported significantly more housing problems than owner-occupiers and those renting from housing associations.

Ethnicity
White families were more likely to be living in a home which was suitable to their needs than non-white families. Differences exist in the types of problems likely to be experienced by different ethnic groups.

Impairment
Difficulties with housing are reported by all types of families with disabled children: housing is not just an issue for children with physical impairments. For example, problems with the location of the home and difficulties with safety in the home were more likely to be reported where the child had behaviour and/or learning difficulties. In contrast, problems with access and equipment storage were most common among families with a child with physical impairments and/or a serious health problem. However, a lack of family space was likely to be experienced by any family.

Children with 'less severe' impairments

This research focused on families with a severely disabled child: children who typically have more than one type of impairment. However, the findings from this survey can also inform our understanding of the housing needs of children with less severe or single impairments as, for the first time, this research was able to explore the associations between impairment and housing need. In addition, the known association between disability and poverty means that the issue of poor housing condition is likely to remain present among families with less severely disabled children.

Dealing with unsuitable housing

Most families had already moved at least once in response to their child's needs or their needs as carers. The vast majority had made these moves without any professional advice or assistance.

At the time of completing the survey, half the respondents said they wanted to change their housing in some way so that it better suited their needs. Overall, moving home, as opposed to adapting the current home, was seen as the preferred option. Home-owners were the only group where adapting the current home was the preference.

Different problems with the home can require different solutions. A preference for moving was most strongly linked to living in a difficult location; needing larger toilet and bathrooms; and/or needing more space for using and storing equipment.

Housing need assessments

Interviews with relevant practitioners revealed that it is rare to find housing need being included in a needs assessment carried out under the auspices of the Children Act.

Three-quarters of families said they had not had their housing needs assessed by an occupational therapist. Among those who had been assessed, the outcome varied by tenure (see Table 2). Home- owners were most likely to have had changes made to their home. Almost a half of housing association tenants reported no change as a result of the assessment.

Funding changes to the home

One in ten families had received financial assistance with the costs of adapting their home (typically a Disabled Facilities Grant) from their local authority. Of these families, a third had had to make their own contribution to the funding of the adaptation.

Among those families who had been assessed as needing to make a contribution to the costs of an adaptation, one in three had been unable to meet these costs and the adaptation had not been carried out.

Table 2: Outcome of housing need assessment (percentages)

Outcome	Owner-occupier	Rent from Local Authority	Rent from housing association
Changes made to current home	60	51	39
No change	30	27	43
Moved/waiting to move (tenant)	N/A	6	16
Other	10	8	2

The current system

Interviews with practitioners and managers provided information about current practice in dealing with the housing needs of families with a disabled child. This revealed a number of examples of 'good practice'. However, this was not widespread and did not permeate the entire process of meeting housing needs -through identification, assessment, funding and delivery.

The effectiveness of current practice is undermined by a lack of clarity about the roles and responsibilities of the various (and potentially numerous) practitioners and departments involved in meeting the housing ne.eds of a family with a disabled child. Typically, no single agency or department takes overall responsibility for ensuring the housing needs of disabled children living in their authority are met.

Finally, a significant barrier to improving service provision and delivery is the lack of data currently collected by local authorities that could be used to map housing needs and inform planning and budgeting. In addition, there is no routine evaluation of the outcomes of adaptations or rehousing.

About the project

The project comprised of a survey of parents of one or more severely disabled children who were identified through the Family Fund Trust database. The sample is representative of the great majority of parents caring for a severely disabled child in England. Parents completed a postal questionnaire and a response rate of just over 60 per cent was achieved, yielding a sample size of 2,941 families. In order to compare the housing needs of families with a disabled child with those with non-disabled children, secondary analyses on datasets of families with dependent children and with similar income profiles were carried out. The datasets used for the comparative analyses were the 1999 Poverty and Social Exclusion Survey and the 1998/1999 Surveyof English Housing. In addition, interviews were conducted with senior housing, social service and occupational practitioners working in over thirty different housing authorities and related social service departments.

The full report, Housing matters: National evidence relating to disabled children and their housing by Bryony Beresford and Christine Oldman, is published for the Foundation by The Policy Press (ISBN 1 86134 483 X, price £13.95). It is available from Marston Book Services, PO Box 269, Abingdon, Oxon OX 14 4YN, Tel: 01235 465500, Fax: 01235 465556, email: direct.orders@marston.co.uk. (Please add £2.75 p&p for first book and 50p per book thereafter.)

Getting Help To Adapt Your Home

Contact a Family

If you need to adapt your home to make it easier for you or your child to manage then you may be entitled to a Disabled Facilities Grant, or Home Improvement Grant if you live in Scotland. For anyone considering an application for a grant, it is advisable to seek further help and advice as the system can prove quite complex.

England, Wales and Northern Ireland

Disabled Facilities Grants (DFG)
To be eligible for a DFG you must be an owner occupier, tenant (private, Local Authority or Housing Association) or landlord with a disabled tenant. The grant can help with the cost of the works such as building safe play areas, installing a stair-lift, adapting a lighting or heating system, or building a new bathroom facility. Whatever the proposed works it must be agreed that these are 'necessary and appropriate' in meeting the disabled person's needs and also 'reasonable and practicable'. The question of 'reasonable and practical' takes into account the age and condition of the property.

The grant is means tested and therefore income and savings will be taken into account. In the case where the disabled occupant is either aged under 16, or over 16 but under 19 and for benefit purposes is treated as a 'dependant', the income and savings of the parent(s) are taken into account. In most other cases it is the disabled occupant that is assessed.

There are two types of Disabled Facilities Grant:

1) Mandatory grants are awarded where the works are considered essential to enable better access and movement around the home and to essential facilities within it. The grant can also cover the costs of building essential facilities where necessary. The maximum grant payable is £25,000 in England, £20,000 in Northern Ireland and £30,000 in Wales. Note that if the costs exceed the limit then the council has the discretion to give a further grant covering the full costs of the mandatory works.

2) Discretionary grants (England and Wales only) can

be awarded to make the home more suitable In meeting the disabled occupants accommodation, welfare and employment needs. There is no maximum discretionary grant. Note that from July 2003 the Discretionary Grant will be abolished and replaced with Local Authority discretionary powers.

How to apply

Application forms are available from the local housing or environmental health department Applicants in Northern Ireland ShoUld contact their loCal housing executive office for a Preliminary Enquiry Form.

Once a formal application has been made the council must make a decision within 6 months In reaching a final decision the housing department is required to consult with the Social Services department to be satisfied that Ihe works are 'necessary and appropriate' It is important to note that applications for works already carried out will not be considered.

A further possible SoUrce of financial help is a Home Repair Assistance Grant This is a discretionary grant to help meet the CoStS of smaller scale works including adaptations, up to the value of £5,000 As well as being an owner-occupier or private tenant applicants must also be in receipt of a means tested benefit such as Income Support or Working Family Tax Credit or be 60 or over, or be disabled You may also qualify where the grant is to enable someone who is 60 or over, or disabled, to be cared for This might apply where the applicant provides regular respite care for a relative For further details and an application form contact the local housing authority.

Scotland

Home Improvement Grants

Grants are currently available from the housing authority to help with adaptations, but only If you're a homeowner or private tenant Local Authority and Scottish Homes tenants should approach their landlords for help to adapt their homes Housing Associations should arrange to have the work carried out for those tenants assessed as needing the adaptation.

Before any assistance can be considered the social work department will carry out an assessment of the disabled person's needs to determine if the works are necessary It is also worth noting that grants can be awarded for works to property where the disabled occupant intends to move to once the works have been completed.

There are two types of Home Improvement Grants available:

Mandatory Improvement Grants cover the provision of standard or adapted amenities such as a fixed bath and shower or additional amenities to meet the needs of disabled occupants. The maximum paid is half the cost up to £3,450.

Discretionary Improvement Grants (also referred to as a disabled grant) cover works to make the house suitable for the welfare, accommodation or employment of a disabled person. The grant will cover up to 75% of the costs of the works. With the maximum approved expense limit being £12,600, this means that the maximum grant payable is £9,450. If the cost of the works exceeds this limit then the Local Authority can apply to the Scottish Executive for an increase in the grant limit.

How to apply

The local council will give you an application form and tell you what information they will need to process the application. This may include information about income and savings.

New Grants in Scotland from April

In Scotland the system for receiving help with adaptations will change dramatically in April 2003. Current grants (Mandatory & Discretionary Improvement Grants) will be replaced with a new Housing Grant which could meet 100% of the cost of works. The most significant change is the introduction of a means test for applicants. At the time of writing very little information was available about how the system will operate, but it is understood that a guide will be published by the Scottish Executive in April.

Challenging decisions

If you are unhappy about the outcome of an application for a grant, or the length of time it has taken to reach a decision, then seek further advice. Ways of challenging decisions include talking to your local councillor, complaining using the Local Authority complaints procedure or asking the Local Government Ombudsman to investigate. The local Citizen's Advice Bureau or Home Improvement Agency (see below) may be able to advise further.

Further Help and Advice

Home Improvement Agencies

Home Improvement Agencies, sometimes referred to as 'Care and Repair Agencies,' exist to help home owners or private tenants improve their living conditions. This involves giving technical and financial advice as well as help and guidance through the application process. They may also be able to identify other sources of financial help if the grant is insufficient.

For information about local agencies in England contact Foundations, Tel. (01457) 891 909. For Scotland contact Care and Repair Scotland, Tel. (0141) 221 9879. For Wales contact Care and Repair Cymru, Tel. (029) 20576286. If you live in Northern Ireland a local disability advice project should be able to help further. The Family Fund Trust The Family Fund Trust also produce a helpful factsheet entitled 'Adaptations to Housing' for each nation country. For a copy, Tel. 08451304542.

Fashion For All

Contact a Family

Does finding practical, comfortable, affordable clothing for your child mean sacrificing on style? Are High Street retailers catering for your needs? This article by Awear first appeared in RNIB Eye Contact magazine, Issue 34 (Autumn 2002).

Awear is an organisation of disabled people and fashion professionals who are promoting the importance of fashionable clothing for everyone.

We are challenging the fashion industry to change the way they think about design and manufacture so disabled people can have full access to fashionable, affordable clothes that fit well and suit their lifestyle. Fashion is important from an early age in fitting in with peers and developing a sense of individualism and style.

As the choice is so restricted on the High Street, many fashion conscious teenagers choose to sacrifice comfort for style. They want those "must have" pair of jeans, that "everyone at school has", despite the fact that they may cause pressure sores from the seams or are not cut for someone using a wheelchair.

At Awear, we believe thatHigh Street retailers and manufacturers should change the way they think about who their customer is. There are currently 9.1 million people with disabilities in the UK, with a spending power of over £40 billion a year (Yourable.com). However the disabled customer still experiences discrimination when it comes to access to fashion. Poor access into and around shops, narrow aisles and inadequate changing facilities, added to the fact that the majority of garments are not designed with inclusion in mind, makes the shopping experience of most people with disabilities extremely frustrating.

Realising potential

Awear is therefore helping retailers realise their full market potential by introducing a national Accreditation service which rewards retailers that provide good access to the physical environment and good customer service, and will eventually highlight inclusive product design. By introducing low level changing mats big enough for older children and adults in their Milton Keynes store, Debenhams for example, showed a commitment to inclusive shopping for all and have been awarded an Accreditation Certificate.

With a grant from the Community Fund, Awear is developing computer aided design (CAM/CAD) software that can make patterns to individual body shapes. This groundbreaking technology creates the possibility of High Street fashion for all, as it works with mass-production methods, as well as being useful for the smaller business or home sewing market. It is our vision that one day it will be possible to purchase made-to-measure garments on the High Street for the same price as mass-produced clothing.

Awear evolved out of the National Association of Clothing Workshops, which was set up in the late 1980s. This small group of non-profit Clothing Services still exists providing a unique service to disabled people and their carers. Each service provides a made-to-measure and alteration and adaptation service to customers, without passing on the full cost of the production of the garments. This provides a valuable resource for families of young children especially as the cost of replacing out-grown or worn out clothing can prove to be a huge drain on the income of a family of a disabled child.

Finding solutions

With more disabled children now attending mainstream school, the need to fit in becomes all the more important. The Clothing Advisors at each clothing service are able to work with their customers to find solutions to everyday clothing difficulties. By changing fastenings on shop bought garments, children can gain more independence in dressing, or by clever design, clothes can be made that enable children who rip or remove clothing to stay dressed while the clothing remains damage free.

Design challenge

Last year. Awear set the first Design Challenge to students from Nottingham Trent University. The challenge was to design a collection for a high street retailer that had inclusion incorporated into the designs. The project brought together fashion students, clothing services, made-to-measure retailers and disabled young people who worked as models and acted as advisors on the project. The result was fantastic. Clothes were designed that were both practical and wearable by people with various impairments as well as being fashionable and suitable for the made-to-measure market.

While people with disabilities have to compromise on their individual style because the choices available to them are limited, they are being restricted from expressing their true selves. As one person put it, "Disabled People both need and want to look good. The beauty of Disabled People has now to be emphasised. We are gorgeous but we need the right gowns for the ball".

How you can help

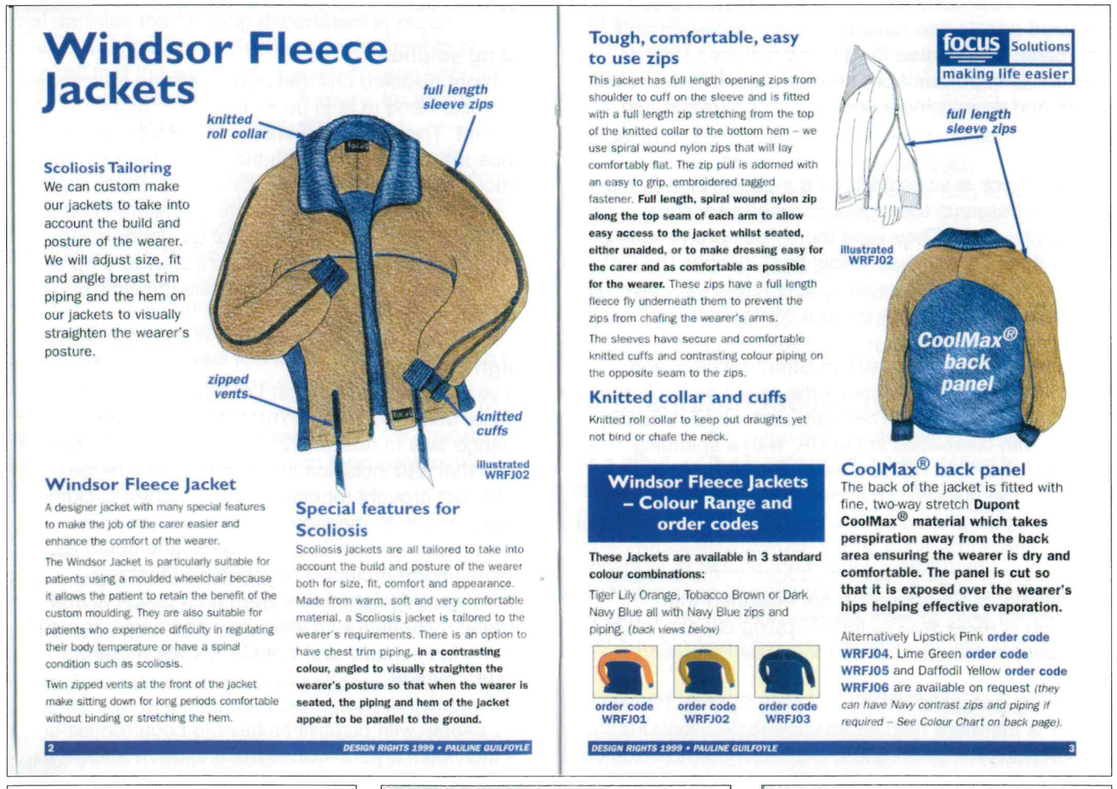
We would like to see every child wearing something they feel good in at the next school disco or family event, which is why we need your opinions and experiences. In order to ensure that our Accreditation standards are representative of all disabled people, we encourage you to inform us of any good or bad practice you encounter via Awear's Annual Clothes Shopping Survey. We ask you to tell us about retailers you have visited and each reply is entered into our £250 worth of clothing prize draw. The information gathered from the survey enables us to compare the level of service

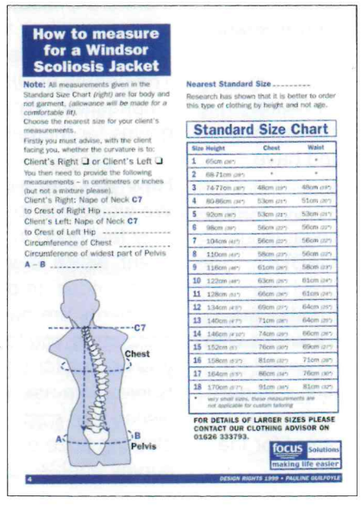
experienced by disabled and non-disabled people within UK retail outlets and monitor progress towards inclusive shopping. For a copy of Awear's Annual Clothes Shopping Survey questionnaire or just more information please contact: Awear (UK) Ltd, Notts International Clothing Centre, Annesley Road, Hucknall, Nottingham NG15 BAY.
Tel. (0115) 953 0439
e-mail: enquiries@awear.org.uk,
Web: www.awear.org.uk

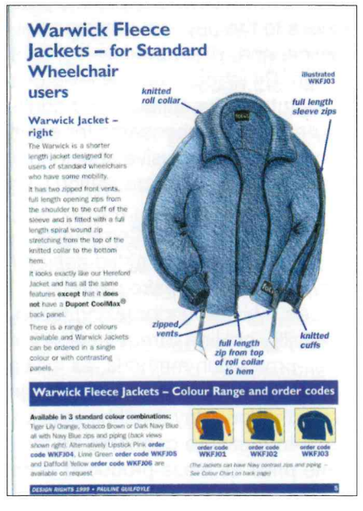
Specialist Clothing - Your Advice

Further to our question in the Winter 2002 Newsletter, we have had a number of answers with regards to specialist clothing. Julia Duckmanton used to buy all her daughter's clothing from, Fashion Services For People with Disablilities, Greefield Centre, Green Lane, Baildon, West Yorkshire, BD17 5JS, Tel: 0274 597487.

Anne Hill has sent us a brochure from Focus Solutions, a company who produce designer clothing which can be custom made. Solutions for the Disabled, 116 Queen Street, Newton Abbot, Devon TQ12 2EU, Tel: 01626 202517







Transportation

London Congestion Charging

Congestion charging...

Individuals or institutions in receipt of a Blue Badge or Orange Badge are eligible for the 100% discount from congestion charging. This applies to individual Blue Badge holders throughout the European Union. After registering and making a one-off payment of £10, holders of this discount are not required to pay the congestion charge when they enter the congestion charging zone.

For more information visit www.cclondon.com or phone 0845 900 1234

Parking boost for blue badge holders

The government has agreed to press ahead with improvements to the disabled persons parking scheme.

The improvements, part of a UK- wide study of the blue badge scheme, include reinforcing the eligibility criteria for badges and considering the feasibility of a national database of badge holders.

The changes were put forward by the Disabled Persons Transport Advisory Committee, an independent advisory body.

The government has also announced cash support for the "Baywatch" campaign that encourages the protection of disabled persons' parking bays in supermarket car parks.

Meanwhile, disabled people in London are to receive a further boost from 1st April when they will receive 24-hour free travel on tube, bus and tram services. At the moment they are entitled to free travel only after 9am. This brings them into line with the benefits offered to blind people.

Direct payments

A woman recently launched a test case at the High Court to allow her to use direct payments from her local authority to pay her husband to care for her.

Currently the Community Care (Direct Payments) Regulations 1997 forbids direct payments being authorised to secure services from a person's partner or close relative ie. parent, brother or sister.

A government consultation on the Community Care (Direct Payments) Act 1996 which closed at the end of 2002 also states that the regulations prohibit direct payments to a partner or other close relative living in the same household.

Recently the woman was granted leave for a full judicial hearing. If successful, other people who have been refused direct payments on similar grounds could also challenge their authority.

New MPSI Web Site

A new website has been developed to provide those affected by MPSI, their families and professionals information on Hurler disease and the attenuated forms of the diasease, commonly known as Hurler Scheie and Scheie disease. This website gives valuable information on the disease phentypes and details of on going clinical trials. Visit www.MPSIdisease.com

Sure Start Guidance

Supporting families who have children with special needs and disabilities.

During the later part of 2001, Contact a Family was asked by the Government's Sure Start Unit to produce a guidance document on working effectively with families who have young disabled children. This publication became available in November 2002. The guidance is for all Sure Start Programmes in England, although Contact a Family is currently discussing the possibility of adapting it for use in Scotland, Northern Ireland and Wales.

The purpose of the guidance is to:

Ensure that all families who have disabled children have access to and get a good quality service from Sure Start programmes.

Ensure that all partners in Sure Start programmes consider access and quality issues when designing Sure Start programmes.

Help programmes develop an awareness of and respond appropriately to the needs of families who have disabled children.

Help programmes to build and share knowledge about existing Information and support available to parents and carers of disabled children.

The guidance has been produced so that it will become a practical guide to staff and organisations in Sure Start programmes. As well as providing advice in areas such as how to support parents through diagnosis, key - working and accessing financial help there is a pullout 'Good practice checklist' which can be displayed in offices and other Sure Start facilities.

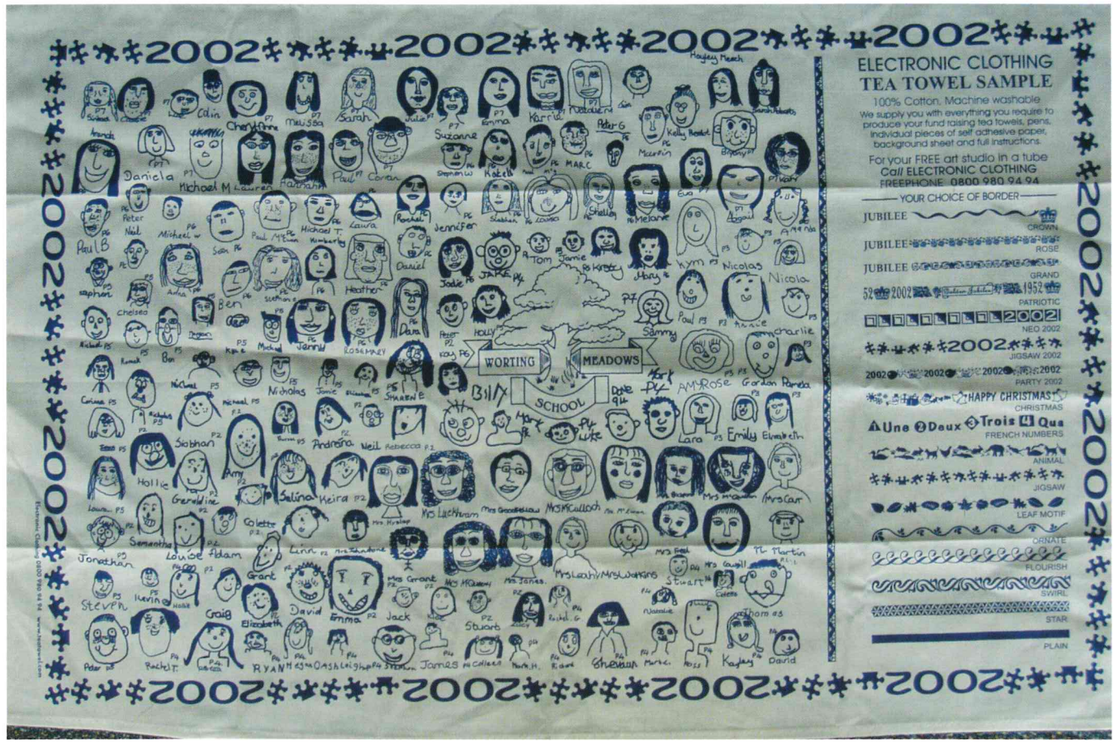
There are also helpful appendices made up of a glossary of terms, initiatives and programmes that support families, useful organisations, and references and resources. All Sure Start programmes in England have been sent copies of the guidance along with some information about Contact a Family.

If you would like a copy of the document it is available free of charge from DfES Publications, PO Box 5050, Sherwood Park, Annesley, Nottingham NG15 ODJ Tel. 0845 6022260 e-mail: dfes@prolog.uk.com Web www.surestart.gov.uk

Jeans For Genes Appeal

Christine Lavery - Director

Help MPS To Create Its Own 21st Birthday Tea Towel



The Society is inviting all affected children and their brothers and sisters to draw a picture of their faces rather on the lines of the tea towel above. If bereaved brothers and sisters want to draw a picture of their MPS sibling for the tea towel this would also be welcomed. In order for this to be a success we need to receive at least 200 faces.

The Tea Towels will be available at the MPS National Conference and through mail order to the Society.

Email, telephone, fax or write to us for your special pen and paper. Ask now as we can't send this out after the 15th April. We must have your drawings by 25 April 2003.

Caversham Charity Folk Festival 20-22 June 2003

This year we are holding the 10th CCFF and again it is Free Entry but be prepared to be pestered for a donation to our charity fund supporting: Berkshire Autistic Society; Berkshire Multiple Sclerosis Therapy Centre; British Red Cross Society; Cruse Bereavement Care; Cystic Fibrosis Trust; The Guide Dogs for the Blind Association, Reading; The Society for Mucopolysaccharide Diseases.

One way to give and avoid the pestering is to purchase a Collection-Free Zone Badge for just £5! All the artists and facility providers give their services free and so almost every penny collected goes to help the charities.

The main Festival site is on the south bank of the Thames at Caversham, in Reading, adjacent to the Holiday Inn hotel on the A4155. It is an ideal location for camping at £7/head (book with Mike Tierney on 0118 975 1016).

Additional events, including blues sessions, take place at local bars and at the Holiday Inn, where the Sunday evening concert with guest Roy Bailey is hosted by The Readifolk Club. In the main marquee the headliners for the weekend are Jez Lowe, Paul Downes and Mike Silver, supported by Alan Franks & Patty Vetta, Mouse, JCB & Betty Davila, Kath Tait, Ron Trueman-Border, Warblefly and a host of our favourite local acts.

There will be a craft fair, children's entertainment, real ale & singarounds in the beer tent, dance performances from local morris sides and the opportunity to enjoy a boat-trip down the Thames to the strains of local musicians. For a location map for the site try www.multimap.com on the internet using the postcode RGl 8BD and for the latest information see our web-site at www.ccff.org.uk or email info@ccff.org.uk.

Donations

The D'Oyly Carte Charitable Trust
DM Charitable Trust
DLM Charitable Trust
Amersham Plc
Mrs Elizabeth Jenkins
James Radford
Wessex Water
Shawforth Methodist Church
Bunney Reckitt trust
Wilkinson
Rio Tinto plc
Charity Flowers
Gone to Pieces
Mr and Mrs Lloyd – West Midlands
Heath Townswomens Guild
SIG plc – Sheffield
Yvonne and Kevin Puddy
Mark and Debbie Burniston
Mrs L Alexander
B & N Carpets Ltd
Mr V G Richardson
Porsche Cars – Great Britain Ltd
Land Securities Group PLC
Tomkins plc
TKT Inc
TKT 5s
Genzyme Corporation

COLLECTION BOX
Huddersfield Bus Station
Mary Wearing
Mr Mervyn Albert Short
James Bernard Edwards

Fundraising

The Society is grateful to the following who held fundraising events

Vicki & Samantha Brockie – Chocoholics Party
Kathy Parkinson and Colleagues – In Lieu of Xmas Cards
Judey Lloyd – Solihull
Towersey Morris Men – Mummers Play
Northgate – Recycled Paper Sale
Solihull Borough Council – London Marathon 2002
The Boomerang Golf Society – Peter Stuart
Darwen Moorland High School – Lancashire
Mr and Mrs Cocoran – Charity Race Night
First Group Huddersfield
Woodland Glade Sports and Social Club – Huddersfield
Nationwide Building Society – Huddersfield

Stamps & Foreign Coins

Hillside Pre-School, Dorset
Wiltshire County Council
Jennifer Norsworthy, Plymouth
Mr and Mrs Garthwaite, Jersey
Rachel Todd
Ben Williams Family & Friends

In Memory

Robert Mayhew
Aaron Martin
Kim Eggleton

The Daniel Thwaites Charitable Trust have donated £257-50 for the Society to purchase and donate a 21" colour TV and video recorder to the Enzyme Replacement Therapy Treatment room at the Willink Genetics Unit, Royal Manchester Children's Hospital.

MANAGEMENT COMMITTEE

Chairman Barry Wilson

Vice-Chair Judy Holroyd
Bob Devine

Treasurer Judith Evans

Trustees Ann Green
Sue Peach
Wilma Robins
Adam Turner
Chris Holroyd

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Alison West	Development Officer	a.west@mpssociety.co.uk

YOUR HELP NEEDED

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.

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

NEWSLETTER DEADLINES

SUMMER
30 June 2003

Autumn
30 September 2003

Winter
17 December 2003

Spring
31 MArch 2004

SPECIAL DAY OUT AT UNBEATABLE PRICES
ALTON TOWERS

FOR MPS MEMBER FAMILIES

ATTENDING THE MPS SOCIETY'S AGM

ALTON TOWERS HOTEL

9.30am SATURDAY 10th MAY 2003

All MPS parent and adult members can purchase a maximum of 2 adult tickets and a ticket for each of their children for a total of £20.

This exceptional rate is dependent on the parent members attending the Society's AGM and tickets will be handed to families at the end of the meeting.

The AGM will start promptly at 9.30am and it is anticipated that it will be over by 10.00am.

MPS families may reserve accommodation at the Alton Towers Hotel on Friday 9th May and/or Saturday 10th May 2002 for £170 per room per night for up to 4 people including continental breakfasts. Please book directly with the hotel stating that you will be attending the MPS AGM.

Notice of Annual General Meeting

Notice is hereby given that the Annual General Meeting of the Society for Mucopolysaccharide Diseases will take place at the Alton Towers Hotel, Alton, Staffordshire on 10th May 2003 at 9.30am

HOW TO BOOK FOR THE ALTON TOWERS HOTEL

You need to have this information ready as you phone:

- The dates you wish to stay
- The number of children and adults in your party
- The number of rooms required
- Any special requirements you may need
- Your method of payment
- State that you are attending the MPS AGM

Now phone 08705 00 11 00