NEWS

LETTER

mps

THE SOCIETY FOR

MUCOPOLYSACCHARIDE DISEASES

FREE TO MEMBERS

YOUR COUNCIL OF MANAGEMENT FOR 1982/3

CHAIRMAN	Mr G.C. Nichols, CBE, DFC, 'Mossville' Cokes Lane, Chalfont St. Giles, Buckinghamshire.
HON. SECRETARY	Mrs. Christine Lavery 30 Westwood Drive Little Chalfont Buckinghamshire. Tel: Little Chalfont 2789
HON. TREASURER	Mrs Catherine Grant 'Roselawn' 43 Woodside Close, Amersham, Buckinghamshire. Tel: Amersham 4799
HON. FUND RAISING OFFICER	Mrs. Susan Heath 'Prospect House' 133 White Lion Road, Amersham Common, Buckinghamshire. Tel: Little Chalfont 2029
MEDICAL OFFICERS	Dr. D. Garrow, BM, FRCP. Dr. B. Neal, MBBS, MRCPG, D.Obst. RCOG.
GENERAL MEMBERS	Mrs. M. Hourigan Mr. R.J.O. Lavery.

AREA SUPPORT FAMILIES

Mary and Colin Gardiner, 35 Church Rd. Banks, Southport, Merseyside.
Derek and Pat Kirkman, 63 Higher Shady Lane, Bromley Cross, Bolton, Lancs
Robin and Christine Lavery, 30 Westwood Drive, Little Chalfont, Bucks.
Neil and Jane Reid, Meadowlark, 9 Huddleston Way, Sawston, Cambs.
Robin and Anne Ridley, 2 George Road, Lutterworth, Leicestershire.

IMPORTANT NOTE: The articles and views expressed in this publication are not necessarily the views of the Society.

AIMS OF THE SOCIETY

1. To act as a parent support group
2. To further research into MPS Diseases.
3. To educate the public about Mucopolysaccharide diseases.

NEW YEAR LETTER FROM THE CHAIRMAN

When the Society got off the ground six months ago no one them involved, either the area support families or the council of management, had any idea that its effect would be so immediate. We had a shrewd idea based on the experiences of families with MPS children that a support group of sorts was indeed necessary.

The number of sufferers around the country is not precisely known but an intelligent guess is about 400. Since the population of the UK is about 56 million our children are something of a rarity, and rather special. Of this estimated count nearly 100 families have of their own volition contacted the area support groups, and new approaches continue on, at least, a weekly basis.

The first newsletter was circulated to 225 people/families, including a great many in the medical world, who had themselves been in touch with us. The first print run of 1000MPS Information sheets has long been exhausted. Besides the medical profession, teaching and general hospitals and medical faculties of universities these have been dispensed to individuals, voluntary organisations clubs and commercial companies who have already rallied round to support with fund raising activities or donations.

The Aims of the Society are stated in the Newsletter. Already we have gone a long way to put parents in touch with each other and have been surprised and joyed by the response from the public. We have been looked on with favour by the medical profession. Quite apart from the support from our Medical Advisors, we have been approached by hospitals around the country for background information or to suggest that we might contact their families with 'lay' information. In this Newsletter, Dr. Philip Benson of Guy's Hospital has kindly contributed an article which I known will be read with keen interest. The Society has also been featured in the Health Visitor Magazine.

As the early months were concerned with making contact with families and setting up the Society we did not expect to make much headway in attracting funds towards further research. Therefore, it is a considerable achievement that by the end of December 1982 almost £2,000 had been banked and we are grateful to those who have entrusted money to MPS. Susan Heath drops a number of ideas in the Newsletter which if taken up on a regional basis should see us set to acquire funds of a considerable amount by the year's end. The operating costs of the Society have been minimal since we have relied on voluntary effort. Only in December did we think it right to have produced letter headed paper and even this is marred by the fact that Christine insists on bashing out (no better description is appropriate) letters on an old and battered typewriter which cost her £8. Through the kindness of the Sainsbury Memorial Trust who late in 1982 donated an additional sum of £900 for the express purposes of supporting the administrative printing, postage and publicity expenses of the Society for 1983, We should for the foreseeable future be able to give the assurance that the funds you have been able to raise will not be diverted to the running of the Society, even though I view this as a legitimate and worthy claim on a proportion of the funds.

And what of 1983? Christine has already circulated tentative ideas for an MPS family weekend in September/October. This must be an instructive, happy and worthwhile occasion for all, particularly as whatever venue is chosen it will mean hundreds of miles of travelling for many. And it will take some organising. She will be in contact soon. Considering that various clubs and organisations with very little knowledge of MPS have responded handsomely we know that we can spread MPS awareness and fund raising much further in 1983. We will wish to maintain our constructive links with the Medical profession, and invite all readers to correspond or contribute to the Newsletter. Whilst the UK is now independent with regard to prenatal diagnosis and the furthering of research and treatment of MPS we appreciate what international cooperation can do. We will continue to foster overseas links in addition to our present contacts in Australia, South Africa and the United States. I won't pretend that reading the experiences of MPS families in the Newsletter is anything but harrowing. Their frankness is commendable when we are all seeking instruction and example. One can only try to imagine the hard slog and weariness which is a common feature in providing the love which an MPS child deserves. Nevertheless, I know that most MPS families have positive attitudes which rise above these pressures. If you reflect, no matter the child's physical or mental condition he/she is special with a funloving personality bursting to get out from behind the facade. They deserve a dignity and a quality of life which you and this Society will do its best to develop in 1983.

A Time for a Break

There were many times before our little daughter Sarah died that I, especially, felt desperate for a break but this was made impossible by the unavailability of anyone who I felt confident enough to look after Sarah - if only for a day.

Now we no longer have Sarah with us I feel left with lots of experience and understanding but unable to use it. We are lucky to have Mark who is a normal happy 2½ year old.

We feel we could give other parents a well-needed break even if only for a day or weekend, would be completely happy taking on any of our special little children to give them the same devoted care too as we gave our own little girl Sarah, in our happy family environment.

If you as parents feel that we could help you in this way, please do not hesitate to contact us by phone or write.

Anne, Robin and Mark Ridley, 2 George Street, Lutterworth, Leics. Tel: Lutterworth 2405.

Dear Friends,
I'm Mary Gardiner, my husband is Colin and we enthusiastically volunteered our help as a support family to Christine and hope that we will be of value to you in the years to come.

Colin and I have a daughter Catherine who is eight years and healthy. We did have a little boy Richard who had Hunter's Syndrome, and as you know that's not easy to accept. When Richard was diagnosed at two years we were told his and our only hope of a healthy life for him was through a bone marrow transplant. With the information given we weighed the bone marrow transplant risks against the prognosis if he was left untreated, and decided it was in Richard's best interest to go ahead. As Colin, Catherine and myself were unsuitable donors there followed six months of travel to Westminster Childrens' Hospital searching for a perfect unrelated donor from the Anthony Nolan Lab. Richard had 11 matches and 19 near-matches. Unfortunately, all these proved incompatible once matched in the Lab, so the attention was again on the family and this time they used a technique called autoblast immunisation on Colin and one of my brothers. Eventually they decided upon Colin.

Richard entered the Westminster and had his B.M.T. on April 22nd 1982. There is a lot to a B.M.T. in terms of drugs, making a child and his environment as sterile as possible and the affect all this has on a child and his family. The enormity of it all is too much to expand on at this point and I hope another parent will give those details. What I can write about is the way Richard accepted and coped bravely with it all. Richard was nursed initially in a Vickers isolator which was roughly the size of a small bedroom with a kitchen at one side of it which left little room for him to get about as freely as he would have liked. However, he and I managed and both of us kept as cheerful as possible.

Fortunately, the new marrow grew straight away and seventeen days after the transplant Richard was tucking into yoghurt to recontaminate him with germs. Three days later and once again Colin and I could take hold of our little boy without the protective plastic between us. I remember the medical team telling Richard he could come out and him climbing up onto his bed and announcing "I like it here" - laughter broke out as they had had three weeks of him and me asking when he could come out. Eventually Richard did come out through the rubbish bin and into the arms of his daddy. We all cried and laughed together, it was a wonderful feeling. All the months of worry and anxiety seemed to lift and we could, at last, look to the future with new hope. Richard had come through the transplant and he had the missing enzyme.

In the time I spent at Westminster I met other families searching for the same chance of a life for their child. We shared the same urgency that everything must be alright and somehow we got through those long agonising days.

Richard's main concern was to go home and he plagued the doctors every day with the same question. "Can I go home?" Richard did come home and his face was a picture of happiness. He searched the house checking that nothing had changed.

Sadly for us, all was not well with Richard and he returned to the Westminster where he died on the 3rd July 1982 after a very brave battle by him and the medical team.

There are many reasons why everything went so wrong for us, but we accept the medical team's view that it was a candida infection associated with the B.M.T. We considered that Richard had had enough whilst alive, and refused a post-mortem.

As some of you know the loss of your precious child is something that you never 'get over'. We consider ourselves blessed that Richard was our child, and he has left us some wonderful memories. As the days pass we try very hard to think of them and not the sad and emptiness his death has left.

For the love of Richard let us help you.

Mary, Colin and Catherine.

Fundraiser's Report

First and foremost, a big thank you and congratulations to Audrey Hodges, Lyn Windsor and Jean Towning who have raised large sums of money for our cause. Keep up the good work. A lot of their success has been due to local publicity in their home towns from the press, so if any of you are prepared to give an interview to your local paper, do get in touch with them, preferably with a fund-raising scheme up your sleeve, and see if any local groups, churches or schools will help raise money as a result.

Now for our big national fundraising effort which will hopefully take place this Spring - Christine and I are organising a 'coffee snowball', whereby we start off with eight people inviting seven people to coffee at a charge of 50p. per head; those seven then invite six, and so on down the line. Provided nobody drops out, we should be able to raise a staggering £54,000, though our initial administration costs will have to come out of that. Some of you have already been invited to participate by proxy, as obviously we want this to take place all over the country, but if any others want to be involved, bearing in mind the people you invite must be prepared to continue the chain or hundreds and possibly thousands of pounds will be lost from just one person dropping out, please contact us as soon as possible.

If any of you are having bring and buy sales, it might be worth contacting your local manufacturers and shops to see if they would be prepared to donate anything towards a tombola or a raffle. It's also worth seeing if any local organisation would be prepared to collect tin foil or newspapers on your behalf, provided you can get a good price for them from the recycling dealers.

Happy New Year to you all, and happy fundraising.

Susan Heath
Fundraising Officer.

PRENATAL DIAGNOSIS AND GENETIC COUNSELLING IN THE MUCOPOLYSACCHARIDOSES

There are many types of genetic counsellors. The process starts with the family doctor, who because of the complexity of biochemical genetic disorders usually refers the patient to a consultant paediatrician. From the symptoms and physical examination, X-rays and other investigations, the paediatrician can usually make a shrewd guess at the diagnosis, but before accurate counselling is possible, sophisticated biochemical tests are essential. These are carried out in only a few laboratories in any given country, which are usually part of specialised genetic centres. The final test which clinches the diagnosis is measurement of enzyme activities either in the blood, or in cultured fibroblasts, that is cells, grown from a small piece of skin. For some of the mucopolysaccharidoses (MPS) for example Morquio disease type A or Sanfilippo disease type C, only one or two laboratories in the United Kingdom have the expertise to make an accurate diagnosis. For these reasons, the final counselling is best carried out by a geneticist with experience in the interpretation of the biochemical findings.

The questions most commonly asked by parents at counselling clinics are 'will it happen again?' 'how long will he/she live?' is there any hope of treatment?' 'why did it have to happen to us?' The last question is often associated with a feeling of guilt, knowing that the mother in Hunter's desease or both parents in the other MPS diseases were the carriers.

Having made a precise diagnosis, the counsellor answers these questions as accurately as possible. In Hunter's disease, both the mild and severe forms, the genetic risks are that one in two of the brothers will be affected, and one in two of the sisters will be carriers. In all the other types of MPS, irrespective of sex, the chances are one in four that brothers or sisters will be affected.

In some families there are younger children who appear normal at the time the diagnosis is made in their sibling. Parents usually ask for these to be tested. Awaiting the result of the tests is a dreadful experience for the parents. I know of two families where an older child was found to have a form of Sanfilippo disease and on testing all the younger children who were considered to be normal, three in one family and two in another were found to have preclinical Sanfilippo disease. In one of these the mother was pregnant and requested amniocentesis to test if her foetus (or unborn child) was affected. It was, and the family unhesitatingly requested termination of the pregnancy.

Termination of pregnancy (or medically-induced abortion) is a controversial procedure. Some religious groups such as the Roman Catholics will not allow it. Others, such as the Jewish, are not completely dogmatic - some Rabbis will accept it, others won't. I know of a Catholic family and a Jewish family who were not allowed amniocenteses by their priest and Rabbi respectively, and consequently had a second affected child. When the mothers became pregnant for a third time, they abandoned their religions and requested amniocentesis. The test is usually carried out at 16 weeks of pregnancy in hospital by an obstetrician under direct visualisation of the foetus and placenta by ultrasound scanning.

The mother returns home the same day. The baby is in a sac surrounded by amniotic fluid. Some of this fluid is aspirated through a syringe. The cells in the fluid are mainly from the foetus; some are alive and can be grown in the laboratory. Three to six weeks later, the enzyme test can be carried out on the cells. Since the baby passes urine into the amniotic fluid, this is tested for excess mucopolysaccharides, which if present allow for a prenatal diagnosis within a few days after amniocentesis. This is a useful second test because in about one in 200 times, cells from the mother overgrow those from the foetus and can lead to an error in diagnosis.

Twin pregnancies can present further problems. At amniocentesis, each sac must be aspirated separately, and mistakes have occurred. In a pregnancy not in the UK, one foetus was found to have Hurler disease and the other to be unaffected. At the parents' request, air was injected into the heart of the affected twin, who died. The pregnancy continued until the birth of the normal twin.

The risk that amniocentesis can itself cause abortion is small - about 1% above the average risk of spontaneous abortion,

Some parents who reject abortion decide to adopt further children or (except in the case of the Hunter diseases) opt for artificial insemination.

It is the counsellor's responsibility to explain these biological and procedural risks. It is them up to the parents to decide on what action to take in planning their future family.

Philip F. Benson
Paediatric Research Unit,
The Prince Philip Research Laboratories,
Guy's Hospital Medical School,
London SE1 9RT.

FOOD FOR THOUGHT

Just suppose every MPS family in the Society managed to raise £120, by this time next year we could afford the salary of a full-time researcher solely looking into MPS for one year.

It is worth looking into the party-plan idea, such as Tupperware, make-up parties, clothes parties, Usborne books etc., as most of these will give you at least 10% profit on takings if done in aid of a charity. If anyone wants more ideas, please drop me a line.

Susan Heath

We are Michael and Pat, and our two children are Richard 7 and Helen 3 and 9 months. To anyone meeting us we are a perfectly normal, run of the mill, family. Nothing particularly exciting or outstanding about any of us and yet like so many of you reading this we have had experiences that have altered our 'ordinary' lives so drastically, we never can be 'normal.' We have a daughter with Hurlers Syndrome.

At this present time I am not going into detail of Helen's diagnosis, treatment or the traumatic times we have had in the past 3½ years, many of which you will be, or have, experienced yourselves. There is such a tremendous difference in communicating with fellow sufferers than with general outsiders. However, I would like to write about the brighter side of a little girl we, as parents, feel very humbled to have as our daughter.

Helen and I were admitted to Westminster Childrens' Hospital on 17th June 1981 for a bone marrow transplant using Richard as donor. He was not a good match but at least a sibling and the best available. When we made the decision to use Helen for this experimental treatment, only one child had been treated and by the time Helen was admitted, another boy had received a successful transplant from a perfect match donor. Within an hour of being admitted to WCH, treatment began in earnest and we were to undergo two transplants with Michael being the second donor before success. Helen was dreadfully ill during these months and we lived, and still do, on a day to day basis.

At the end of October, we finally returned home after what seemed a lifetime of having our family separated. Trying to pick up the threads was constantly interrupted by frequent planned and emergency trips to London, but almost 18 months have passed since the second transplant and the obvious question is: How is she? The most effective way to answer that is in a purely medical format.

Liver and spleen: were enlarged and are now back to normal size. This was the first improvement.

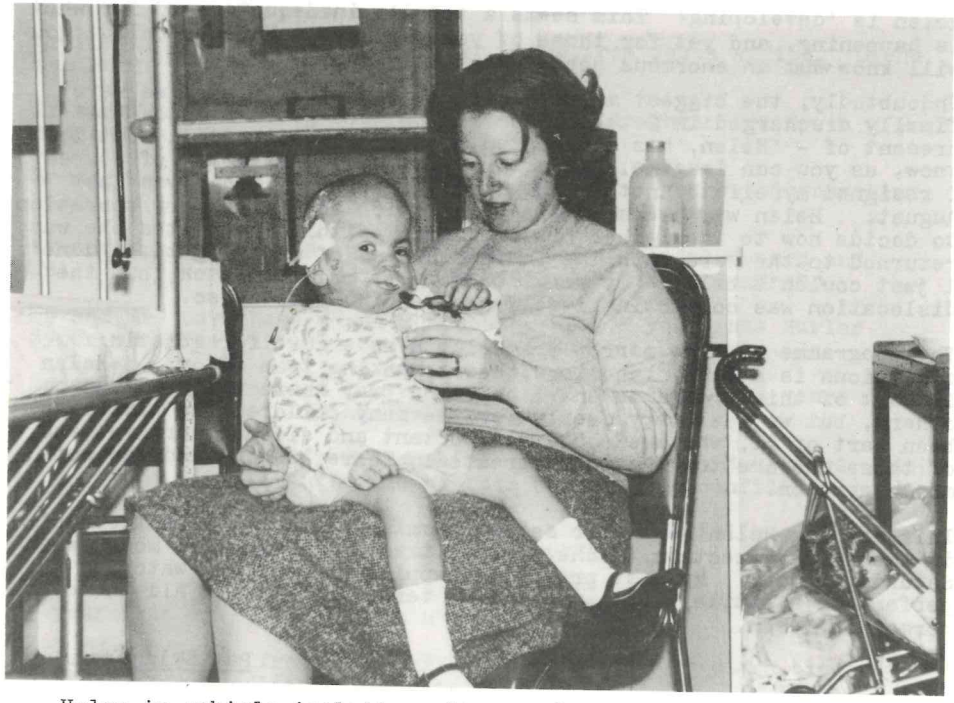
Corneas: were quite cloudy although vision was not impaired. They are now brighter and clearer. A forthcoming examination will give more accurate results.

Joints: were stiffening and mobility becoming more difficult, fingers and toes becoming 'clawed.' If I say Helen uses her bed as a trampoline, attempts the splits, walks on tip-toe and generally has very few restrictive movements, it shows they are most certainly not getting any worse but showing very promising signs of improvement.

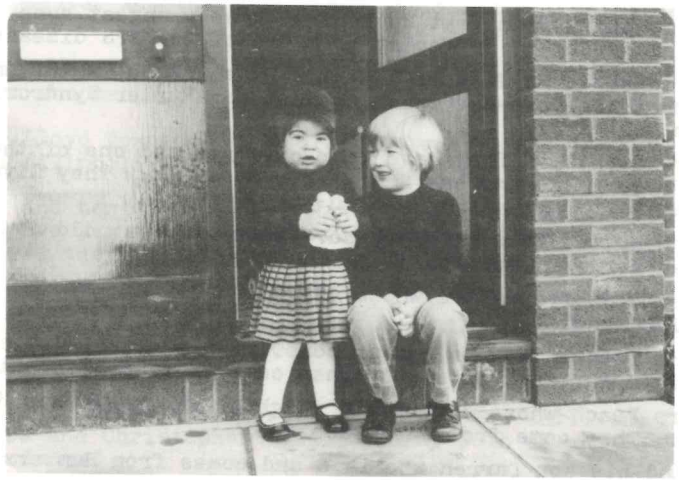
I.Q.: Groan, groan! A very frustrating thing to assess accurately I feel. A Ruth Griffiths test was carried out in March 1980 which proved her to be 14 months behind (pre-transplant it was approx. 3 months) so her period of isolation was taken into consideration. Just before Christmas, another one was done, of which I do not yet know the accurate results, but it seemed she had caught up a great deal and was almost normal.

Facial Features: began to deteriorate at 18 months and are now reversing quite considerably, which the photos may show. During and after the transplant she was almost unrecognisable, due to high doses of steroids and side effects of other drugs.

There are many changes occurring which are difficult to describe.



Helen in cubicle isolation after coming out of the bubble. The tube is because she refused to take her drugs and the spots are Host versus Graft.



Helen, on her third birthday, 6 months post Graft.

Helen is 'developing' This seems a totally inadequate word for what is happening, and yet for those of you who have a Hurlers child, you will know what an enormous achievement this is.

Undoubtedly, the biggest miracle must be her hips. When we were finally discharged in October from WCH we were given a 'good-bye' present of - 'Helen, has dislocated hips'. I just didn't want to know, as you can imagine, but after a few months at home, of course I resigned myself to another long spell in London. This was last August. Helen was prepared for theatre for an exploratory operation to decide how to tackle her hip problem. Within 40 minutes she was returned to the ward with the fantastic news of 'nothing to be done'. I just couldn't believe my ears, there must be some catch, but the dislocation was correcting itself and continues to do so.

The programme of bone marrow transplants as a cure for genetic conditions is a long, laborious, heartbreaking team effort. Helen is part of this programme and I hope and pray there will be many others, but we feel very deeply for the many children who have also been part of it, who are equally important and did not survive, and of those who are too old to be treated. There is hope and the work must carry on.

This morning Helen began her second term at our local infant school, in the nursery section. She adores it and we have great moans when Saturday comes. My most proud and happiest moment was watching her perform as a little pig in the end of term concert with all the other normal children.

Pat Skidmore

We would like to welcome these families to our Society:

Mr and Mrs. Bryan from Tasmania. They have Becky aged 3 years who has Hurler Syndrome.

Mr and Mrs. Baird from West Yorkshire whose youngest sone aged 20 mths has been diagnosed as suffering from one of the MPS diseases.

Mr and Mrs Hartley and their daughter Emma. Emma is 16 months old and has just had bone-marrow transplant for Hurler Syndrome. They come from Cheshire.

Mr and Mrs Stanley. They have four children and one of their sons, Paul is 8 years old and has Sanfilippo Syndrome. They live in Hampshire.

Mr and Mrs Huxted from Oxfordshire. Their 10 year old daughter Rachel has Hurler Syndrome.

Mr Moon from Staffordshire who wrote to me saying that he suffers from Hurler/Scheie.

Mr and Mrs Stenson who have four sons. Their eldest Jamie (6 years) suffers from Hurler Syndrome. They come from Hertfordshire.

Mr and Mrs Peach whose 3 year old daughter Elizabeth has Hunter Syndrome. They come from Warwickshire.

Mr Carr and his son Darren who is 6 and comes from Humberside. Darren suffers from Sanfilippo Syndrome.

Mr and Mrs Ryan from Lancashire. They have just learned that their 2 year old son Paul has Hunter Syndrome.

Mr and Mrs Bam and their daughter Diane from South Africa. Diane has Hurler Syndrome.

Mr and Mrs Borrett from Hampshire. Their daughter Tracey is 2 years old and has Hurler Syndrome.

Mr and Mrs Bradford and their daughter Sally from Nottinghamshire. Sally is 17 years old and has Sanfilippo Syndrome.

Mr and Mrs Broome and their daughers Louise aged 12 years and Julia 14 years. They both suffer from Sanfilippo Syndrome.

Mr and Mrs Duvenhage whose son Johan, aged 2 years has Hurler Syndrome. They live in South West Africa.

Mr and Mrs Hampson and their son Andrew. Andrew is 8 years old and lives in Cheshire. He has Sanfilippo.

Mr and Mrs Headland from Hampshire. They have two daughters, Katie aged 7 and Victoria aged 11 who both suffer from Sanfilippo Syndrome.

Mr and Mrs. Palmer and their son Neil who has Sanfilippo Syndrome. Neil is 12 years old and lives in Derbyshire.

Mr and Mrs Piron who live in South Africa. Their son Patrick is 5 years old and suffers from Hurler Syndrome.

Mr and Mrs Scott from Kent. Their daughter is 20 years old and suffers from Sanfilippo Syndrome.

Mr and Mrs Skidmore and their daughter Helen. Helen is 3 years old and has Hurler Syndrome. She was the first girl to received a bone marrow transplant at the Westminster Children's Hospital for Hurler Syndrome.

Mr and Mrs Soutar and their daughter Christine. Christine is 13 years old, suffers from Sanfilippo Syndrome and comes from Shropshire.

Mr and Mrs Thomas and their son Shaun from South Africa. Shaun is 9 years old and has Hurler Syndrome.

Mr and Mrs Lloyd Towning and their son Mark. Mark is 9 years old and has Sanfilippo Syndrome. They live in Essex.

Anne Trigg and her husband from Kent. Their eldest son, Simon, died some five years ago from Hunters Syndrom aged 7 years. Nevertheless, they feel that they would like to support the Society. Anne has two younger healthy sons.

Mr and Mrs Walker and their daughter Samantha from Lancashire. Samantha is 4 years old and diagnosis is ?Hurler/Sheie or ? Maroteaux-Lamy.

Mr and Mrs Rock from Lancashire. They have two children, Lorraine aged 8 years and Christopher aged 5 years, both of whom have Morquio Syndrome.

Mr and Mrs Bramford and their four daughters. The youngest Toni has Sanfilippo Syndrome. Toni is 5 years old and lives in Wiltshire.

Scott

Our family consists of husband John, Scott born in June 1973, Vicki born in April 1980 and myself, Sheena. Scott has Hurler/Scheie Syndrome, Scott seemed to be normal at birth, and progressed well, he walked at a year, gained weight and was a very contented baby. Speech was slow, his abdomen was distended, and his fontanelle had hardly closed when we finally convinced our GP to refer us to a paediatrician, who told us his fontanelle would take up to 4-5 years to close, and that he was a normal child.

At three years of age we noticed his hearing was getting worse and he was referred to a Hearing Clinic who recommended removal of tonsils and adenoids, and grommets inserted. He was always chesty and his breathing is loud and uneven. At four years of age his eyes started to go cloudy and he was referred to an Eye Specialist and was admitted for investigation. They told us he was partially-sighted and they also did extensive tests and discovered Scott had Hurler/Scheie Syndrome. We were quite shocked as by this time Scott has five years old and was about to start school.

He was Mentally Normal but Partially Sighted and we decided we wanted him to have a chance to try normal primary school, so we enrolled him at the local primary, though the Eye Specialist told us we could not put a child with such poor eyesight in a normal school. The school psychologist supported us as did the school medical officer, and the school eye consultant, so to normal school he went.

Scott is an extrovert loves life and makes friends easily, and loved going to school, and though playgrounds and some schoolwork like writing was not easy because of his sight and his stiff fingers and joints, and he tired easily because of his heart, two years went past and he was still at the same school. Vicki was born after I had an amniocentesis.

We moved to our present house and again Scott started at the local primary school, though by this time his sight was deteriorating, and his fingers and joints were stiffer. He is still there and doing well. He went through a spell of wanting to be like other children as he was conscious of his height (he has not grown in four years) and he wanted to see like his friends, but with time and patience he has come to terms with these things.

He is so stiff now he cannot raise his arms any more, and his fingers are stiff, especially his thumb, which is completely turned in, and makes his life a little bit harder. Scott had an Amniotic Implant in March 1982 at Yorkhill Sick Children's Hospital, Glasgow, and at first his fingers were more supple and tests showed signs of working but after 4 weeks everything was at it was before. We were asked again to consider another implant by injection this time in his chest at both sides, unlike the first which was in his abdomen. So in June 1982 we went to Glasgow for the second implant and once again blood tests showed signs of it working but urine tests showed nothing. He is still having blood tests hoping for signs of improvement, though we have noticed his appetite improving and there has been no more deterioration over the past nine months.

Scott plays football at school and goes horse riding and loves television. He won a local Lego building competition, all despite his disabilities and is a happy affectionate child.

Sheena McCarthy

Dear Readers,

My name is Jean Towning and my husband is Lloyd. We are the parents of Paul who is 12 years old, and Mark who is 9. Mark has Sanfilippo Syndrome Type A. He is a very happy little boy with mischief in his big brown eyes. He is also deaf, but with his hearing aid he can hear us and can say 'Mum', but that is all now. Mark can stand with help and walk about four steps, but like Pat Kirkman we have faced many problems, accepted the challenge and find Mark so loveable and very special. We still hope that perhaps something might help him even now.

We are going to start fund-raising as well, and are certain that if we all pull together we could do some good. Please accept £4.00 to help with the administration.

Jean Towning.

Dear Friends,

Recent research at the Westminster Children's Hospital has suggested that MPS children may be helped towards a healthy life through Bone Marrow Transplantation.

As you may know, most of those children already treated by this method are progressing well, but it is early days. Their progress is encouraging to all of us. However, as the Westminster Unit is the only Unit actually pioneering this treatment, it goes without saying that this single unit cannot cope with all of our children.

We have been in touch with the necessary medical teams in the North West with a view to a BMT Centre here. It would appear that these units are currently under consideration in Manchester and Liverpool. In fact several children with various diseases have already been transplanted, although special units do not yet exist.

One hospital we have contacted has expressed their desire to do BMT's but would only consider adult leukaemics. While we agree all are important we have emphasised our MPS children's needs.

We would like to pass on some advice given to us by the consultants to enable MPS parents to secure a future for their children. Parents must endeavour to inform their Regional Medical Officer of their child's condition and the treatment necessary, that the treatment is only available, at present, in London and that waiting lists are far too long. We would also hope that parents of MPS children who have sadly missed their opportunity would also contact their Regional Medical Officer.

Pressure is being brought by the consultants themselves for BMT Units but extra pressure from parents could well tip the balance in our favour and so help children in the future.

Mary Gardiner

Dear Readers,

After having three normal healthy chidlren, I began to suspect things were wrong with Toni as she didn't seem to respond to the things normal babies do.

It all started when Toni was three weeks old, it took 2 hours to give her 1½ ozs. of milk. No sooner had she taken it that she vomited violently. The next day she was admitted to hospital for tests, the results of which were put down to a chest infection, only to find out at 2½ years old, when admitted for tonsils and adenoids that she had bronchitis at 3 weeks old.

When Toni was three and a half (she was talking up to the age of three and learning to be potty trained) she messed herself and daubed it everywhere and started to eat it. It was only then that we realised something must be wrong, and I admitted to the health visitor that I could have killed her. The health visitor came to visit us and saw Toni as she really was. The health visitor got me an appointment at the hospital and here we were back for tests. Soon after (Toni being just four years old) I had a telephone call asking us to return to the hospital and bring along a sample of urine; we were told that the blood samples they had taken were normal.. Nevertheless we were sent off to Great Ormond Street Hospital fo Sick Children to see Dr. Rosemary Stephens who told us straight and to the point that Toni was suffering from Sanfilippo Syndrome.

It came as a terrible blow and it was difficult to accept that Toni wasn't going to live long and end up being bedridden, incontinent and totally dependant on us. But the worst trauma so far began on 15th February this year when Toni suffered a severe fit lasting nearly 3 hours. This happened whilst Toni was at school. Now she is on Epilim Valergran at night and codeine phosphate to help her loose bowels. We have been told the fits don't normally start until eight or nine years of age but for Toni she had her first fit at three although very mild.

Toni goes to Tadworth Court Hospital 3 or 4 times a year just to give us a break.

I hope this information is of interest to the members of the Society

Sue Bramford.

We wish to extend our deepest sympathies to the families of the following children. Our thoughts are with them.

Simon Kirkman
Treena Silvey
Mark Farrington
Rachel Huxted.

We wish to thank the following people for their generous donations:

Mr. & Mrs. Soutar
Mr. & Mrs. Scott
Mrs. Liz Payne
Mrs. Masters
Mr. & Mrs. Towning
Fran (Middlesex Hosp)
Mrs. E. Perfect
Mr. & Mrs. Headland
Mr. & Mrs. G. Southwood
Mr. & Mrs. D. Russell
Mr. & Mrs. C. Ackle
Mr. & Mrs. C. Winsborrow
Mr. & Mrs. E. Clifford
P.C. E. Raison
Mrs. M.A. Jeffrey
Edie & Ron Gardiner
Jenny & Roger Broome
Mrs. Q. Beecham
The Companions Club of Worthing
The teachers and pupils of St. Symphonious Church Sunday School.

Our thanks also go to:

The Amersham Ladies' Circle for sponsoring a thousand copies of our Information Sheet.

Heritage House Special School, Chesham, Bucks., for kindly donating the proceeds from their Harvest Festival Celebration.

Our thanks in the last newletter to the Amersham Lions should have read Amersham and Chesham Lions. We also owe thanks to the Amersham and Chesham Lions for meeting the cost of the last newsletter and enabling more of the money we have raised to go into the research fund.

We wish to thank the family and friends of Mrs. Vida Windsor for their kind donations in memory of the late Mr. John Windsor, Grandfather of Gary Windsor who has Hurler Syndrome.

The Diplomatic Service Wives Association, Tokyo Branch for kindly donating the proceeds from a fashion show held at the British Embassy.

Eight Medical Secretaries from Worthing Hospital who weight-watched in aid of the Society.

Sue Bramford donated her cash prize for having her letter about the Society published in the "Liveliest letter" column of the Sun Newspaper.

Lyn and John Windsor, Richard and Audrey Hodges, and Jean and Lloyd Towning who have worked very hard to fund-raise.

The Tuesday Toddler Group, Worthing, for their very generous donation as the result of a successful raffle and Christmas sale.

Mrs. Roberts and her friends from Amersham Quakers who so kindly gave a coffee morning. Also Mrs. Jill Williams who kindly donated a beautiful dried flower arrangement for the raffle.

Mrs. S. Marshall-Taylor of Little Chalfont who donated the proceeds from a Tupperware morning.

De Lisle Middle School Loughborough who made a generous donation from their charity fund.

The Mums and children at Rose Wilmot Toddler Group, Worthing, who donated the proceeds from their Nativity Play.

Our thanks also go to:

The Sainsbury Singers who held a Christmas Party for their members and donated their profits to the Society.

Jean Silvey and Glen who held a raffle.

Jane Palmer aged 10 and her friend Alison aged 11 held a fun afternoon for children and raised £7.00.

Mr and Mrs Palmer who held a Christmas raffle.

THE WINDSOR'S

My husband John and I (Lin) have two sons, Stephen who is three years old, and Gary aged fourteen months. Seven months ago we were told that Gary has Hurler Syndrome. When the nature of the disease was explained to us we just couldn't believe it. To us it was a nightmare. About three months ago Gary had an amniotic implant at Guy's Hospital; at first he didn't show any signs of producing the enzymes, but a few weeks ago we were told that he is producing the enzyme. Nevertheless John and I want Gary to have a Bone Marrow Transplant, but like everything else this takes time.

Recently, we decided to help fund raise for the Society, so we got in touch with our local newspaper and they printed our story. It is very hard to fund raise when there is only you in that area; even so I have organised a giant raffle and tombola at my local toddler group and that looks like being a great success. To do anything bigger needs such a lot of organising, not easy with two young children and numerous visits to hospitals. As a result of the newspaper article the Society for Mucopolysaccharide Diseases got good publicity and we received numerous offers of help from local organisations and donations in excess of £250.00

I recorded 'A chance in a lifetime' and my health visitor is bringing another health visitor to watch it. Well, I feel the more the medical profession know about MPS the quicker other children might be detected.

Anyway keep battling on.

Lin and John.

Half a penny is better than none

How about getting your local pubs to collect ½p pieces in jam jars on the bar? This can be extended to shops as well, and would be a fun idea for children to collect thoselittle annoying coins. You'll be amazed at how quickly they add up.