



Niemann-Pick News

The Newsletter of The Niemann- Pick Disease Group (UK)

Autumn 2006

Volume 13 No 3



Roger Franklin completes the Scafell Pike Challenge (page 19)

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Chairman's Chat



This newsletter has a lot about research in it; the content of which is of course, of interest to many of you. However, the fact that so much is going on is of interest to everybody who has the desire to see progress in the fight against this group of diseases – it provides us all with hope.

News too of the wonderful efforts many of you have made when it comes to fundraising – organising events large and small – we can't all be involved in the science, but we can make a difference. We are, in the NPDG (UK) and all the other support groups around the world, involved in making a difference. In this newsletter you will see articles about this year's Annual Family Conference, at which the theme was 'Forging New Relationships'. The more we can work together on these new relationships, families, scientists, doctors and support groups, the faster we will make progress.

Speaking of which, this newsletter contains a request for feedback from families and supporters. Please, if you possibly can, take a few moments to forge a new relationship and fill it in.

I hope you enjoy the newsletter.

Jim

Jim Green
Chairman NPDG (UK)

Research News

Ara Parseghian Medical Research Foundation

NP-C Natural History Study

National Institutes of Health – Bethesda, Maryland, USA.

Glen Shepherd writes: We are pleased to announce that a Natural History Study of Niemann-Pick Type C patients began at the National Institutes of Health (NIH) in July 2006. We have been working with Dr. Denny Porter as well as Dr. Bill Pavan, who serves on our Scientific Advisory Board, for more than a year to help launch this important study.

Following, please find a brief summary of the NP-C Natural History Study for families who may be willing to consider enrolling their son or daughter in the study. For families whose NP-C children may qualify for the Study, we urge you to consider taking part. The information gathered from this study will be utilized in many important ways to help everyone afflicted with NP-C.

The study IS open to all NP-C patients, including those from the UK, with the caveat that NIH can only cover expenses once the patient and a parent get to a port of entry: i.e. they can't pay for the flight to and from the UK, but will pay for internal flights in the USA.

General Information on the Study:

The NP-C Natural History Study will capture much-needed clinical information to determine markers that will help track the disease process for drug trials, provide clinicians working with NP-C patients across the country with better information to care for their patients and will also help with research.

1. All NP-C patients from the US will be considered for inclusion
2. Potentially NP-C patients from outside the US may be considered depending on the response from American families. If a patient from outside the US is enrolled in the study, the family would need to pay for their flight to a port city in the US. The NIH would cover all expenses from that point on. If patients from outside the US are needed to complete the cohort of 20-25 NP-C patients desired to be in the study, there is a possibility funding might be found to cover flight expenses for patients from outside the US.
3. Study began in July 2006

4. 20-25 NP-C patients will eventually be involved. As of September 18, 12-14 patients have been enrolled.

5. Patient and one parent/guardian will visit NIH twice a year for ongoing study

6. Expenses paid by NIH – roundtrip flights or equivalent cost of ground transportation for US patients, accommodations, and daily food stipend

7. Most families and children will stay at the new Children's Inn which is directly across the street from the new Paediatric Clinic and offers beautiful sleeping rooms as well as community kitchens, living rooms, game rooms, libraries, etc.

8. NIH cannot pay for lost wages while parent is away from job, but can write a letter to encourage employers to co-operate with NP-C parents

9. All travel logistics will be handled by the NIH, length of stay about 4 days

10. The NP-C patient will be involved with a number of tests including audiology, swallowing, eye movement, fine motor, MRI, blood and urine samples will be taken, and others.

11. As with any clinical study, there could be certain risks involved. Please contact Dr. Porter and/or Nicole Yanjanin, RN at the NIH for any medical and scientific questions you may have about the study, and to indicate your possible interest in having your child(ren) participate.

NIH Contacts:

1. Nicole Yanjanin, RN (301) 594-1765, nyanjanin@mail.nih.gov
2. Forbes "Denny" Porter, MD, Ph.D., (301) 435-4432, fdporter@mail.nih.gov

Thank you.



Glen Shepherd

Any UK families considering taking part in this study are urged to consult with Jackie Imrie at the Willink for a full explanation of the tests involved. Jackie can be contacted on 0161 922 2414 or by email jackie.imrie@cmmc.nhs.uk

Update from Genzyme.

ASM Deficiency (Niemann-Pick Disease, Type B) Phase 1 Trial

As we end the summer, Genzyme and our collaborators, Drs. Desnick, McGovern, Wasserstein, and Schuchman at Mt. Sinai School of Medicine (MSSM) have reached an important milestone in the development of recombinant human sphingomyelinase (rhASM) for treating ASM Deficiency (Niemann-Pick Disease, Type B). Everything is in place to begin the first ever clinical trial of rhASM. A multi-day training session for all of the study personnel at MSSM is scheduled for the end of September. This session will go over all of the operational details of the trial with everyone who will be involved in its conduct; both those involved directly with the trial participants as well as those who will be working behind the scenes to collect and input the data. Patient screening will commence shortly after the training session and treatment will follow once appropriate patients are identified.

The Phase 1 clinical trial will be conducted entirely at MSSM. The primary objective of the study is to assess the safety of rhASM when administered once to adults with ASM deficiency. Individuals who, based on their medical records, are potentially eligible for the trial will be contacted by Dr. McGovern or her staff to determine their interest in participating. Those who choose to do so will be screened at MSSM to determine whether they meet all of the eligibility criteria for the study. Persons who meet these criteria and agree to participate will be given additional information about the study. Additional information is available at www.clinicaltrials.gov

Different groups of patients will receive a single administration of different doses of enzyme starting with



the lowest dose and proceeding to the highest. This type of study is called a sequential dose escalation study because the data from each group of patients must be analysed before deciding whether to proceed to the next. The decision about whether to proceed to the next dose level will be based on pre-specified criteria. Under the best of circumstances, it is expected to take at least 9 months to enrol all of the patients into this study and several months after that to collect and analyse all of the data. The data from this study would then be used to design future studies that will measure the effectiveness of the investigational drug in different groups of patients. Such efficacy studies will take several years to complete and must be submitted to regulatory authorities around the world as part of comprehensive drug registration package.

Genzyme is looking forward to the participation of patients in this next crucial step in the development of rhASM.

Paul L. Kaplan, Ph.D., M.B.A.

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The Changes in NHS Funding of the treatment of Lysosomal Storage Disorders

Geoff H Apperley, a Commissioning Manager at Addenbrooke's Hospital, explains the changes in funding for the treatment of Gaucher's disease and other lysosomal storage diseases since April 2005:

'The centralised funding of treatment for lysosomal storage diseases such as Gaucher's disease, which was introduced in April 2005 by the Dept of Health's National Specialist Commissioning Advisory Group (NSCAG), has dramatically reduced the time between confirmed disease diagnosis and delivery of effective treatment for initially Gaucher's and Fabry's and most recently, Pompe's patients. Now what used to take many months, in most cases, has been reduced to a single notification to NSCAG of the start of therapy. In

my opinion, the shift in funding has been a success and nothing reflects this more than the benefits this has had on improving access for patients to effective treatments.'

'I have been a NHS Commissioning Manager at Cambridge University Hospitals NHS Foundation trust (Addenbrooke's Hospital) since 2000 and am responsible for managing agreements between the Trust and commissioners such as primary care organisations, the independent sector and agencies such as NSCAG. I have been particularly involved in obtaining funding for the treatment of lysosomal storage disorder diseases since 2001.'

A "Lottery"?

'Before 2005 NSCAG designated and funded the services of four Gaucher's Centres to provide diagnosis and management of Gaucher's disease alone. Treatment, such as enzyme replacement therapy, was prescribed through the patient's local GP, hospital consultant or clinician at a designated Centre. Payment for their treatment came from their own local Primary Care Trusts or from a Consortia (group) of Primary Care Trusts. This meant that funding decisions on high cost treatments were subject to the 'lottery' of separate local decisions. If at first funding was not granted, the decision was often challenged by the patient's doctor and family with the support of the relevant patient advocacy group. Much time and many letters were expended in seeking funding to allow treatment to start not to mention pressure on patients and their families.'

Shift in Funding Responsibility

'On 1 April 2005 the 'treatment landscape' changed with a radical shift in the funding responsibility in England centrally to NSCAG which began to fund both services and treatment on a national scale through six designated centres (listed below). Treatment funding is obtained by NSCAG by top-slicing PCT allocations and reimbursement to the designated centre. In addition, the coverage of these centres was extended to all lysosomal storage disorders and currently provides funding for Gaucher's, Fabry, MPS1 and Pompe's treatments with therapies for other disorders such as Niemann-Pick, Sandhoff and Hunter's under review.'

'The most important reason for the change was the inconsistent funding of treatments and consequent inequity of access. The Department of Health recognized these inconsistencies and the additional funding pressure placed on local trusts by the increasing burden of rare diseases that require high-cost drugs.'

Six Centres

'The six designated centres diagnose, manage, prescribe and deliver treatment to all existing and new patients. They are listed below with A for Adult and P for Paediatric coverage:

- Addenbrooke's Hospital: A/P
- Central Manchester and Manchester Children's Hospital: P

- Hope Hospital, Manchester: A
- Great Ormond Street Hospital, London: P
- University College Hospital, London: A
- The Royal Free Hospital, London: A.

'Adolescents are managed through a partnership between adult and paediatric centres. Other metabolic centres such as at Birmingham are applying for NSCAG designation in 2007-08.'

Treatments

'The funding available covers enzyme replacement therapy and the newer oral substrate reduction therapy. Only the above designated centres are allowed to prescribe these treatments. Patients can choose which centre to attend subject to referral by their GP.'

'The change has simplified administration at centres so that decisions regarding a change in dose or the commencement of treatment for a newly diagnosed patient, who meets the agreed protocol and criteria for Gaucher's, Fabry's, MPS1 or Pompe's disease can be made directly by the designated centre clinician and notified to NSCAG. It also means that decisions regarding new therapies and off-label use of existing therapies can be assessed by a Lysosomal Storage Diseases Advisory Group of Clinicians and NSCAG at a national level.'

Differences in the UK

'Although there is one National Health Service, differences apply within the UK. The funding mechanism described has only been adopted in England and there are other funding arrangements in Scotland, Wales and Northern Ireland.'

'NSCAG already has an arrangement whereby Scottish patients can be seen in the designated English centres for diagnosis and management. But the decisions on funding treatment still remain with the patient's Local Health Board and is obtained through the National Services Division in Edinburgh.'

'Welsh and Northern Irish patients can be seen in designated English centres for diagnosis and management, although costs for this access and treatment costs are funded through the Health Commission Wales and the Northern Ireland Health and Social Security Boards which take the respective decisions on treatment funding.'

'European Economic Area and overseas patients can

be seen for diagnosis in the designated centres under an E112 arrangement where there is a reciprocal bilateral health agreement or as private patients. However treatment costs are the responsibility of the patient's country of origin.

'The new NSCAG centrally supported system offers improved and speedier access to appropriate existing treatment for English patients and better support for designated centres throughout England.

The future

This initial agreement supporting this model of treatment provision was due to expire in March 2007 however we have been recently informed by NSCAG of its continuation from April 2007 to March 2008. I hope that this new "central" model continues beyond 2008 and will be adopted throughout the UK. It is effective, responsive and delivers patient treatment in

a timely manner. There remains issues of cost but, the use of agreed protocols and criteria for treatment in the centres, ensures cost-effective use of NHS funding in a challenging high-cost environment. For me as a commissioning manager in a designated centre, the most radical changes have been the delivery of equity of access with significant reductions in time to start effective and licensed treatment of patients by my clinical colleagues. This has resulted in patient benefits of equity of access and decreased anxiety together with a dramatic saving of my time in letters, telephone calls and follow up.'

This article first appeared in the June 2006 issue of Gauchers News. Thanks go to Geoff H. Apperley who kindly updated the article and gave permission for us to reproduce it.

The Peter Carlton Jones Memorial Award 2006 – Winner

The winner of this years Peter Carlton Jones Memorial Award was Emyr-Lloyd-Evans, who collected his Award at NPDG (UK) Family Conference from Peter's widow, Lesley Carlton Jones. An abstract of his winning submission follows:

Lay summary: We have discovered that NPC1 lysosomes have less calcium than normal lysosomes. This lysosomal calcium problem occurs as an early step in the disease process leading to the lipid accumulation, including the build up of cholesterol and glycolipids. If we compensate for the calcium problem in the NPC1 mouse by treating it with a drug that alters calcium levels the mice survive longer and the disease process is slowed. This research suggests that treating the calcium problem in NPC1 may offer a new route to the development of an effective therapy.

Defective late endosomal/lysosomal calcium homeostasis is an early event and a potential therapeutic intervention point in Niemann-Pick C1 disease pathogenesis.

*Emyr Lloyd-Evans, Dan Sillence and Fran Platt.
Department of Pharmacology, University of Oxford,
South Parks Rd., Oxford, OX1 3QT.*



Niemann-Pick type C1 (NPC1) is an autosomal recessive neurodegenerative lysosomal storage disorder. NPC1 is caused by mutations in a late endosomal/lysosomal protein (NPC1) of unknown function leading to accumulation of glycosphingolipids, sphingosine, sphingomyelin and cholesterol in these compartments. How mutations in the NPC1 protein leads to lipid storage and how lipid storage leads to neuropathology remains unclear.

Some of the sphingolipids stored in NPC1 cells and tissues have been reported to alter endoplasmic reticulum (ER) calcium homeostasis in primary sphingolipid storage disorders, contributing to neuropathology (Gaucher, Sandhoff and Niemann-Pick A). In order to see whether NPC1 cells share common calcium induced neuropathological mechanisms with the primary sphingolipidoses we analyzed the calcium content of intracellular NPC1 organelles using the intracellular fluorimetric probe Fura-2AM.

We have discovered that NPC1 null cells have a decrease in total cellular calcium. Calcium uptake and release from the ER, the largest calcium store in the cell, is normal. Mitochondrial calcium content is also normal, indicating that NPC1 mitochondria can correctly sequester calcium despite a reported defect in their membrane potential. We discovered that the defect is caused by a large reduction (~70-80%) in the late endosomal/lysosomal acidic calcium pool. This novel finding was confirmed using multiple pharmacological agents to specifically empty those stores.

In order to discover the relevance of this finding to the disease process we used secondary amines (U18666A, sphingosine) and siRNA against NPC1, to induce the NPC1 phenotype in control RAW macrophage cells, and recorded the events that occurred over a 48h time course. Perturbation of late endosomal/ lysosomal calcium is the first event that we detect following amine treatment. This is followed by abnormal transport of a fluorescent glyco-sphingolipid, BODIPY-lactosylceramide, to late endosomes, with cholesterol and glycosphingolipid storage occurring last in this sequence of events. Silencing of the NPC1 gene leads to the same chronology, indicating the validity of using secondary amines to mimic early events in NPC1 pathogenesis.

To validate the possibility that late endosomal/ lysosomal calcium depletion is an early event in NPC1 pathogenesis we treated control RAW macrophages with BAPTA-dextran (a plasma membrane impermeable calcium chelator that can then be internalized by endocytosis and chased into different endocytic compartments) to chelate calcium in the late endosomal/lysosomal system. Chelation of calcium in these compartments resulted in mistrafficking of sphingolipids and storage of free cholesterol, indicative of NPC1. As depletion of calcium could induce an NPC1 phenotype we determined whether increasing

cytosolic calcium by treating NPC1 cells with calcium modulating agents (calcitriol, thapsigargin, curcumin) could reverse the cellular abnormalities. All three reagents were able to reverse the endosomal transport defects leading to a reduction in lipid storage back to control levels. We are currently testing in the NPC1 mouse model the efficacy of curcumin, a natural product that is tolerated at high doses. Treatment with curcumin leads to an elevation in cytosolic calcium that corrects defective vesicular fusion and transport at the level of the late endosome (potentially caused by reduced calcium efflux out of the organelle). Early results indicate improved functionality and increased life expectancy in the curcumin treated NPC1 mouse.

This study indicates that abnormal late endosomal/ lysosomal calcium in NPC1 cells is an initiating factor in pathogenesis and a target for therapeutic intervention with drugs that modulate calcium homeostasis.

[Supported by the Ara Parseghian Medical Research Foundation]



Niemann-Pick Disease

The Peter Carlton Jones Memorial Award 2006/7

Up to £1000.00

It is proposed that an annual award of up to one thousand pounds (£) will be made to an individual who is engaged in either research or teaching or treatment or care, within the public or private sectors in the United Kingdom.

The award to be granted in response to the submission of a 'research project' (not a literature review) which provides an original contribution to the scientific or public understanding of the Niemann-Pick Diseases and / or their treatment or cure.

The applicant's submission of a completed project should be in the form of an abstract, of one side of A4 paper (~ 600 words), which includes clear statements on the work undertaken, methodology, 'findings' and principal features in regard to the nature of the project, as referred to above.

The decision on the award will be determined by an Award Committee of the Niemann-Pick Disease Group (UK). The Committee comprising; the Group's Chairman, the Medical Advisor, the Trustee with a research remit and the Clinical Nurse Specialist (Niemann-Pick Disease).

Applicants are invited to submit for the 2006/7 Award, with a view to the Award Committee short listing and possibly seeking further information and / or a project report, prior to a final decision being made.

Submissions to: Toni Mathieson, National Development Manager
Niemann-Pick Disease Group (UK)
11 Greenwood Close, The Pastures, Fatfield
Washington, NE38 8LR
E-mail: niemann-pick@zetnet.co.uk
Tel: 0191 415 0693

Closing date for submissions: 28th February 2007

It is envisaged that the successful applicant will be invited to the Annual Family Conference of the Niemann-Pick Disease Group (UK), to be held Summer 2007, to formally receive the award.

We would like to thank all those who submitted to the 2005/6 Award.

The award has been established in memory of Peter Carlton Jones, a Trustee of the Group, who died suddenly while attending the 2004 Family Conference in Telford.

Niemann-Pick Disease Type C in Yarmouth County, Nova Scotia.

By Dr. Wenda Greer

Dr. Wenda Greer, PhD, FCCMG, is Associate Professor of Molecular Biology, at Dalhousie University, Canada.

Founder effects resulting in an uncommonly high frequency of a mutant gene are often noted in isolated rural populations. One well studied example is Niemann-Pick C disease in Yarmouth County, Nova Scotia. Early work by Winsor and Welch (1978) described 15 patients, all of who lived within a 10 mile radius of one another and whose ancestry could be traced back to 3 couples who were born in Nova Scotia in the late 1600's. Due to the high carrier frequency, many in this community are at high risk of having an affected child, and until recently, there was no reliable test to distinguish carriers from non carriers. Fortunately, through collaborative efforts of my laboratory with others at Dalhousie University, the genetic defect responsible for disease in these patients was identified and a simple blood test was developed to identify the mutation. Because of their common ancestry, the mutation is expected to be identical in all patients and carriers; therefore the development of a carrier test was straightforward. With this test, it was possible for individuals to make informed decisions with regard to family planning and reproductive choice. In the absence of a cure for this devastating disease, carrier testing is one avenue to prevention.

Because of the extensive personal contact with approximately 200 first degree relatives of patients and the newspaper and radio coverage of our work at the time, we perceived that the community was well informed. We met with the family twice to describe the objectives and results of our research, a letter describing the availability of a carrier test was sent to all participants and family physicians in the region, and a genetic counselling session was provided to approximately 50 interested parties where results and implications of individual carrier testing was explained. We anticipated a huge number of referrals for carrier testing, but it didn't happen.

Because of this lack of response to the availability of a NPC carrier test, a study was conducted by a graduate student, Tracey Russnak-Redden, to assess factors influencing the acceptance and utilization of genetic testing in this community. A questionnaire was distributed to a random sample of the general population of Yarmouth County (450 surveys mailed, 128 completed and returned). It was found that one third of the subjects expressed an interest in determining their carrier status, but only 9.5% intended to be tested, and only half of these intended to do so within six months. No one came forward for testing as a result of participating in the study. Only approximately half of subjects had heard of NPC before receiving study materials, and only 21% knew that a genetic test was available to detect carriers. Most felt that their risk was low so would not be tested and those past child bearing years felt that there was no need for them to be tested. The participants agreed that the community should be well informed about the availability of the test, that the test should be voluntary and that it should be made available locally such that they would not need travel 3 hours to Halifax. From this study, it is clear that there is a need for more education in the Yarmouth region in order to increase the awareness of NPC as a genetic disorder and the availability of a genetic test. Our centre has recently acquired several additional genetic counsellors which should enable the development of an education program that will allow members of the community to make informed decisions regarding NPD genetic testing.

Despite the disappointing lack of awareness of NPC and apparent lack of interest in genetic testing in Yarmouth County, each year our centre receives a number of very appropriate referrals of young couples with a family history of disease and who are considering pregnancy. Thankfully, this seems to be having an impact because no-one in Nova Scotia is currently affected with NPC.

NPDGUK Calendar Competition

Thank you to all who sent in pictures for the calendar competition.

The pictures were all fantastic and we hope the finished product will be available soon...

Research Report by Bill Owen, Research Co-ordinator

Family Trees

These Newsletters seem to come around very quickly and as usual I am scrambling to meet the publication deadline and trying to recall the various research events of interest that I have had an involvement in since the Summer Newsletter. One day I will get organised! In my last article I suggested that families could make a contribution to disease prevention by data gathering in the form of constructing a family tree which is not necessarily a simple matter as I have discovered. With both my parents dead and my wife's mother pretty hard of hearing, she is 94, we have no option but to fall back on paper information which seems to have the ability to disappear without trace. The other option is to attempt to trace long forgotten relatives and have the problem of explaining what you are doing, particularly with regard to the disease. The message is don't leave it too late! It is worth remembering when looking through old family records to note any instances of premature deaths. Whilst these cannot be evidentially linked to Niemann-Pick, and it is unlikely that Niemann-Pick was the cause, such events are additional data for the archive.

Charity Research Involvement

Earlier in the year we held an additional meeting of the Board of Trustees to review our activities and to determine if there were other ways in which we could improve the way we work, provide greater assistance to families and improve our involvement with research into the Niemann-Pick Diseases. In this latter area we have become more active in 2005/6 although not at the expense of the charity core management functions and the Family Support Service. Our ability to fund research is limited by our funding capability and, we are very much aware that conducting research is costly. We have however been able to make modest funding grants for some interesting research projects and I provide a brief update of these below. We are not alone in this respect and in discussions with the US National Niemann-Pick Disease Foundation (NNPDF) we agreed that our most appropriate method of funding research projects for charities such as ourselves, is to provide 'seed corn' grants. These provide researchers with sufficient effort to conduct a pilot study, gather supporting data and make a supported case for a more major study to organisations able to provide greater amounts of funding. In addition, we agreed that when projects are submitted for funding approval, we would use the NNPDF Scientific Advisory Board (SAB) to peer review the proposals, thereby taking advantage of the considerable expertise of the SAB members. This is what we have done.

- NOS2 Regulation – This project was undertaken by

Dr Thomas Kelley at Case Western Reserve University, in Cleveland, Ohio. From initial findings in NOS2 (nitric oxide synthase) regulation between cystic fibrosis and Niemann-Pick Type C it was proposed to study the cell signalling pathways leading to NOS2 reduced expression. The work completed earlier this year and identified changes in proteins Ras and RhoA GTPase which are involved in cell proliferation. This is unsustainable in neurological cells and may influence the disease pathology. A further proposal is being prepared to construct a more neurologically relevant model.

- Substrate Reduction Therapy (SRT) Monitoring – The current trial using miglustat has few endpoints that be objectively measured and Dr Fran Platt at the University of Oxford has, for some time, been working on methods of evaluating the efficacy of miglustat at cellular level. A number of cellular changes were identified as a result of miglustat administration and these were validated in a single patient study undertaken by Dr Robin Lachmann then at the University of Cambridge. Funding for the ongoing work at Oxford is often a problem and because of an impending discontinuity, the work was about to come to a stop. Following discussion with Dr Platt we were able to provide short term funding and, this was subsequently increased by a grant from the NNPDF. These funding actions have allowed this important work to continue and an application for a larger grant has been submitted to a Medical Research Charity.
- Research Nurse – In addition to the provision of the scientific funding, we are also interested in clinical research. Those who were at the Family Conference this year will have met Liz Jacklin who has been appointed to the 2 year Niemann-Pick Research Nurse post, at the Willink Unit, Manchester. Amongst other things, she will be collecting data on liver disease, seizures, effects of pregnancy, psychiatric manifestations in addition to mutation correlation.
- Cellular Calcium Study – A young scientist, Emyr Lloyd-Evans working at the laboratory of Dr Fran Platt, presented work he had submitted under the late Peter Carlton-Jones Award scheme which is in its first year of operation at our Family Conference. This is exciting work which identifies defective calcium homeostasis in NPC sub cellular organelles which in turn is followed by other established defects. We hope that the existence of the award helps to create other exciting papers in the future.

Ara Parseghian Medical Research Foundation

I was able to attend the annual reporting round held by the Foundation in Tucson, Arizona in June and was reminded once again, of the great effort expended by family and friends into raising funds and, to promoting Niemann-Pick research on a broad front. This was reflected in the range of presentations and posters exhibited. It is difficult to take in all that was presented but from my point of view I was pleased to see more of an emphasis on the neurobiology of the disease. The loss of neurons, particularly cerebellar Purkinje neurons is well established and the details of the process leading to cell death, known as apoptosis, has yet to be understood. This is a complicated process involving many signalling proteins but the identification of these together with a method of intervention would be likely to result in cell survival even if neuronal function remained impaired.

An example of the detail that scientists must go to in order to resolve the mechanism of protein action, was illustrated in work presented on the NPC2 protein. This protein defect causes NP disease and thought to act in the same pathway as NPC1. After the discovery of the gene which was also known as HE2, the three dimensional

protein structure was identified and it was thought that this would bind a molecule of cholesterol and was located in lysosomes. It was already known that cholesterol was delivered to lysosomes by LDL particles and, that the cholesterol was separated from other molecules by an enzyme called a hydrolase. But how does the cholesterol get out of the lysosome? This is where NPC2 is thought to operate but it was not clear whether NPC2 could bind cholesterol and what it did then. Although there is some way to go in understanding exactly what happens, it has now been shown that NPC2 is able to transport cholesterol between membranes by what is described as a collisional mechanism. Another step taken?

In another project, studies have shown that NPC1 cells have reduced calcium levels in the late endosomal/lysosomal (E/L) system. Introduction of calcium into the cell cytosol restores a number of defective processes caused by the NPC1 defect.. It is proposed that this additional calcium, corrects transport problems in the E/L system. One of the agents used for this is curcumin, a constituent of tumeric the yellow powder use extensively in Indian cuisine! In work with NPC1 mice, the use of this agent has shown functional benefits and this was the case irrespective of the age when treatment was started.

Research Nurse Update

Well I have been in post for 6 months now and the time has flown. I am really enjoying working with Jackie [Imrie] and being part of the team at the Willink and of the wider Niemann-Pick Team.

The highlight of my first 6 months has to have been the family conference. It was lovely to meet everybody especially the patients and their families. I had a great day at the theme park and can't wait for next year!

My first priority when I took up post was to learn as much as I could about Niemann-Pick Disease and how it affected not only the individuals but also their wider family and friends. I have started to do this and have been grateful for being allowed to visit patients at home with Jackie and also attend meetings at a school with her. This has helped to give me a real insight.

My first task was to complete an audit of all our patients with Niemann-Pick type C disease. This is a piece of work which details the presentation, symptoms and progression of the patients together with any genetic information we have. This work was nearly complete but needed finalising for publication. This has been done and we are waiting for final confirmation of its acceptance for publication in an academic journal.

The Group identified a number of areas which they would like me to concentrate my work on. I have begun with a study entitled "A retrospective study to look at the effects of puberty, menstruation and pregnancy on

Female Patients with Niemann-Pick type C disease". The protocol for this has been written and has received full ethical approval to go ahead. The first letters to relevant patients and their families went out last week. I hope to get this project well and truly under way in the coming months.



I recently attended the INVOLVE (INVOLVE promote public involvement in research) conference which has led to new research ideas which I will be exploring in the coming months. I hope the next six months are even more productive than the first have been and that I too can soon start to make a difference.

Liz

Elizabeth Jacklin RGN

Niemann-Pick Disease Research Nurse

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The post of Clinical Research Nurse is funded by the NPDG (UK) in collaboration with Manchester Royal Children's Hospital.

An update on Genetic Testing and Insurance

By Dave Roberts, NPDG (UK) Trustee

The last article we published on this topic was in the spring of 2004. In that article, Bill Owen, our research co-ordinator, outlined a concern that any person who was the carrier of a recessive gene and who wished to take out a health / life insurance policy would be subject to discrimination.

The underlying issue is the use of genetic testing by the insurance industry as a screening process and as a way of predicting the risk of someone developing a rare, or maybe even a common, disease at sometime in the future.

Those of us with families affected by Niemann-Pick Disease may wish to take a genetic test at any time in the future for a variety of reasons - it may be to test if we are a carrier or we may want to take part in a family exercise to track the gene defect. Whilst we do not know of any health implications ourselves, we may be concerned that an insurance company may take a different view. To date this does not seem to be an issue but, as a Charity Committee, it is worth monitoring the ongoing discussions between the insurance industry, the Government and other interested parties. This article provides a further update.

Putting this into context, the first code of practice covering Genetics and Insurance was introduced by the UK insurance industry in December 1997. In October 2001 the Association of British Insurers in agreement with the Government, announced a moratorium (i.e. a voluntary ban) on access by member companies to applicants' predictive genetic results.

This moratorium was renewed in March 2005 and took into account the views of the Association of British Insurers, its member companies and the Government, the Genetics and Insurance Committee, the Human Genetics Commission, patient groups and other interested parties.

From this review it was agreed to extend the Moratorium by five years to 1 November 2011, and to create a policy and framework that was subject to independent oversight and which provided transparency and fairness in the way that insurers used genetic material. This policy and framework would be reviewed in 2008.

Within the moratorium, the main things you need to know about insurance and genetic tests are that:

- Insurers will not ask you to take genetic tests in

order to get insurance.

- The broad classes of insurance for which genetic testing may be relevant are confined to life insurance, critical illness insurance and income protection insurance.
- Until November 2011, you can apply for a total of up to £500,000 of life insurance, £300,000 of critical illness insurance and £30,000 annual benefit of income protection without having to tell the insurer the results of any predictive genetic test you have previously taken.
- When answering insurers questions, you will only need to tell your insurer the results of a predictive genetic test if you want higher amounts than this (totalled across all your insurance policies), but only where the test has been approved by the Government's Genetics and Insurance Committee as being relevant to the type of insurance you want.
- At this stage, it is important to note that the only test result to be approved by the Genetics and Insurance Committee is for Huntington's disease when applying for Life Insurance.
- If one of your relatives has taken a genetic test, you do not have to tell your insurer when looking at an application form. But if the insurer asks, you will need to tell them about any illnesses that run in your family.

At face value everything looks re-assuring and people should not be put off having genetic tests. However, there are still some serious concerns that need to be addressed. It would appear that there is uncertainty by individual insurance companies about how to adopt the policy on the use of genetic test results and some insurers may still have in mind the "ten tests for seven conditions" which existed before the moratorium was introduced. Indeed, the reason for extending the moratorium, once again, was to provide a breathing space, during which the industry, the Government and other interested groups can discuss and agree on a long-term solution. These discussions are ongoing and the outcome is not clear as yet.

Other issues surround the fact that whilst an insurance company will not ask you to take a genetic test, you must give the insurance company any information you know about any hereditary (genetic) disease that you or your family members have had if it is requested on the application form - insurers can refuse to pay a claim later on if you deliberately don't fill in the application form fully and correctly. Further still, in the

conversations about predictive tests for a genetic disease there appears to be no distinction between late-onset disorders, caused by single genes, such as Huntington's Disease (of great interest to insurance companies) and carriers of a recessive genetic disorder such as Cystic Fibrosis and Niemann-Pick (which should have no interest to the insurance companies).

There is clearly the potential for a complex swirl of emotions, between all of the interested parties, when trying to reach agreement on these matters and things will only become clearer over time. Our interests are represented through membership of the Genetic Interest Group, which is an umbrella charity for genetic diseases. The more vocal Gene Watch UK has called on the Government to introduce legislation to:

- Prevent discrimination by employers and insurers on the basis of genetic test information;

- Prohibit employers and insurance companies from using genetic test results to make employment decisions, deny health coverage or raise insurance premiums;
- Guarantee genetic privacy and the right of people to refuse to undergo genetic testing;
- Ban insurers from requiring genetic tests, from requiring the disclosure of genetic test results, and from using the results of tests to increase insurance premiums or deny coverage.

It may well take five years to reach agreement with all of the competing interests but in the mean time, genetic testing can offer important health benefits for many people and no-one should be put off having a genetic test because they fear discrimination by insurance companies.

Christmas Card Competition

We are pleased to announce the winners of our Christmas Card Competition, judged by attendees at Conference 2006. Thank you to all who entered the competition, all of the pictures we received were wonderful and it was very difficult to choose a winner! A small prize will be on the way to our budding artists very soon....

THE WINNER



The Winner – Annie Pyne, aged 10 – 'Christmas Night'.

The Runners Up – Edward Hurst, aged 3 – 'Snowman';

William Brooks, aged 6 – 'Angel'

THE RUNNERS-UP



The cards are available in packs of 10 for a suggested donation of £3.00 per pack + postage and packing. Please contact the Office for more details on 0191 415 0693 or email niemann-pick@zetnet.co.uk. Alternatively, please complete and return the order form included with this newsletter.

C o n f e r e n c e 2 0 0 7

SAVE THE DATE!!

NPDG (UK) Family Conference 2007

23 / 24 June 2007

Venue to be confirmed

Comments from Conference 2006:

‘Excellent. Very informative – great variety’

‘As a family we really look forward to our weekend away’

‘The venue was very good – easily accessible with spacious accommodation’

‘It was very useful to speak to other families in our situation’

‘The Children’s Programme was excellent and kept them entertained’

Watch this space... more details coming soon...

Fundraising News

Awareness Week 2006

This year we decided to move Awareness Week from October to July, to enable participants to hold more outdoor events, such as a ‘Niemann-Pick-Nic’. This proved to be a popular decision, with many ‘Pick-Nics’ and other events taking place around the country and attracting the attention of local media. Thank you to all who joined in the fun, here are just a few of the events that helped to make Awareness Week 2006 a great success:

Mary Winetroube held a Niemann-Pick-Nic in her garden, she writes:

“Friends of Lucy Mathieson, age 3 NPC, got together on one of the hottest days of the year to raise £380.12 for NPDG (UK) at a Niemann-Pick-Nic. Great fun was had by all from the Sunderland ‘Tiny Talk’ group at the event, which was held at Burdon Village.

Friends and neighbours joined in the fun with the little ones playing on the bouncy castle and in the paddling pool - it was so hot a few of the mums had a paddle too! A big thank you to all who donated the fabulous prizes for the raffle and tombola, and to those who bought and scoffed an enormous amount of coffee and cakes.

We look forward to doing it all again next year!”



Toddlers, Scouts and South Africans join in Balloon Race!

During Niemann-Pick Awareness Week Liz Jacklin organised 3 balloon races. She writes:

"The first started out as a small family gathering, my dad's wife had family over from South Africa and she was hosting a family get together for them. I saw this as a perfect opportunity to have a party with a difference and we sold balloons to all the party guests. We sold 49 balloons and sent the visitors back armed with literature about Niemann-Pick.

On 24th July Denton District Scouts held a Fun Day and Challenge day in the Town's park. We sold balloons at all the groups in the district leading up to the event and on the day. The afternoon was filled with activities, SUMO wrestling (with inflatables!) assault courses, individual challenges. DECATHLON donated a pop up tent and we held a competition to see who could erect the tent in the fastest time. The winner was another scout leader who put up the tent and placed two roll mats inside and lay on them in 30.4 seconds! All proceeds were donated to the Niemann-Pick Group. We ended the day with a Teddy Bears Picnic and then let off 150 balloons. Everybody had a really good time (and in good scout tradition got some badges!)

The last day at nursery for my son Alex and his classmates was marked by Niemann-Pick Day. All the nursery staff and children dressed in yellow. In the nursery entrance was a display explaining about Niemann-Pick with literature for parents to take away. In the afternoon all the children had a teddy Bears picnic with all the food either yellow or teddy bear shaped! At the end of the picnic we let off over 100 balloons to the delight of all the children.

The local press came and featured our events.



The winning balloon was found in Denmark! We have sent a Niemann-Pick T Shirt as a thank you to the person that returned it and the person that bought the winning balloon won a picnic hamper with chocolates and wine, donated by Tesco's and Morrison's.

A huge thank you to everybody that got involved it was great fun and we raised £390!"

A day for Annie

By Sam, Annie's friend.

Having known Annie for a number of years and obviously knowing about her illness, I wanted to do something for her and her family. The idea came to me when my sister and I were talking about Annie, and all of a sudden I knew what I wanted to do. I had an idea buzzing around in my head for another day or two but was not sure whether Kait and Norman (Annie's parents) would like the idea. I approached Kait and she seemed very keen, but would speak to Norman and get back to me. They agreed and by this time my sister had put together a leaflet about what the day would entail.

I was so excited about what the day would bring. The leaflet would briefly let people know that the day was to raise money for Niemann-Pick Disease and also that I knew of someone who is suffering from it. Breakfast would be provided for £2 which would include Tea/Coffee, croissants, strawberries and orange juice. Also there would be homemade cakes for people to buy or take home for a small charge. My sister Marie would provide leg and feet massaging for £5 and all the proceeds would go to the cause. I had other friends that had card and jewellery stalls, quilts from abroad that were handmade and also a raffle and a bring and buy table. All of my friends and my sister donated items for these stalls as well as their time and effort. They will never know how grateful that I am to them!

I only had about 3 days to put it all together and there were times that I thought it was not going to happen, but because I have great friends and a fab sister, it all



worked out really well and everyone realised that the cause was very dear to my heart. People started to arrive shortly after 10am the tea urn was ready, the oven was on for the croissants. It was a really hot day, we had two gazebo's in the garden, one for the children to play under and the other for the bring and buy tables. The garden was full of people browsing around or eating and it was a very happy atmosphere - it was a really fun day. The raffle went very well, lots of people went home with prizes and then we started to clear up. We all had a sense of achievement by this time and an idea of what we had raised, but never expected to raise over £500!!!! I am so chuffed that I and lots of people dear to me helped to raise this huge sum of money for an extremely worth while cause!

Generous Gestures...

Michelle Maguire requested donations instead of presents at her 40th birthday party, charging guests to attend and holding a raffle and a tombola. Michelle also laid on entertainment in the form of a comedian, ensuring a good night was had by all and raising over £300!!

Margarita Rutson also requested donations instead of presents at her 40th birthday party and her husband John, along with children Thomas and Madison, competed in the Green Park Challenge as part of Reading's half marathon. The two events raised awareness of NPD and a fantastic £175!



Coffee Morning

By Rose and Ron Thompson



We are a small community, consisting of three blocks of flats. On hearing about Lucy Mathieson age 3 who suffers from Niemann-Pick Disease Type C, we felt we could do something to raise funds.

As myself and Sylvia run a small community room, we thought this the ideal venue. We printed out leaflets and put posters up in each block. All the people who attend the community room brought all kinds of things along and we were able to have a giant tombola, raffle and bric-a-brac stall. One lady gave a beautiful doll and we had a 'Guess Her Name' competition.

Sylvia baked a mountain of pies and cakes. We also had tea, coffee and home made scones (I made them). We had a great morning. Even before the event people were giving money – they wanted to help in any way possible.

At the end of the morning, I could not believe we had raised the fantastic amount of £522.17 – we are looking forward to our next fundraising day. Great big thanks to everyone.

Rebecca Walters held a Niemann-Pick-Nic in her garden, she writes;

"With the weather being so nice we decided to hold a Niemann-Pick-Nic in our back garden. Then what should happen on the morning of Saturday 22nd July – the heavens opened!!! We still went ahead with it though and a fab day was had by all. My dad put three gazebos up in the garden to shield the guests from the wonderful British weather. We invited mostly friends and family and charged them a £5 entrance fee, this included food and drink which had all been donated.

There was a fancy dress competition, a raffle and tombola. We were given 30 2006 World Cup memorabilia pint glasses and asked people to make a

donation for them. We also had a 'Guess the Weight of the Cake' competition and the balloon race.

Unfortunately, the balloon race was a bit disastrous – once the luggage labels were attached the balloons didn't fly!!! But after raising £432 in total – what the heck!!! Everyone was very understanding about the whole thing – thank Goodness!!

Well I think that's it by the way Captain Jack Sparrow and the Umpa Lumpa won the fancy dress competition!"



Dianne Lish and Julia Gothard organised a 'Niemann-Pick-Nic' at their local church in Sunderland. The Children enjoyed many games themed around teddy bears, with a bouncy castle and a magician, as well as a delicious picnic served outside due to the fantastic weather! The day raised over £200!

Tony Jellings writes:

On 5th May at St George's Church in Kemp Town, Brighton, two outstanding young musicians gave a charity concert in aid of the Niemann-Pick Disease Group (UK) – sixteen year old clarinettist Julian Bliss and award winning pianist Julien Quentin.

The large audience was treated to a superb performance by these two virtuosos who regularly give recitals all over the world. It is the second time that Julian Bliss has given a concert for the NPDG (UK) and once again he "wowed" the audience with his amazing talent.

On 19th July, Julian played with the BBC Symphony Orchestra at the Royal Albert Hall in a Promenade Concert celebrating Her Majesty the Queen's 80th birthday. It is only the second time the Queen has attended a Promenade Concert during her long reign and therefore this engagement was a great honour for Julian.

Many in the audience for our concert on 5th May commented to me that they had been privileged to witness the performance of Julian and Julien and were proud to be able to say 'I was there'!

The support of these two young men brings nothing but credit and increased awareness among the public to



The Aronowitz Ensemble

our charity and helps to draw attention to the need for public support. The concert, which was sponsored by Sotheby's of Bond Street, London, raised a sum of £2,307 for the NPDG (UK).

Much of what I have written about the 5th May concert applies equally to the second concert this year in aid of our charity, held on 14th June in St John's Church, Wimbledon, featuring

the immensely talented Aronowitz Ensemble. The ensemble consists of seven award winning musicians who gave a stunning performance to a large, enthusiastic and appreciative audience. Among the musicians playing were our Patron cellist Guy Johnston and his violinist brother Magnus.

The concert was again sponsored by Sotheby's of Bond Street and also by Robert Holmes and Company. Lesley Carlton Jones, widow of our late Trustee Peter, helped to organise the concert which was a great success.

The two concerts raised a net sum of over £5000 for the NPDG (UK). My grateful thanks go to all the musicians and the teams of willing helpers.

Tony Jellings is a former NPDG (UK) Board member and Fundraising Co-ordinator who continues to support the Group with his innovative fundraising efforts.



Richard Rogerson, Vice Chairman of the NPDG(UK) thanking the two outstanding musicians for giving their time and considerable talents to support the NPDG(UK)



l-r: Julian Bliss, Tony Jellings and Julien Quentin relaxing in the Jellings' home after a highly successful concert which raised over £2,000 for the NPDG(UK)

Raising Money for Niemann Pick

By Shani Miller

My name is Shani, and I am 12 years old. I go to Ellen Wilkinson High School for girls.

Since January my class mates and I have been selling sweets to raise money for Niemann Pick and have raised £90.65 already - but we are aiming for a larger amount of money. My class mates and I have been spending our Monday, Wednesday and Friday break and lunch time going to different classes selling

sweets, getting donations, telling people about the disease and giving out leaflets, copies of when I was in the newspaper and the Niemann Pick magazine and also Niemann Pick stickers.

My class and I even want to do an assembly about Niemann Pick so the whole school knows about it and then they will donate money and we will get a lot of donations for the Niemann Pick Disease Group.

I can't wait.



Scafell Pike Challenge

Roger Franklin set himself a challenge for Awareness Week, to climb the infamous Scafell Pike in the Lake District. He successfully completed the challenge at 11.13 a.m. on Tuesday 8th August. He writes:

"Bill Owen and I set off at 7.30 to drive the 80 miles to Wasdale Head at the foot of the Scafell massive. We took a short cut along a very minor road which appeared to run straight across the Lake District. It was an interesting route with double chevron inclines and declines on sharp left and right hand bends. However the views over Wrynose Pass (dubbed Rhino's A*** by Bill) and Hardknott Pass were spectacular. Bill's seriously impressive driving skills brought us to Wasdale Head at around 10.30 and we set off for the mountain an hour or so later.

The plan was that Bill would accompany me on the first section of the climb, along the flanks of the fell, and then make his way back to the Hotel to await my triumphant return. From the road the path seemed to cross the fell at a comfortable slope but within 10 minutes we were both breathing heavily. 'How many hours have we been walking?' asked Bill as some measure of the task began to dawn on us. A short while later Bill stopped for a breather and ushered me

onwards as it looked as though the attempt would take longer than planned.

The path continued upwards to a sharp left hand bend into the ravine of Lingmell Gill. Ahead and above was the gill crossing. When I reached the crossing I was desperate to stop and so decided to take a couple of photographs as an excuse for an extended breather. The dry summer had reduced the gill to a trickle and one could cross dry shod. The guidebook suggests that in normal conditions some wading is necessary and when in spate one may expect to get thoroughly wet.

Beyond the crossing a thread-like path seemed to wind vertically upwards to dark brooding cliffs whose tops were shrouded in mist. Now the true measure of the climb became apparent and I confess to being not a little daunted. Would my woefully inadequate training be sufficient to get me to the top? I set off at what I thought was a sustainable pace but within minutes was blowing like a whale. As the minutes passed my breathing became more regular and the pain transferred to my legs. For the next hour I saw little of my surroundings, my mind being focussed on the path a few feet ahead of me. When I did stop to

gasp and rest I saw large shed sized boulders just ahead of me. This must be the Hollow Stones described in the guide and I was close to where the track divides.

The split in the track is not obvious. The path I was following appeared to bear to the left and this should have been my preferred route but the mist had descended and I could see nobody on this path. Ahead of me was a steep scree slope below mist covered cliffs. To my surprise six or so figures were making their way up the scree. Their route looked to be the more direct route that led between Sca Fell and Scafell Pike. Not feeling sufficiently confident to go any where on the mountain alone I chose the shared difficulty of the more direct route.

Climbing on scree is hugely frustrating, as you push up the scree slides away and you get nowhere. Also you set off small avalanches of stones that upset those below you. Eventually I caught the group above me and was amazed and humbled to discover mum and dad with their eleven year old son. Dad was not sure of the route but was following the couple above us who, he assured me, had climbed the mountain before. We struggled painfully on and slowly came upon John and Claire. Claire confessed she had not climbed the mountain. John had, but in the dark.

We had stopped below a steep, intimidating, boulder filled gully which John said was the way ahead. The gully faded into mist and I asked John if he was sure he had scaled this gully in the dark. 'Pretty sure' said John cheerfully and set off. Surprisingly the steepness of the gully eased the climb. Now I could use both hands and legs like climbing stairs using a handrail. We emerged from the gully onto the flat stony area of Broad Stand and gazed out on the valley of the river Esc. To the right a path led up to Sca Fell hidden in cloud. To the left the path wound upwards past the brooding cliffs of pulpit rock whose jagged peaks emerged and disappeared as the mist swirled about them. Some debate ensued as to how high we were but for me it was academic. I knew we were around 900m which meant I had less than 100m to the summit and was now sure I would make it.

The last 100m, over large boulders, was very tiring on my weakened legs but finally the slope eased and the summit cairn came into view. I arrived at the summit at 11.13am an hour and 50 minutes since my start. Our little party shook hands and congratulated each other with a pleasure only the completion of a shared endeavour can bestow. I wanted to announce my triumph to the world and particularly to Bill who was waiting patiently in the pub at Wasdale Head but my mobile phone would detect no signal so the world and

Bill would have to wait my return. We ate our sandwiches, took photographs and discussed the route down. At 11.30 I set off down the mountain but that is another story.

Many thanks to all who supported me and NPDG (UK)

Roger Franklin

Thanks to Roger's energetic efforts to date we have received £430. Well done!



Highland Extreme 2006

It was that time of year again, the time to decide what to undertake in order to raise money for NPDG. Cycling coast to coast Scotland in two days was the agreed challenge, a challenge we felt was gruelling for ordinary men like us. We trained from January, culminating in the completion of 850 miles in the saddle.

On the 24th & 25th June, I and 7 other ordinary men completed the challenge. The journey was a total of 217 miles, with 18630 feet of ascent, and was completed in the two days. The event started at the most westerly point of mainland Britain (Point of Ardnamurchan) and concluded in the east coast town of Stonehaven. The route took us through the Highlands, with some breath taking scenery.

We have raised £6700 from this event, raised via a combination of sponsorship and raffle. Having a family member with NP Type C, we try to raise as much



8 Ordinary men at Stonehaven, completing the challenge

money as we can each year, to support this worthy charity. In addition to the money, we are raising awareness of NP which we feel is just as important.

Ian Birchnall

Great Manchester Run

Rob Unsworth completed the BUPA Great Manchester Run on 21st May 2006. He writes;

"The race was brilliant; my finish time was 55 minutes and 58 seconds which I was extremely proud of. The day itself was a really nice day (except for the weather) and it was really humbling to see so many people doing so much for all of the different charities out there. This was my first race so I didn't really know what to expect but the crowds all along the route really helped and the rush of adrenaline when I saw the finish line was amazing.

I'd like to thank everyone who sponsored me, running the race gave me a buzz

but doing it for Niemann Pick gave it a real purpose."



Flora London Marathon

Charlie McLean (NPC) and his parents, Coral & Andy would like to thank the many friends and family who contributed so generously to two events that in total raised over £6200 for the Niemann – Pick disease group.

In April Charlie's uncle, Graham Pattison, bravely completed the Flora London Marathon, in just over 4 hours, an incredible time and a brilliant effort. Graham said, "I really wanted to raise some money to help Charlie and was amazed at the tremendous support and generosity I received". Charlie travelled to London to cheer along 'Uncle Graham' and it was a truly memorable day for all.

In a second event, Charlie's friends at Bradley Stoke Mums and Toddlers organised a 'Toddle Waddle', which involved the children completing a sponsored obstacle course. There was a raffle and a cake sale helping to make the event a huge success that everyone really enjoyed

Both events received extensive local press coverage, raising awareness of the disease, which in turn



resulted in an unexpected donation from a local charity (BCWA), after reading about Charlie and NPC in the Bristol Evening Post.

The Marathon and Toddle Waddle were a huge success and Charlie and his Family would like to express sincere thanks to all those involved – well done!



Thank you!

Thank you to everyone who has been busily fundraising for the NPDG (UK), please keep up the good work; your efforts are very much appreciated. Please contact us at the office on 0191 415 0693 or email niemann-pick@zetnet.co.uk if you would like more information on fundraising or a fundraising pack.

Changing places - Changing lives

A new national campaign, Changing Places has been launched by a consortium of organisations including charities Mencap and PAMIS, and is calling for support from people across the UK. Thousands of people with profound and multiple learning disabilities need Changing Places toilets. These are rooms equipped to allow people to use the toilet with assistance or have their continence pads changed. They include an adult-sized height adjustable changing bench, a hoist, a toilet with space either side for a carer, and plenty of space. Without these facilities carers are often forced to change family members on a dirty toilet floor with little or no privacy – this is unhygienic and dangerous.

However the alternative is to limit outings to a couple of hours or to not go out at all. There are almost no public Changing Places toilets in the UK. The campaign is calling for Changing Places toilets to be installed in all big public places, including city centres, shopping malls, and arts venues. As disabled artist and author Alison Lapper explains:

"The Changing Places campaign is so important because it is about more than just toilets; it's about people's quality of life."

For more information visit www.changing-places.org

Benefits News

Contact a Family – New Pamphlet

Parents of a disabled child are often entitled to a series of benefits and other help. Contact a Family have published a new pamphlet which contains a selection of the most common, briefly explaining

what they are, and who is entitled to claim them. A checklist for parents can be downloaded in Adobe Acrobat pdf version 5 format at <http://www.cafamily.org.uk/Checklist.pdf>

Free advice on baby bonds

Charity Contact a Family has joined forces with independent financial advisers, Peppermint Financial Solutions, to bring a unique investment service to families with disabled children.

Families are being offered free advice on investing Child Trust Fund vouchers in the most appropriate accounts - a service that would normally cost upwards of £100 an hour.

"Parents were increasingly calling our helpline for advice on investing their vouchers; advice we're not qualified to give and they couldn't afford elsewhere," said Jill Harrison, Contact a Family Director of External Affairs. "But we felt strongly that as one of the most disadvantaged groups in society, families with disabled children desperately need this advice, to help them make the most of their money."

Since 2002, all babies have received Child Trust Fund

vouchers, worth £250 or £500 depending on income, to be invested in a dedicated account.

Vouchers not invested immediately lose interest, and are eventually paid into a government selected account that is not necessarily the best for the family.

But now, under the new scheme, which is funded by Her Majesty's Revenue and Customs, callers to Contact a Family's Helpline will be able to book free, hour long advice sessions with Peppermint.

"Our sincere thanks to HMRC for making this innovative service possible and also to Peppermint for joining with us on this new venture," added Ms Harrison.

Parents wishing to book a telephone advice slot with a financial adviser should ring Contact a Family's freephone Helpline on 0808 808 3555

Useful Publications –

Contact a Family – New Fact Sheets

Finding and Paying for Childcare

This tells parents everything they need to know to help them arrange suitable childcare - not always easy when you have a disabled child. It runs through the childcare options for disabled children of all ages, explains how to find out what's available locally and includes a handy checklist to help parents decide if a childcare place is suitable. It also gives full details of the benefits, tax credits and grants that are available to help parents pay - as childcare for disabled children can sometimes be extremely expensive.

Educational Maintenance Allowance (EMA)

Explains exactly how this benefit works and who is eligible for it. Broadly speaking, EMA is for 16 to 19 year olds from lower income families in full time education, or on certain training courses, and funds some of their day to day expenses. Students are eligible for weekly payments, plus possible bonuses and should apply as soon as possible, as payments cannot always be fully backdated.

Both fact sheets are free, and are available from

Contact a Family's freephone helpline on 0808 808 3555, or from the charity's website at

www.cafamily.org.uk

A Guide to Genetics

This booklet is the fully updated third edition of 'The Progress Guide to genetics', first published in 1999 now in a handy pocket size format. This accessibly written reference source is essential reading for teachers, health professionals and their patients, genetic counsellors and anyone wishing to increase their understanding of what genes are and how they work. Topics include genetic testing, gene therapy, preimplantation genetic diagnosis, genetic testing in pregnancy, genetic counselling and ethical issues, plus a comprehensive glossary and details of support groups and genetic centres in the UK. 'A Guide to Genetics' is recommended for any reader aged over 16 and costs £6.49 (including postage) to customers in the UK, and £7.59 (including postage) for customers outside of the UK. For more information and details of how to order follow: <http://www.progress.org.uk/Publications/Index.html>

Positive Aspects Issue 4 September 2006

Family Focus

In Loving Memory

We remember



Orenda Davison-Hulme age 8

In Loving Memory of Orenda Davison-Hulme

Orenda was amazing and brave. She was my special little mermaid sister. I loved doing things with her and I miss her so much. Orenda, and her twin sister Freya, were the best sisters that I could ever have. But they were too good for this world. Be free and come visit me – *her big sister Seraphina*.



Orenda was an amazing sister and a little warrior who looked like a princess. She had to fight for everything and then try to fight to keep it. She didn't have to fight for anybody's love though, as that came naturally. Everything about her was an inspiration to me and she has given me the strength to face anything. Orenda left me millions of happy memories, which will always stay with me. For example, how cool she would act on her walking frame, keeping a smile on her face, despite how tough she found it and how much concentration she had to put in. We always made each other laugh, especially by saying funny sounding words. This had got harder as time went on, but I still loved the unimpressed faces she would pull as I jumped around in front of her, trying to make her laugh! Then recently she giggled again as her chair ran over my toe! I also loved coming home from university unexpectedly to surprise her. Rooms don't seem as bright when she is not in them. I miss how we held each others hands, whether it was on the beach, watching TV or even when they were covered in sticky chocolate mousse. We carried on holding hands as she struggled for her last breath. I look forward to the day when I will see her again with Freya, and they show me what they've been up to since we've been apart. – *her brother Tyrone*

I remember seeing Orenda for the first time, lying in hospital next to Freya. I was amazed at how small she was and how delicate her features were; however, this was only the first of many things that amazed me about her. Orenda had great depth of character. She was very strong-minded, at times quite wild (we used to joke that she was like Mowgli from the Jungle Book), but extremely caring and sensitive to how others were feeling. My fondest memories are those

of her lying on my lap, where she would be so peaceful that she made you forget the world and any troubles for that while. I loved watching her with Freya, holding hands together exploring the ground floor of the house - bottom shuffling while collecting things together in the bags they carried. On our family holidays, she always loved a visit to see the animals. She was never afraid to touch them or be close to them, the noises of the donkeys made her giggle. While the tranquillity of an aquarium would give comfort and capture her imagination. Even later, when she lost the use of words, her personality was so strong that you could really

share what she was feeling. As I sat next to her with my family, as hard as it was to say goodbye, it felt right that she should be back with her sister as when I first saw her. I stroked her long hair, held her hand and felt such pride for my sister who underneath her delicate features was the most unbelievably courageous special person and that's why I'll always be inspired and amazed by her. – *her Big bro' Jesse*

My daughter lived up to her name (which meant '*magical/mystical power*') all her life. Orenda was a complete joy to be with. Even as the disease gradually took over, all I wanted was to be by her side just like the lyrics 'I can't take my eyes off you'. She was a hell of a tough cookie, and a real fighter, with a love for life, even though it wasn't always kind to my wonderful girl. She had a mermaid/siren like quality that drew people to her because she was beautiful inside and out, very loving and sweet in nature with a cheeky sense of fun, always chuckling and smiling. However, she was mischievous and rebellious as well and very stub-born, when she made up her mind about something you couldn't change it! But she would look at you with her big eyes and her two fingers in her mouth and you would forgive her anything. Not that I would have ever got angry because the petty little things, like all my tapes being emptied onto the floor yet again, didn't matter as long as she was happy. In the earlier days I imagined her in the jungle, like a baby Jane, because she

seemed a child of nature who didn't like restrictions. She was always removing her clothes and bottom shuffling happily in her nappy – until that was removed as well! Orenda's trademark was she would wink at the people she liked – it was great! Orenda went, with her twin sister Freya, to a motor-skills group, Rubery Nursery and a mainstream school in Turves Green where they were both loved and accepted – the children took it in turns to push them around the playground in their wheelchairs. My daughters were very special and inspired love, and brought the best out of everyone who came to know them. They were very close and beautiful together. When we lost Freya in her sleep it was Orenda who was lying next to her and holding her hand. It was devastating but on top of that the doctors thought we would lose Orenda shortly after Freya. I picked myself up and would only have positive feelings around Orenda. We had quite a bit of medical equipment at home now, but Orenda got stronger and for months she didn't need to use the oxygen very often and we only put her on the monitor occasionally during the day. I loved the moments when she slept in my lap and nothing relaxed me as much as when I was in tune with her breathing. There was medical equipment to check at night and Orenda missed sleeping with her sister so I took it in turns with her dad to sleep with her. Orenda had a silent strength and amazed the doctors by living a year and five months after Freya. I lost my wonderful, brave daughter on the 14th July, she left this world in my arms, but I felt no separation. This is because I know she lives inside my heart and will forevermore. I believe that she is with Freya now, happy again and getting up to mischief. My Peter Pan girls are reunited and free with the fairies. No restrictions anymore! - *her mommy Gaye Davison-Hulme*

On the 14th July 2006, my last act of love, in this world, for my adorable daughter Orenda was to lift her into her mother's arms and to disconnect the tube from the machine that was supporting her breathing and then gently remove it from her throat. Of course I didn't want to do it, but I hadn't wanted to sit her on my lap, four years earlier, while a nurse stuck a needle all over her arms trying to get a blood sample; neither had I wanted to pass all those nasogastric tubes down her. However, paternal love knows no bounds, we had always been fiercely independent in the way we cared for our twin daughters, and if this was the last thing I could do for her I wasn't going to let anyone else do it.

Orenda was an exceptionally good child. I don't remember her doing anything naughty. She was inquisitive and would get into things and tip things out of cupboards etc, but she never did anything nasty. Freya, probably trying to assert her own independence, went through a very short stage when she would do unkind things to Orenda, but I can't remember Orenda ever retaliating. She would just look hurt and wonder why Freya was being unkind. The phase soon passed and they were extremely close to each other again. She smiled with her whole face and her joy seemed at times too much for her to contain, it was as if her excitement was going to lift her into the air. I always found joy with Orenda, at first in all the normal things fathers do with their children, then later I found as much pleasure in the most simple of things, pushing her through the park, plaiting her hair, falling asleep holding her hand and awakening the next morning still holding it.

Orenda put up with all the medical stuff better than Freya did. While Freya would remove her nasogastric tube, and sometimes even Orenda's! I think Orenda trusted it was for her own good, and would even pat and laugh at her 'drinks machine'. When they were both quite young, they caught a very nasty sickness and diarrhoea bug and as they were slight in build they became dehydrated quite quickly. Orenda was affected worse and was gnashing her teeth when I came home from work. We always tried to avoid taking them into hospital, but on this occasion I took Orenda and stayed the night with her. Once she was on the drip she immediately felt better and she gave me such a look of gratitude. I always sensed that a special link developed between us from that time and I think Orenda always deeply trusted me. It was Freya who was taken first, slipping away in her sleep holding hands with Orenda. I'm not sure that Freya would have coped with the last 17 months as well as Orenda did. The medical equipment increased and life undoubtedly got more difficult for Orenda, but she had a real inner strength and her love of life within our family kept her fighting right to the end; even when the tube was removed she breathed unaided for over half an hour, but it was time for her to go and nestling in her mother's arms, with all of us stroking and kissing her, she slipped off to her sister. I feel such a sense of loss, but that is only a reflection of how much I had. - *her dad Derek Hulme*



Emily Bushaway

I am Emily's nanna, Lisa's mum. The picture is of Emily at three and a half before we knew that she had Niemann-Picks C. As you can see Emily is a perfect little girl and a delight to everyone. It wasn't until she got to around five years old that we noticed little things, like clumsiness, and lots of tears. I can remember Lisa telling me that something was wrong, and my other daughter Jacqui agreeing. I was in denial and would not admit to noticing anything. Of course I had to accept the diagnosis from Great Ormond Street Hospital eventually. Emily is my first granddaughter and is very special. Her name is actually Emily Irene after me and I am very proud of that.



Her sister Sarah is also special as are my grandsons.

Sometimes you think the grief is so unbearable that you cannot survive it, not only do I cry for Emily but also for my daughter, but then Emily will catch my eye and smile and you see what a special gift she is. I can never express my pride in Lisa and her partner Mark. The love and dedication they show to her is beautiful. I know they feel privileged to look after her. We and Jacqui often offer to look after Emily so that they can have a break, but they rarely take up the offer.



We recently went to Disneyland Paris with Lisa Mark and the children. It was a lovely trip and one we will all, always cherish.

I would like to say that sometimes when I look at Lisa I cannot believe that this strong young woman is mine. She and Mark have fought to get all that Emily needs, and at the same time give all the love and attention they can to their other children. Harry and Sarah must be praised too. Sarah, although the baby at eight has never really been the baby, she has had to make allowances for Emily and is like a second little mum to her. Harry is a typical protective big bother.

I want Lisa to know how proud her dad and I are of her, she and Mark have three beautiful children, and we especially thank them for Emily Irene.

Irene Bushaway

Administrative Changes

New Administration Assistant

My name is Sue Lowe and I am the new Administration Assistant for the Niemann-Pick Disease Group (UK). I took over the post in May 2006 and my first four months with the Group have been very enjoyable and rewarding. I really hope I will be an asset to the Group, helping to raise awareness of Niemann-Pick diseases and providing the essential admin support to facilitate the Group in achieving its aims and objectives.

During my career I have worked for The Kennel Club, where I helped to organise Crufts Dog Show, the original teletext company ORACLE and Radio 1 back in the eighties, where I worked with DJ's Peter Powell and Simon Bates – do you remember "Our Tune"? Well one of my tasks when working with Simon was to help pick the daily "Our Tune".

I have lived in Washington, Tyne and Wear, with my two

daughters, Alex (13) and Emily (11), my partner Will and his son Mick (13) since July 2005. Prior to that I lived in Bedfordshire, working in customer relations for a local car dealership. As I worked from home, I was able to bring my job with me when I moved to Washington. I spotted the advertisement for an Admin Assistant with the NPDG (UK) in a local paper and thought it was just what I was looking for - a more fulfilling role. It sounded perfect, especially as 'the office' was just a five minute walk away! I applied and the rest is history.

Attending the Family Conference in Northampton this year was a wonderful introduction to the Group, it was lovely to meet so many of you there and to finally put faces to names. I have really enjoyed working with Toni and supporting the work of the Group. Please feel free to contact me at the office on 0191 415 0693 or email niemann-pick@zetnet.co.uk



National Development Manager Report

It is now September and my first year with the Group is drawing to a close. It has been one of the busiest years I have ever had, full of opportunities and new experiences. As I look back I realise how much I have learned and also how much I still have to learn!!

Our new Admin Assistant Sue Lowe came into post in May - only six weeks before Conference. She proved to be a great help - even though it must have been a real baptism of fire! We are now getting to know each other and it is nice to have someone to talk to other than my computer!

A lot of work went into organising this years Family Conference and I would like to say a big thank you to the many people (and photocopiers!) that helped to make it a great success. I really enjoyed meeting families I had previously only spoken to over the telephone and it was especially good to have delegates attend from around the world. This year the Group said goodbye to Kris White, who was a Trustee of the Group and Children's Programme Co-ordinator for over three years. Kris did a wonderful job with the Children's Programme and she will certainly be missed. We are already planning Conference 2007 and would be pleased to hear any suggestions you may have to ensure it is an enjoyable and beneficial experience for all who attend.

In July the Group were nominated for the Communiqué Patient Association of the Year Award. Jackie and I were delighted to be invited to attend the awards dinner, at the Grosvenor Hotel in London. It was a very glittering occasion, we were pleased to have each others company as otherwise it would have been rather daunting! The other groups nominated for the Award were a lot larger than NPDG (UK) and we were very pleased to receive a commendation as runners up. I would like to thank Sante Communications for nominating us and for their support at the event.

Also in July I had the pleasure of meeting Suzi Dietz, who is the new German Niemann-Pick Support Nurse. She visited the Willink along with her colleague Hans Kluenemann to find out Jackie's experience of the job. Their visit co-incided with the first adult NP-C Clinic to be held at Hope Hospital, at which I met some old friends and new.



The response to the third annual NPDG (UK) Awareness Week was fantastic this year, with many of you taking advantage of the good weather by holding 'Pick-Nics' and events around the country. Many of your events captured the attention of local media, helping us towards our aim of raising the profile of NPD. It is great to see so many people joining in the fun - well done to you all and keep up the good work!

The coming months are going to be just as busy, with meetings planned to progress the Care Manuals for Type B and C, new funding applications to make and work underway to complete the new look website - I wonder how I will fit in Christmas shopping! At least I do not have to look for cards - as you will have seen earlier in this newsletter the children have done a wonderful job designing the Christmas cards, I hope you will support us by ordering a pack or two.

Please continue to contact me at the Office if you feel I can help in any way, on 0191 415 0693 or email niemann-pick@zetnet.co.uk

Toni

Toni Mathieson
National Development Manager

Feedback

How can we make it easier or more interesting for you to give us feed back?

By Dave Roberts, NPDG (UK) Trustee

The NPDG (UK) Board of Trustees meet at least four times a year to consider the work of the charity and how well we're performing against our objectives. We are committed to providing effective support through care, information and research and the work that we do takes input from 'around the table'. We rely on each other's experience and expertise in formulating policy and decision making, supported by medical and clinical advisors and other expert opinion as appropriate.

However, it is also important that we understand the views of our members and build these into the decision making process. We have tried to do this by asking for feedback. So far, whilst we have not been very successful in gaining feedback we are now in a better position to compare different approaches.

In the last edition of the newsletter we featured some Questions and Answers about the Niemann-Pick disease with a request for feed back. In response to the newsletter article, we had no feedback but were more successful when we asked face to face. We also asked for feedback about the web site and requested members try out the new forum. Again, apart from Vik, Toni, Clive and Dave trying it out, we have had no other users and no feedback.

With little or no response, it makes judgement and decision making more difficult. For example, should we add the Questions and Answers to the information pack? Are they aimed at the wrong age group? Should we produce an A3 version or should we do nothing? These are all valid questions with cost implications against each one, get the answer wrong and it's money down the drain.

Again, I know that the web site development is behind schedule and there is a lot more work to be done, but how important is it to our members and to other interested parties? Should we spend more money on further enhancing the site? Will members be more likely to access the site and use it to give feedback? I guess the answer is 'we don't really know, but we will all have differing views'. How best to move forward on these topics will be for the committee to decide but, without knowing our members views, the outcome may not be the best for all concerned.

In stark contrast, at the Family Conference, we had a 97% feedback rate. This was because the feedback forms were included in the pack of information and we 'encouraged' you and set aside time on the Sunday for you to complete them. Maybe we can learn from this experience and have feedback sessions as part of the Family Conference agenda. The outcome of these sessions could then be fed into the next Board Meeting. – Please let us know what you think!

To complete the picture, the respondents within the age group that the Question and Answer leaflet was aimed at, gave the following responses:

- The questions and answers were very clear and added a lot to our understanding of the disease and the issues affecting us. Just having the information made us feel better.
- We don't know if there were any mistakes but we could not find any apparent contradictions in the leaflet. The layout and order of the questions were very good to the extent that as a question formed in our mind, it always seemed to be the next question that was answered.
- There were no more questions but we thought that awareness, outside of the family, was still an issue.
- We thought access to a regularly updated information pack, when required, would be useful to individuals and professionals, - perhaps it could be loaded onto the website.

Finally, feedback is a two way process, and we are most grateful for your support and hard work that goes into fund raising, contributions to the newsletter and support for the Family Conference. Please keep up the good work.

I think it is both our jobs to understand how we can better encourage feedback and involvement in the work that we do on your behalf but this can only be done with your willingness to participate – let us know what you think by email, internet or letter.

Many thanks

If you would like to respond to this article please email niemann-pick@zetnet.co.uk or write to the NPDG (UK) Office, 11 Greenwood Close, The Pastures, Fatfield, Washington, NE38 8LR

NPDG (UK)
14th Annual Family Conference
23rd & 24th June 2007

We Need your Input

We would like to involve all of our families and friends in helping us to make the 2007 conference a success. As we are still in the planning stages, we would love to hear your thoughts and suggestions. Perhaps you would like to hear a particular speaker, or you have an idea for a Break Out Session, please let us know. If you have not yet attended our conference, we would be interested to find out if there is anything we can offer that might encourage you to come along.

We received positive feedback after Conference 2006 regarding the choice of venue and the content of the programme. However, there is always room for improvement and the suggestions you made to us on your evaluation forms are high on our list of considerations. They include:

- Looking at ways of helping families deal with day to day life with NPD.
- How does the NPDG spend the funds available to them?
- Who are the Board of Trustees and what is their role?
- Relationship issues – When a diagnosis is received, often the whole family need assistance in coming to terms with the way their life will change.
- Research, research and more research – a very popular issue!

Most of you thought holding a two day conference allowed time for families to get to know each other, would you like more social time included in the programme?

Children and Young Adult's Programme – after the success of the programme at the last conference, especially the trip to Wickstead Park, we hope to be able to do this again. What would your children enjoy? What activities would you like to see included in the programme?

As the Break Out Sessions at our last conference proved to be very popular, we propose to run these again. Some of the suggestions we received for Break Out Sessions after last years Conference are:

- More practical sessions – how to manage symptoms of NPD.
- Sibling workshop – coping with the issues faced by siblings
- Employment Rights
- Provisions for carers
- Carrier testing as a form of prevention?

This is your conference; help us to make it as beneficial and informative as possible.

The suggestions I would like to make are:

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Please return your suggestions to:
Toni Mathieson, NPDG (UK) Office 11 Greenwood Close, Fatfield, Washington, NE38 8LR.
Alternatively, please call 0191 415 0693 or email niemann-pick@zetnet.co.uk

Jackie's Journal

Hello again from sunny Manchester with the photograph to prove it!

Actually this photo was taken earlier this year when we were visited by Hans Klunemann the Neuropsychiatrist who cares for adult patients with NPD and the new German Niemann-Pick Nurse Suzi Dietz. They joined us at 2 clinic sessions and then we visited a family in Birmingham so they had a chance to see a bit of England. Helena and Toni were here too so this is truly the Niemann-Pick team, unfortunately minus Liz as she was not in that day. Hopefully Hans and Suzi will be available to come to our conference next year.

Talking of conference I think that all felt that this year's weekend in Northampton went very well. All the families and professionals I have spoken to had a good time especially those new families who had been worried about attending. This conference is for you so please do let us know what we did right or wrong and if there is anything you would like us to do next year.

Another conference I attended was the International MPS and related disorders conference. MPS (mucopolysaccharide diseases) are lysosomal diseases with similarities to NPD and the research into these other diseases will hopefully benefit all. It was a chance to meet up with nurses and scientists working with metabolic diseases internationally and to share our experiences. Also it was in Venice so I could occasionally top up the tan!!

More recently I was privileged to go to the American NPD conference in Dallas. It was lovely to meet up with old friends as well as some new ones. My main talk while there was discussing dementia in children and young adults and resulted in a lot of discussion at the time and later over coffee. The Niemann-Pick world is very small so I feel it is vital that we have close links with all the other countries so we can all work together and have a bigger voice.



Over the next few months we will be contacting quite a few of you as Liz gets on with the vital research work that she has got her teeth into. Liz has written her own report so I won't say much more, other than how did we manage without her!

Back to work now, or is it morning coffee time? For those of you who are early risers I am usually in the office by 7.45 so if you want to contact me before you go off to work I should be around. Alternatively I usually have my mobile with me, mainly for office hours but if you have a crisis outside these times leave me a message and Toni or I will help you.

Take care

Jackie

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Niemann Pick Disease
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From the editor...

I should like to thank all of those who have contributed to this Newsletter. Please continue to submit articles, stories, poems, management tips, advice, children's contributions, fund-raising ideas and anything else you feel may be of interest, especially photographs!

Send your contributions to: Toni Mathieson, 11 Greenwood Close, The Pastures, Fatfield, Washington, NE38 8LR
or Email to niemann-pick@zetnet.co.uk

Please send your articles for the Summer Newsletter by 1st March 2007

Disclaimer: Information which appears in this Newsletter is for the express purpose of raising awareness and does not necessarily reflect the views of the NPDG (UK). All medical information should be reviewed with your doctor before being acted upon