



Niemann-Pick News

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

Winter 2006

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Ben Alexander and family

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Chairman's Chat



recently read an article about what we can give for Christmas. For so many giving has come to mean something material some-thing financial. Money was originally developed to allow us to exchange goods, services and time. Before money was in common use we could give our time, our skills to making something or our effort to support a friend or someone in need. It's time to remember that, and I hope that is what happens as you read this newsletter.

Thanks to the efforts of many of you who have exchanged your time effort and skills helping to raise funds we are now embarking on a process of exchanging that for family support, clinical research and the development of a network of similar groups to carry forward the exploration of answers to this group of diseases. Recent fund-raising and the Niemann-Pick Awareness week have given a real boost to our funds and we are now investigating four new projects.

i) The employment of a clinical research nurse: this will enable us to begin to collect much needed data regarding clinical aspects of this disease. This would be a first for this group of diseases any where in the world. This has the potential

to improve understanding and help with patient support and therapy development.

- ii) Support for the purchase, in partnership with others much needed equipment help with the understanding of the underlying genetic faults in specific patients. This would add to understanding and speed up diagnosis.
- iii) The further development of our website in terms of keeping it up to date and active.
- iv) The formation of international alliances in terms of Niemann-pick disease and lysosomal storage diseases which would allow us to tackle some of the bigger issues such as dealing with governments, health boards, drug companies and research concepts.

What Christmas presents these make!

This is about the efforts of many people being translated from love and care into making a difference. I hope as you read this newsletter you can feel the underlying message of working together, bringing whatever we can, and exchanging it to support a common purpose. I hope 2006 will see those efforts rewarded.

Jim Green

Chairman
Niemann-Pick Disease Group (UK)
Dec. 2005

Research News

Press Release from Actelion - OGT 918 Clinical Trial

One-Year results on the use of miglustat (Zavesca) in Niemann Pick Type C presented at the annual conference of the American Society of Human Genetics

ALLSCHWIL, SWITZERLAND – 27 October 2005 – Actelion Ltd announced today a poster presentation at the annual conference of the American Society of Human Genetics (ASH-G) of one-year results from a randomized controlled study evaluating safety and efficacy of miglustat (Zavesca®) in patients with Niemann Pick Type C (NP-C), a rare genetic disease associated with multiple neurological manifestations.

Although not statistically significant, the 29-patient-study showed trends toward improvement or stabilization in terms of saccadic eye movements (by electrophysiological assessment) and swallowing and audition (by clinical assessment) in patients receiving miglustat compared to standard of care. The study will continue as planned for another 12 months, during which time all patients will receive miglustat. A complete discussion of currently available study findings are contained in: Patterson M., Vecchio D., Prady H., Ait-Aissa N., Abel L., Wraith E. - Poster 2505/T, ASH-G 2005 (Salt Lake City).

Marc Patterson, M.D., FRACP, Professor and Head, Division of Paediatric Neurology at Columbia University New York/USA, commented: "We know already that miglustat crosses the blood-brain barrier and accesses the brain. For the first time, we have been able to observe some potentially beneficial effects in patients with NP-C. I am also encouraged with these observations as they could suggest some restoration of function in neurons that have been altered by the disease process. As such, this study is another step forward in our quest to understand and potentially treat this complex disease".

The safety profile in this study evaluating miglustat 200 mg three times a day was consistent with safety information generated with the use of miglustat 100 mg three times a day.

Details on study and results

The study is being carried out at both Columbia University, New York (US) and The Royal Manchester Children's Hospital (UK). Twenty-nine patients (>12 years old) with NP-C were randomized to receive either miglustat 200 mg three times daily (twenty



patients) or standard care (nine patients) for 12 months and a series of clinical, electrophysiological and quality of life assessments were conducted at regular intervals. The dose double that recommended for Gaucher Disease type 1 (GD1), in order to increase brain exposure to miglustat.

Twenty-five patients have completed the first 12-month study period and four have discontinued treatment (three in the miglustat group, one in the standard care group). Results from the 12-month data showed improvement in horizontal saccadic eye movements (by blinded centralized assessment) and improvement in swallowing, none reaching formal statistical significance. The analysis also showed stabilization in auditory acuity in treated patients compared with deterioration in the untreated group.

The safety profile of miglustat observed in this study was consistent with the results obtained in patients with GD1 treated with miglustat 100 mg three times daily, particularly regarding weight loss and gastrointestinal disturbances.

Based on these results, the study will continue as planned for a further 12 months during which time all patients will receive miglustat and will be followed with the same set of assessments.

About Substrate Reduction Therapy with miglustat

Miglustat is a small molecule with a large tissue distribution and is administered orally. It works on the principle of substrate reduction therapy, by reducing the rate of formation of glucosylceramide, a precursor of glycosphingolipids that are stored in NP-C. It is the storage of these lipids that contributes to neuronal dysfunction and ultimately neuronal death in several areas of the central nervous system.

As you can see, the results for the first 12 months of the over 12yr olds trial of OGT 918 (miglustat / zavesca) are encouraging, showing trends of improvement.

We have always been very aware that families of patients with NPC have waited a long time for the study and subsequently the 'results'. We hope that, although the improvements reported did not reach statistical significance, you are encouraged by the improvements reported and also reassured by the safety reports.

The trial will continue as planned and we look forward to the next set of results from the month 12 – month 24 study.

Best wishes

Nikki West

Clinical Project Manager
and

Helena Prady

Research Nurse / Trial Co-ordinator



Update from Genzyme.

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

The Genzyme team is looking forward to beginning the first ever clinical trial of recombinant human acid sphingomyelinase (rhASM) for treating ASM deficiency (Niemann-Pick Disease, Type B). The clinical trial design, production of the drug for the clinical trial, and preclinical testing were the coordinated effort of well over 100 scientists and physicians at Genzyme working with Drs. Desnick, McGovern, Wasserstein and Schuchman at the Mt. Sinai School of Medicine (MSSM).

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.

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 analyzed before deciding whether to proceed to the next. The decision about whether to proceed to the next dose level will be made by a data monitoring committee that is independent of Genzyme. 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 analyze 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 as part of comprehensive drug registration package.

Over the next several weeks, Genzyme and MSSM will be completing final preparations for starting the trial. We hope to screen the first patient for the study early in 2006.

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

Senior Director of Program Management
Genzyme Corporation (www.genzyme.com)

What rare diseases can teach us

By Steven U. Walkley

Professor of Neuroscience at the Albert Einstein College of Medicine in New York

Research into many maladies is under funded. This hurts patients and prevents science from unlocking the keys to the body

THIS SUMMER, 8-year-old Jessica Leoni was admitted to UCLA Medical Centre for treatment of seizures brought on by a rare neurological disease called Niemann-Pick Type C, or NP-C. After 11 days, her doctors finally identified a drug that could control her seizures, and she returned to her Westchester home with her relieved parents.

But this family's relief is bound to be tempered by the knowledge that there is no known cure for NP-C and, because it is so rare, research to find a cure is badly under funded.

And NP-C is only one of about 6,000 such diseases, identified by the National Institutes of Health as ailments that each affect fewer than 200,000 Americans.

Both the government and private drug companies too often look at such numbers and say no to the funding required to finding treatments and cures. But that traditional calculus is incomplete — there are plenty of good reasons to do more to fund rare-disease research.

First, even rare diseases affect a huge number of people. Nearly 25 million Americans suffer from one of the conditions that the National Institutes of Health lists as rare.

The financial and emotional burdens on our society of not adequately treating such illnesses are immeasurable.

In addition, however, such research also has ramifications far beyond the individual sufferers. Rare diseases represent unique windows that allow scientists to peer into the intricate workings of our bodies.

Indeed, 3 1/2 centuries ago, long before the initiation of the human genome project and other riches of modern medicine, the leading physician of his day, William Harvey, implored his colleagues to carefully investigate the rarer forms of disease in order to advance the proper practice of medicine.

Harvey's prescient view was that determining what was missing or altered in an uncommon disease could inform us as to its importance in the normal human body.

Harvey's reasoning remains as sound today as it was in 1657.

Consider, for example, Fragile X syndrome, a leading cause of learning difficulties and autism that, according to the Centres for

Disease Control, affects 1 in 4,000 boys and 1 in 8,000 girls in the United States. This condition has been linked to a defect in one gene on the X-chromosome that codes for a protein, FMRP, believed critical for transporting genetic messages in brain cells. Without the protein the brain looks normal but learning and memory are severely impaired.

Scientists believe that studying Fragile X will lead to the discovery of how the nervous system normally stores information — still one of the brain's great mysteries — and by fully understanding how this protein facilitates learning, we may be able to help not only patients with Fragile X but also those with other, unrelated forms of learning difficulties.

Researching diseases such as NP-C could be similarly illuminating. Brain cells in children with this disease are characterized by the formation of abnormal "tangles" that are identical to those of adults with Alzheimer's disease, though the diseases are not otherwise closely related.

Drugs developed to control NP-C disease could provide critical clues for controlling the progression of tangle formation and neuron death in Alzheimer's.

Perhaps the most short-sighted aspect of marginalizing the so-called rare diseases is that they really may not be so rare. Instead of adding up the numbers based on how many sufferers there are for each disease, we should do the calculations recognizing the commonalities among them.

Once again, NP-C is a good example. It is what is known as a "lysosomal storage" disease — one that involves a tiny structure in cells known as the lysosome. There are nearly 60 types of lysosomal diseases known today — such as Gaucher, Krabbe, Sanfilippo and Hurler. Individually they may affect fewer than 100 patients but, as a group, lysosomal diseases are estimated to occur in about 1 in every 7,500 live births, a frequency midway between two of the best-known "rare" diseases — cystic fibrosis and phenylketonuria, or PKU.

Knowledge gained by the study of one of these diseases is highly likely to provide insight into others in the group.

Many rare diseases, including those in the lysosomal group, begin in childhood, which means that research leading to corrective therapies could rescue individuals from a fate that may include seizures, blindness, learning difficulties or dementia and death, often after decades of declining health. Such research would also lessen the burden that long-term diseases place on our society as a whole.

In the past, rare diseases have remained singular tragedies but, as information sharing has become easier with the Internet, families with rare diseases are discovering one another. They are joining forces to lobby for a different view of the costs and benefits of adequately funding rare-disease research.

This can be seen through the growth of organizations such as the National Organization of Rare Diseases and the Genetic Alliance, as well as smaller groups dedicated to finding treatments and cures for individual rare diseases.

Significantly, new coalitions are also emerging, as is the case with lysosomal diseases, where umbrella groups such as the Hide & Seek Foundation, based in Los Angeles, and the Global Organization for Lysosomal Diseases have been formed.

These organizations, dedicated to finding treatments and cures for individual rare diseases, have a simple message: Investment in research on rare diseases clearly pays dividends on many levels and benefits us all.

Certainly the simplest of calculations — the one that tells us which diseases affect the fewest Americans — shouldn't be the main determinant controlling how we spend our research dollars.

STEVE WALKLEY is currently Chair of the Research Sub-Committee of the NNPDF

This article appeared in the Los Angeles Times on September 3rd 2005

Research Report by Bill Owen, Research Co-ordinator

'Questions and Concerns'.

Getting close to research into Niemann-Pick brings a familiarity that makes it easy to overlook the fact, that many close family relatives do not have anything more than a very basic understanding of the disease or the issues of being a carrier. It is not always easy to sit down with relations and give undivided attention to their concerns. Equally they are often reticent in raising questions in case they appear silly or cause hurt. This was brought home to me by Dave Roberts, a new committee member and my brother in law. I have found it relatively easy to discuss questions with him but, he had noted that our younger relations felt uneasy and did have questions and concerns which they had not raised with me. This is particularly significant as many of our 'next generation' family are at, or reaching the age, when they will be starting families of their own.

Dave had raised this at the Committee meeting in September and set about making a collection of questions from our family. He presented a list at the November meeting and Jackie Imrie our Nurse Specialist, agreed to take the questions to the Willink and provide answers. We also thought about the need to extend the exercise to all involved in the Niemann-Pick charity so that the many questions and concerns could be co-ordinated. The aim is to collect

and collate the questions, arrange to provide answers, and publish these in a separate leaflet and on the website. We will aim to do this by the spring next year which will allow further discussion at our Family Conference in June 2006.

The list of questions to date is provided here as a start. Please check them out with family members and add to the list. You can do this by e mail (niemann-pick@or standard mail to Toni who will collate the list.

ABOUT THE DISEASE

- What is the Niemann – Pick disease?
- What is the history of the disease?
- Is the disease hereditary?
- How many people are affected by it?
- Is it found throughout the world?
- Is it more prevalent in male or female?
- Can it skip generations?
- Can relatives be identified as high/ low risk?
- How many different types of N-P are there?
- What are the differences?
- What are the symptoms of the disease?
- Can it occur at any stage in life?
- Is it more likely in younger / older people?

TREATMENT

- If identified at an early stage, is there any treatment, can it be controlled?
- Can treatment reduce the chances of the disease developing, can it lie dormant, what triggers it?
- Are there cases of full or partial recovery?

CARRIER ISSUES

- What tests are available to determine if a family member is a carrier of the N-P gene?
- What is involved in being tested for the N-P gene?
- What are the probabilities of me being a carrier if one or both of my parents were carriers?
- If only one person in a relationship is a carrier of N-P, will this mean that a baby could inherit the disease?
- How would a baby be affected if only one parent in the relationship had the N-P gene, could the baby inherit the disease or would the baby be a carrier?
- Can babies in the womb be tested for N-P?
- If I was a carrier of the N-P gene, can my partner be tested for abnormalities in his genes or clues that may indicate a possible risk of him being a carrier?
- How many people could be carriers? What statistics are available to allow us to understand the chances of a partner's family having NP and therefore ourselves having a baby with N-P?

SCIENTIFIC

- If there is known to be type C within the family, would it always be type C that is passed down to the next generation or could it change forms and show as a type A or B?
- Is there just one or a number of defective genes that cause N-P?
- Is it contagious in any way e.g. by blood transfusions?
- Is any work being done to develop tests for the partners of people affected by the NP gene?
- What research is being done to find a cure?
- Are there any other diseases related to N-P?

COMMUNICATION

- Could possible carriers have been wrongly diagnosed in the past?
- What questions do we ask the partner's family about their memories of illnesses or bereavements in their families, so as to eliminate the possibility of

the partner's family being affected by the disease?

- Should distant relatives be informed that N-P is within the family? Would they want to know and how should they be informed?
- Does every relative have to be informed?
- People in the medical profession that we have spoken to, don't know about N-P and haven't been able to talk to us about the risk of having a baby with NP. Is anything being done to make professional people more aware of the disease?
- Where do we get information from, if we want to understand more about N-P?

Other thoughts

We have all seen or asked some of these questions before and as a member of an affected family our great concern is that someday there will be a cure for the NP diseases. How and when this will happen remains to be seen and we need to look to the future and ask what else can we do to stop the disease. The understanding of genetic diseases is new knowledge to humanity, especially rare recessive conditions which appear to strike out of the blue and, with no history in families and no obvious pattern as to which child gets affected.

The world is different now, we have a better understanding of how inheritance works but we remain with the problem of, not how to cure the disease but how to stop it in the first place. It may be unlikely that we can ever stop the disease from striking at random in new families but, once it has struck with the tragic consequences, with which we are familiar, a knowledge of what gene mutations are involved and who are carriers of these, may help us in the future. To allow these genes to disappear, only to re-emerge in future and cause misery to our descendants is something that none of us would wish for.

Our basic problem at present is our concern for family members who intend to start a family of their own. They can easily be tested for the family mutation but their partner cannot. At present there is no test to check whether a NP gene from a person selected at random is functional or not. This will not always be the case and at some time in the future tests will become available. When this eventually happens, it will be important to know who those individuals are that carry the defective gene. Those who are carriers and do not know it, already carry 50% of a time bomb and, even though the risk of meeting with the other 50% of the bomb is very small, it is finite and high impact as we know to our cost.

The debate on the value of carrier testing for recessive conditions is an important one and, it is essential for families to join in and express their fears and concerns. Also support is needed as the new tests will not happen unless by good fortune, or pressure from patient groups and affected families.

Volunteers Wanted

Professor Chris Harris and his team at the University of Plymouth have recently been awarded a research grant from the national charity Cerebra - for brain injured children and young people. The research team are developing new methods of recording eye movements in babies and young children with neurometabolic disorders.

As part of this research we are seeking to recruit infants and young children with Niemann-Pick type C disease.

The procedures being developed are straightforward and non-invasive and are conducted in a child friendly and relaxed environment.

Return travel, one night's hotel accommodation, and

You may wish to discuss this article with your extended family. If you have any questions or concerns to add to this list, please contact Toni Mathieson at the NPDG (UK) office or email niemann-pick@zetnet.co.uk

all meal expenses will be paid for the participant and their parents/guardians.

For further details, contact a team member on: 01752 233359 / email: nderbyshire@plymouth.ac.uk or fbudge@plymouth.ac.uk. The website at www.harrislab.com gives details of the work carried out by in the teams' SensoriMotor Laboratory.

Helena Prady adds;

'Until we have clarification from Actelion Pharmaceuticals Ltd, no patients who are currently on the OGT 918 trial should participate in this one'

Contact Helena for more information on 0161 922 2967 or email helena.prady@cmmc.nhs.uk

Ara Parseghian Medical Research Foundation

The Parseghian Family and

The Ara Parseghian Medical Research Foundation



In 1994, just months after learning that three of their four children had a fatal, genetic disease called Niemann-Pick Type C, Cindy and Michael Parseghian and a dedicated army of volunteers founded the Ara Parseghian Medical Research Foundation. Ara, the legendary Notre Dame football coach, is the children's grandfather and is also very involved in the cause.

The Foundation's purpose is to fund research and promote worldwide interaction among scientists, research institutes and universities working on NP-C and related diseases in hopes of finding a treatment and cure. The number of labs working on NP-C has increased from less than 10 to more than 50 worldwide since the inception of the Foundation.

As of July 2005, 21 medical research projects are being funded by the Parseghian Foundation in the US, Canada and Europe. The Parseghian Foundation has funded another 33 grants in the past.

Of the more than 400 rare disease organizations in America, the Parseghian Foundation has led the way in raising funds--totaling more than \$22 million in

eleven years. Administrative costs are held to a bare minimum due to the dedicated support of their Tucson, Arizona volunteers, as well as numerous others from across the US who have stepped forward to help including other NP-C families.

Each year, the Foundation raises approximately \$2.2 million through fundraising events, personal and corporate contributions. More than 94% of all funds raised go to research helping not only those suffering from NP-C, but other cholesterol-related diseases such as stroke and heart disease, as well as Alzheimer's.

The Parseghian's son, Michael, lost his battle with NP-C in 1997 at age 9. Christa passed away in 2001 at the age of 10. Sadly, Marcia died on August 6, 2005 at the age of 16. However, the Parseghian family and Foundation is resolved to keep the battle waging to try to save the lives of hundreds of other NP-C children and young adults around the world.

Please feel free to visit the APMRF website at www.parseghian.org to read a lay summary of each of the presentations given at the 2005 Scientific Conference as well as description of the 20 research projects being funded by the Parseghian Foundation.

New Italian association for Niemann-Pick Disease.

We are delighted to announce the formation of the Associazione Italiana Niemann Pick. The association was constituted at a meeting at Burlo Garofolo Hospital in Trieste on November 19th - 20th 2005. Already their Website is up and running and can be found at www.niemannpick.it

We send our congratulations to our friends in Italy and wish them well in their new venture.



News from the Niemann-Pick Association in Spain

It is with great sadness that we heard of the death of Juan Giron, President and Founder of the Niemann Pick Foundation of Spain. Juan Girón was 42 years old.

The loss of his four-year-old son, Juan Antonio, to NPC in 2001 made him determined, with the help of his wife Juani, to start a foundation that would enable other children to have the quality of life that his son did not have. He wanted to ensure that other families affected by Niemann-Pick disease would have support and information to help them in their journey with this disease.

Juan was a man of great compassion and human sensitivity. He dreamed of a world where no child would have to bear the burden of Niemann-Pick

disease. He has bequeathed a great Project, the Fundación Niemann-Pick de España

We send our love to Juani and to his friends, the Niemann-Pick families of Spain, as they strive to fulfil the challenge to continue the work initiated by Juan.



Newly formed Niemann-Pick Association in Argentina

In Argentina eight families affected by Niemann-Pick disease have formed the Asociación Niemann Pick de Argentina. The families live in Buenos Aires, Quilmes (Buenos Aires) San Justo, (Santa Fe), the Pampas (Cordova) and San Juan. They will provide support

and information to Niemann-Pick families in Argentina. We send them all our very best wishes.

Email: niemannpick@argentina.com

News from GOLD

Annual General Meeting

GOLD's AGM took place on Tuesday 25th October, as an ancillary meeting during the American Society of Human Genetics Annual Conference in Salt Lake City, Utah, USA. Minutes will be posted on the GOLD website, www.goldinfo.org.

Dr Charles Scriver, MDCM FRS, Alva Professor Emeritus of Human Genetics, McGill University delivered the Guest Lecture. The theme of this lecture was "A phenotype Project? What might THAT be?" Dr Scriver described GOLD as a unique organisation, which, by uniting scientists, clinicians, patient organisations and commercial organisations created a network which could be a prototype for other orphan genetic diseases. The abstract is posted on the GOLD website.

GOLD Management Council Elections, 2005

GOLD's Management Council is elected by the membership. Each year, 4 members of the sitting Management Council retire. The ballot for the Management Council elections closed on Sept 30th, and results were announced at the AGM.

NPDG-UK nominated Dr Ed Wraith, our medical advisor, whom many of you will know from his clinics at the Royal Manchester Children's Hospital, for the Management Council. Ed was newly elected to the GOLD Management Council. Sitting Management Council members who were re-elected are Rhonda Buyers (US Gaucher Association), Christine Lavery (UK MPS Society) and Dr Roberto Giugliani (Chief of the Medical Genetics Service of Hospital de Clinicas de Porto Alegre, Brazil).



Registry

As reported in the last NPDG newsletter, planning for an international independent registry using GM1, GM2 gangliosidoses and Niemann Pick C as a model has commenced. A Medical Advisory committee has been appointed: Dr Michael Beck, Dr Roberto Giugliani, Professor John Hopwood, Dr Marc Patterson, Dr Cynthia Tiffit, Dr Marie Vanier and Dr Ed Wraith. Several MAC members met during the European Study group on Lysosomal Diseases to discuss diagnostic criteria and data sets, which are currently being developed.

Website

GOLD's website is updated regularly, and discussion forums are available for anyone who would like to use them.

Membership of GOLD

There are now 99 member organisations, from 23 countries, representing all Lysosomal Storage Diseases.

Ann Hale

Executive Director

GOLD

ann.hale@goldinfo.org

Newly Designated NSCAG Centre in Manchester

What does NSCAG Designation mean?

The shared care management of children and adults with lysosomal storage diseases (LSD), including Niemann-Pick Disease, are based at the Willink Unit, Royal Manchester (RMCH) and the department of LSD, Hope Hospital, Salford. Both sites are designated LSD centres by the National Specialist Commissioning Advisory Group (NSCAG). At the NSCAG centre the initial visit will include a discussion of the disease and its future management. Normally this visit is arranged after a diagnosis is made in the laboratory. Treatments, if available, such as enzyme replacement therapy and bone marrow transplant are organised at the site, and the Nurse Specialist involved will liaise with the family about the future

care. We also are a resource for other health care professionals to contact us as these diseases are still rare. When a diagnosis is made it is often a shock for the families and they will require support and guidance throughout.

If treatment is not initiated here at the NSCAG site then the staff at the centre will arrange appropriate follow up and monitoring of the patients condition.

Most patients with Niemann-Pick Disease attend clinic with Dr Ed Wraith twice a year, but more frequently if needed.

If you would like further information please contact Jackie Imrie at the Willink on 0161 922 2414 or email Jackie.imrie@cmmc.nhs.uk

Niemann-Pick Disease

The Peter Carlton Jones Memorial Award

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, in respect of this first year (2005 / 2006) of the 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
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11 Greenwood Close
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NE38 8LR
E-mail: niemann-pick@zetnet.co.uk
Tel: 0191 415 0693

Closing date for submissions: 28th February 2006

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 on 24th / 25th June 2006, to formally receive the 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.

Family Conference

Save the Date!

2006 Annual Family Conference

24th and 25th June 2006, the Hilton Hotel, Northampton.

The theme for the conference will be

'Forging New Relationships'

We would like to involve all of our families and friends in helping us to make the 2006 conference a success. 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. This is your conference; help us to make it as beneficial and informative as possible.

Contact Toni Mathieson at the NPDG (UK) Office on 0191 415 0693 or email niemann-pick@zetnet.co.uk

Full details of the conference will be available in the next issue of *Niemann-Pick News*.

Fundraising News

Niemann-Pick Disease Group (UK) Awareness Week

22nd-30th October 2005

Last year, the first NPDG (UK) Awareness Weekend was a great success, and it was good to see so many of you join in the fun again this year.

Lorraine Bishop, with the help of her family and friends, raised £762.52 at a variety of events. Lorraine's husband Neil held a raffle at his place of work, Homebase- Lydiard Fields Depot, with prizes donated by the management of Homebase, BCA Swindon and his colleague Jim Church. There was also a cake made and donated by Lorraine and her mum, Wendy Peart. The total raised was £378.00.

Thanks again to Neil who, with the help of Lorraine's step-daughter, Josie and her niece and nephew, Sophie and Tom Titcombe, held a sponsored dog walk around a local beauty spot, Coate Water, on a very cold, windy day, raising £264.00.

Lorraine and Neil hosted a Hallowe'en themed fancy dress party as a way of thanking all of their family and friends for the support they have received over the previous two years. The guests were also asked for a donation, raising £120.52, although Lorraine says 'the value of their support is priceless!'



Val Ridley, grandmother of Lucy Mathieson, worked tirelessly at three different events to raise a total of £1120.00. Val organised a table top sale, a Hallowe'en party and a coffee morning with delicious cakes and pastries made by Lucy's Aunt Dora Tye, and her good friend Theresa. Running three events in one week would not have been possible without the help of family and friends, who rallied around to provide prizes, homemade cakes and much more! A big thank you to Karen Maddison from Tiny Talk UK, who patiently organized games for over 60 children at the Hallowe'en party!



Gwen Clark, 81, did leaflet drops to promote the coffee morning she organised in Newbury, raising £152.00. Gwen and her friends made all of the cakes and snacks themselves, and ensured that all who attended had a thoroughly good time. Well done!

Beth Henshaw launched an Awareness campaign at the Railway Pub in Bolton, informing all of the staff and customers about NPD and encouraging them to buy a wristband, so far raising £60.00. This is ongoing and Beth has assured us there will be more to come!

Wedding Congratulations!

Tom Brooks, a Trustee of the Group, married Alison Margetts on 5th November at Huddersfield Town Hall, with the reception at Hey Green Hotel in Marsden. Tom and Alison met at university. Tom's Aunt Roni Brooks donated £1000.00 to the NPDG (UK) on this joyous occasion.



Pat Dixon and the committee, including Miriam, Winnie, Betty, Joyce and Joe, from the Doxford Park Community Association organized a pie and pea supper and a raffle, raising £164.00

Sheila Simpson and Betty Harris raised £52.00 for Niemann-Pick Awareness. Sheila and Betty spent many a long hour counting the whole £52.00 in 1p and 2p pieces.

Flora and Graham North held a teddy raffle for Niemann-Pick Awareness Week and raised £100.00

Mrs Louise Meade of South Shields has raised awareness of Niemann-Pick Disease and £87.00 with the new NPDG (UK) wristbands.

Mr and Mrs Godfrey sent the charity a donation of £500.00 towards the cost of running the group and the groups continued work towards finding a cure for Niemann-Pick.

Marnie Rutson and 8 year old Madison took part in the Hydro Active 5km run in September. This was Madison's first 5km run and she thoroughly enjoyed it. Marnie and Madison raised £95.00 in sponsorship.

Recipe Book

If you would like to send in your favourite recipe, there is still time to include it in our recipe book. There will be different categories, such as

- Mums / dads favourite
- Wicked pud's
- Children's favourite
- Fund raising favourites
- Cheats special

So please feel free to send in more than one! It would be nice if you could enclose a photograph to

accompany your recipe. We would like the book to be ready in time for the 2006 conference, and hope to sell the book at future fund raising events. Closing date for recipes - 1st March 2006

Janice Brooks

Fundraising Co-ordinator

Telephone 01452 311880

Email: brooks_janice@hotmail.com

The Great North Run



Stewart Mathieson, his colleagues and friends took part in the Great North Run on 18th September 2005. This very popular half marathon starts in Newcastle and finishes in South Shields. This grueling thirteen mile 'fun run' was completed in unseasonably high temperatures. The team members were Stewart Mathieson, Andrew Taylor, David Bradshaw, Alistair Chalmers, Alison Lambert, Andrew Spence, Spencer Glanville and Adam Parker. Altogether the team has raised £3277.50 in sponsorship and spent a lot of time encouraging their sponsors to gift aid their donation, which means that the charity can claim back 28p in every pound.

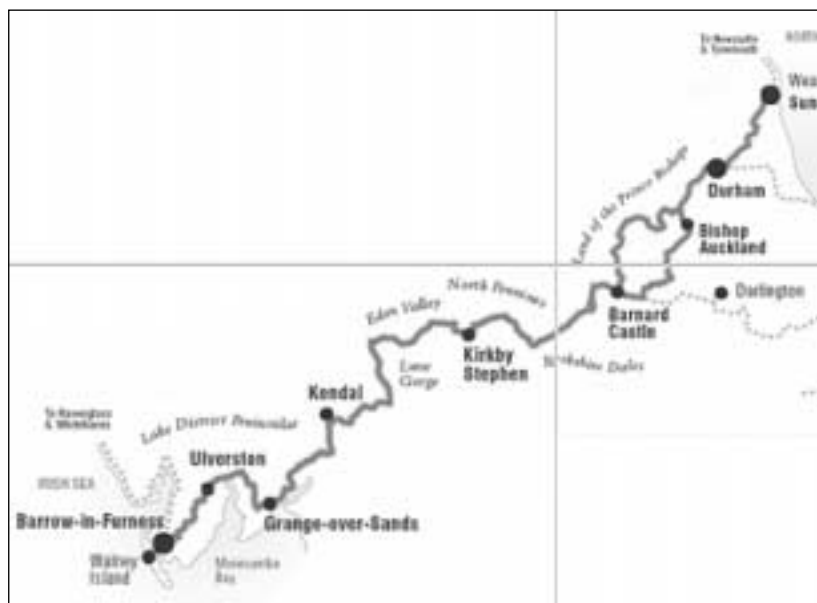
Well done to you all!

W2W – Walney to Wear Cycle Route

Michael Ross from Sunderland along with 9 friends completed the W2W bike ride in July 2005.

Michael writes

'After hearing about this terrible disease and its heartbreaking consequences, a group of friends from around the North East decided to try and make a small difference by undertaking a sponsored cycling event from Walney Island (Cumbria) to Sunderland (Tyne and Wear). This is known as the W2W, lasts 151 miles if you don't get lost, and is regarded as challenging as the infamous Coats to Coast ride. So between the 8th to the 10th July 2005, Stuart Metcalfe, Phil Jones, Gary, Chris Bourn, Barry Stafford, Dave Hodgson, Jo Bainton, Lee Raine, Eric Bolton and I set out to conquer the W2W bike ride.



Day 1 (15:30) – Walney Island

(0 miles). This was the start of the ride and everyone felt fit and well prepared. This optimism often appeared misplaced on numerous occasions over the next two and a half days but with the sun shining down and the Irish Sea behind us we set off.

Day 1 (20:00) – Kendall (42 miles). This was the location of our first night's hotel and all during the previous 42 miles this was where our well deserved pint was located. Needless to say everyone slept very well that night.



Day 2 (13:00) – Kirkby Stephen (74 miles). The proceeding morning had taken us through some of the most beautiful countryside in Cumbria and we arrived in Kirkby Stephen ready to eat a horse. Unfortunately this wasn't on the menu so we had to make do with a pint and a sandwich.

Day 2 (15:00) - Tan Hill (85 miles) – Tan Hill is the location of the highest pub in Britain some 1732 feet above sea level. Even though this was one of the shortest stages of the ride some of the banks that were encountered would test the fittest cyclist. Upon arrival at the pub some serious re-hydration was called for.

Day 2 (18:00) - Barnhard Castle (101) – This was the location of our second night's stay. Considering we were in such an old and historic town we decided to



have some traditional cuisine. This took the form of Indian food and Indian Lager. Unfortunately sleep was not as easy to find that night due to the severe muscle aches experienced by most of the group.

Day 3 (15:00) – Hamsterly Forest / Durham (133 miles) – This stretch was one of the most exciting parts of the ride. After coming through Hamsterly Forest, where we had 2 casualties (Nothing too serious) and saw a mother deer and her fawn, we could almost smell the North East Coast line. This gave us added motivation and spurred us on through the next few gruelling miles.

Sunderland (151 miles) – Home at last. The North Sea looked extremely inviting however looking at the blue coloured children playing at the waters edge we decided to cool ourselves down with a well deserved pint instead.

The total money raised by the group was around £1188.50. The ride was hard and not all of the riders completed it unscathed however to think that this would make even a small difference to the children and families affected by Niemann-Pick Disease made the whole thing worthwhile'



Ernst and Young

Ernst and Young, Citygate, Newcastle Upon Tyne, held a Charity Auction on Friday 23rd September raising £2220.00. Throughout the year the partners and staff at Ernst and Young have held many fundraising events to benefit the NPDG (UK) and have raised an amazing £12,653.34 in total!

Tony Jellings writes

'Readers may remember that my ten year old grand daughter Lisken some years ago on two different occasions designed very successful Christmas cards for our charity, both of which sold out. She attends Firle primary school in Sussex. Recently her local vicar came to address her class and asked the children what they would do if he was to give them five pounds. With one exception all the children had definite ideas on how they would spend the money - on themselves. Lisken replied that she would give the money to a charity, to

the Niemann-Pick Disease Group (UK). The vicar handed her the £5 note. I was so pleased and very proud of her.'

Christmas Cards 2005

Donations

This year nearly £300.00 was donated to the charity in exchange for Christmas Cards. As the charity does not have a trading arm, we are not allowed to sell any items, so we have to recommend a donation amount of £1.00. Those of you who decided that you would like NPDG (UK) Christmas cards have been very generous and helped raise a great amount of money and almost depleted the stock. We now only have a small amount of the Sheep in Snow design left.

We would also like to thank everyone who happily accepted a different order from us, as we sold out of the Children Playing in Snow almost straight away and the Christmas Flower followed shortly after.

Competition Time

Christmas Cards 2006

We are already planning the Christmas Card Range for 2006 and we are running a competition open to Family and Friends aged 16 and under.

If you are a budding artist or you have a family member or friend who likes to paint or draw, let them know and get them to take part. If their picture is chosen it will be sent out to hundreds of people around the world next year!!

We will be judging all of the entries at the Family Conference 2006 in June and the winning entries will be made into Christmas Cards.

If you would like to enter the competition, send your picture on a piece of A4 paper with your name, address, telephone number and age to:

Toni Mathieson, NPDG (UK), 11 Greenwood Close, The Pastures, Fatfield, Washington, NE38 8LR

Closing Date: 1st May 2006

NPDG (UK) Calendar 2007

The Niemann-Pick Disease Group have decided to produce a calendar for 2007 and we will be launching a competition, open to all ages, for each of the 12 pictures that will feature as each month. The picture can be of any media, for example: a photograph, a sketch, a painting, or a photograph of an art piece such as a sculpture you have made, or a piece of needlecraft,

the choice is yours. All we ask is that it represents the season you have chosen.

The competition will be divided into 4 age groups:

Age 0-7 years

Age 8-12 years

Age 13-18 years

Age 19 +

From each category three winners will be chosen, one for each season, Spring, Summer, Autumn and Winter.

Once the final 12 pictures have been chosen, one overall winner will also be featured on the front cover of the calendar.

We will be judging all of the entries at the Family Conference 2006 in June and the winning entries will be made into the NPDG (UK) Calendar 2007.

If you would like to enter the competition, send your picture on a piece of A4 paper or a photograph of your work with your name, address and telephone number to:

Toni Mathieson, NPDG (UK), 11 Greenwood Close, The Pastures, Fatfield, Washington, NE38 8LR

Please also state which season you have chosen and which age bracket you are in. You can enter a maximum of 4 pictures, one for each season.

Closing Date: 1st May 2006

Want to get Involved?

Niemann-Pick Wristband - limited availability!!



The Niemann-Pick wristbands have proved to be a valuable tool in our campaign to raise awareness of the disease, and at the same time raise much needed funds. The bands are periwinkle blue and have 'PERSEVERE' on one side with our website address on the other. There are a limited amount left, in both children's and adult sizes. If you would like a band or would like to use them to raise funds for the group, they are available at a suggested donation of £1, and to help us with costs, it would be very helpful if you send an SAE with your order.

If you would like to support the work of the group by helping us raise funds with the bands, please contact Toni on 0191 415 0693 or email niemann-pick@zetnet.co.uk

Helping us while you Work

The government is promoting a new scheme to increase Payroll giving throughout the country and your company or place of work could become part of it!

Grants of up to £500 are being offered through the Payroll Giving Scheme to encourage companies to start up this form of giving direct from their payrolls. What this means is that your employer will deduct your donation to your favourite charity – hopefully the NPDG(UK) – direct from your pay, before tax, and send it on to our bank account! Because it comes off your salary before tax, it means that, if you choose to donate £10 a month to the NPDG(UK), it only costs you £7.80 as the tax is passed on direct to us as well.

Many companies also match this form of giving so your £7.80 could become as much as £20!. It really is a good way to make regular donations to the NPDG(UK), allowing both you and us to plan regular spending. Every year we calculate the amount of income we will have to provide services to families with NPD and knowing what will be coming in from regular giving really makes a difference to these calculations.

Why not suggest that your employer gets in touch with the Payroll giving Hotline on 0845 6026786 where the Payroll giving Campaign team will explain how they can get it set up and claim their £500 grant.

To those supporters who currently give in this way, many thanks for your continued regular giving, it is much appreciated!

Fundraising Packs

Although Awareness Week is now over until next year, fund raising events continue throughout the year. We are always available to help in the organisation of events, however large or small. We can provide advice on holding many different types of event and can supply you with a fundraising pack, containing; a

t-shirt, stickers, a poster, an ideas sheet, collection boxes, information leaflets and a sponsor form.

Please give us a ring at the office 0191 415 0693 and we will pop one in the post. Alternatively drop us an e-mail at: Niemann-pick@zetnet.co.uk

A big thank you to everyone for your wonderful fundraising efforts!

We would like to thank everyone else who has arranged events, taken part in activities and sent donations for the group. We would love to feature you all, but sadly we just don't have the space. We do, however, really appreciate all of the time, energy and enthusiasm you show, by taking time out of your busy lives to help us continue with the work that we do.

Benefits News

Grants for specialist equipment

Children Today Charitable Trust provides grants for specialist equipment to give disabled children greater independence and the chance to take part in the same sorts of activities as others. Examples include electric wheelchairs, educational toys, communication aids and specialist computers, anything that will improve and enhance the quality of the child's life.

Parents need to apply direct. A Grants Panel looks at all applications and decides whether they can give any financial assistance, up to a maximum of £1000. Best of all, you get a prompt response as the Panel normally meets fortnightly.

Ring 01244 35622 to request an application form or apply online at their website www.children-today.org.uk

Benefits Fact Sheet Updated

The Contact a Family Fact sheet "An Introduction to Benefits and other Financial Help" has been updated to reflect recent changes. It explains the range of benefits and financial support that families may be entitled to, as well as details of other organisations that may provide financial assistance.

You can download a copy from

<http://www.cafamily.org.uk/benefits.html>

Or ring the Contact a Family Helpline on 0808 808 3555

No means testing for Disabled Facilities Grants in England.

The Government has just announced that, from December 2005, families in England needing to adapt their homes to care for a disabled child (and some young persons under the age of 19) would no longer be subject to means testing.

Disabled Facilities Grants can meet the costs of disability improvements to the home, including access ramps, stair lifts, level access showers and home extensions. The grant can be up to £25,000. At the moment, and until the means test is abolished, families resources are taken into account when assessing how much grant is payable and many families are forced to take out loans to cover the costs of works. Families who are about to, or are in the process of

applying for a DFG, can ring the Contact a Family Helpline (0808 808 3555) to discuss the impact these changes may have on their own situation.

The announcement quickly followed the publication of an independent report on DFGs, commissioned by the Government, and a joint review by the Office of the Deputy Prime Minister, Department of Health and Department for Education and Skills of the overall scheme is still under way.

For more information ring the Contact a Family Helpline 0808 808 3555

or e-mail info@cafamily.org.uk

Carer's Rights

Friday 2nd December 2005 was Carer's Rights Day. The focus was on getting carers to claim the benefits they are entitled to. According to a report published by Carer's UK they are currently losing out on £746 million in benefits each year.

These entitlements are vital to help meet the costs of caring, and can be gateway to other benefits such as a discount on Council Tax. High on the list of problems cited by carer's is the financial impact of leaving work early because of caring. Paid employment makes up nine per cent of a pensioner's income, underlining research which shows that the longer a person stays in work, the better their finances will be over the long term. This means that carer's who give up work to care are missing out on an

important source of additional income as well as the social benefits of the workplace

Carer's Rights Day aims to raise awareness of the needs of carer's with policy makers and professionals.

Carers UK has produced a Carers Rights Guide for all carers, You can get these guides free by visiting their website www.carersuk.org or by calling the Carers UK on 0207 4908818

David Wray wrote this poem as an introduction when he was asked to do a presentation (to a group of health professionals) which would highlight the difficulties carers face. It expresses the feelings of isolation and desperation that he felt as a carer. It was used with a series of questions to stimulate a discussion on care issues.

The Prisoner.

From my childhood
I remember a scene in a history book.
A man in chains sits in the centre of a cold flagged floor,
and as if on stage,
is encircled by a pool of light,
which streams into the gloomy cell from a small window, high above.
The thin, tapering cone of brilliant light
is all that links him with the outside world.
Its soothing warmth thrusts aside the cold darkness of the room
and reminds him of the other world that once was his;
a world of freedom, enjoyment and friendship.

But now I am there, for I am that person in the room,
imprisoned not because I have erred,
but because I choose to care.
The picture in my mind is now my reality:
For I am a carer.

D. Wray

Family Focus

Baby Congratulations!

Andy and Sarah Forrester are pleased to announce the safe arrival of Ben Alexander, born on 1st December, 2005 weighing 8lb 11oz. Mum Sarah has NPC.

Baby Ben and all the family are doing very well.



My story about Jade by Shawreen Van Aarde

Given at the Family Conference 2005

Let me first introduce myself. I arrived in England in November 2003 having immigrated from Zimbabwe in Africa. I have lived in Brighton ever since then and my whole family have since come across.

My Daughter Jade had Niemann-Pick Type A. When, after seven years of marriage our first daughter, Alicia was born she was special. Then 2 _ years later when our second daughter, Jade, was born by emergency caesarean on 29th March, 1992 we felt doubly blessed. Weighing in at 8lbs 1oz she seemed to us the most beautiful child ever, and we felt as parents do at this time, that our child was someone special. We called



her Jade which means a precious stone and that indeed she was. It was actually ironic that Jade nearly died at birth from high stress levels and the doctors were amazed at what a fighter she was and her determination to live. So in view of this, I later found it harder to cope with her death as I felt life had dealt us an unfair deal.

At 5 months, Jade started vomiting and this carried on for days. Eventually this became an ongoing pattern that every two weeks or so, she would vomit for a few days. After several blood tests and many hospital visits it was thought that she might have Leukaemia, which is every parent's nightmare. However, after many months of living in hospital and doctors rooms, we were sent to South Africa where we were advised that it was probably Gauchers Disease. However she still was not improving and, after a second visit to South Africa, she was diagnosed with Niemann-Pick Type A. For the first time in my life I felt that life was unfair and cruel because I wanted my daughter. I felt as if my heart had been wrenched out of my body when we were told the soul destroying words "There is no cure".

I was too speechless to cry or show any emotion and, only when I walked out of the doctor's surgery did I give way to all kinds of emotions, not the least of them being anger and hopelessness.

I was very fortunate to have corresponded with a Professor in America who was associated with Niemann-Pick and regret now that I did not bring one of his letters with me when I immigrated here. He was very encouraging that a cure was in the future, but unfortunately it was no consolation to me. Jade was on oxygen by now and the end was near as she was developing little sores all over her body and her stomach was like a round ball and quite hard.

She developed pneumonia and, 6 days later, on 17th March 1994, Jade died - just 10 days before her second birthday.

You never "get over" the loss of a child but somehow you learn to cope with it. The hardest thing I have ever done in my life is leave the grave of my dear Jade so far away. The day we went to the grave for the last time has to be one of the most gut-wrenching days of my life and the feeling of loss was heightened by the fact that I knew this was probably the last time I would ever visit her place of rest. I do still have lots of bad days even though it is just over 11 years ago and some days the fact that she is so far away nearly drives me crazy, but I have another child, Alicia whose future I had to consider and bringing her to England was the best thing I ever did as she has grown from strength to strength.

Rare condition mimics asthma

WHEN Lorraine Bishop couldn't stop wheezing doctors thought she had asthma.

Instead, she had an extremely rare condition called Niemann-Pick Disease.

Only 60 cases have been diagnosed in the UK.

This week is national Niemann-Pick Awareness Week and people are being encouraged to swot up on the potentially deadly condition.

Lorraine, 38, of Harcourt Road, off Ferndale Road, was diagnosed with Type B Niemann Pick Disease in August last year.

"The symptoms are very similar to asthma," said Lorraine, who lives with warehouse operative husband, Neil, 41.

"I get very short of breath and I get a sharp pain in my back.

"There is also a lot of wheezing – particularly at night when I lie down as I tend to prop myself up with pillows.

"Some days are worse than others.

"I have no way of knowing how I'll be feeling the next day.

"I use an inhaler when the symptoms are bad, which does help a little.

"When the symptoms turn into an infection I get prescribed an-

'I can't plan because I never know how I am going to feel'

tibiotics but on a day-to-day basis there is not a lot I can do."

Lorraine, who is not able to work because of her condition, said: "In 2003 I was given antibiotics – they thought it was some kind of chest infection."

"Then in the October I was told I had pneumonia and told to be patient.

"The symptoms just wouldn't

go away, so in 2004 I was sent to London for tests. They took a lung biopsy and in the August I was given the diagnosis."

The condition causes a build-up of white, elastic-like phlegm called casts, which are coughed up.

Lorraine attempts to control this by having regular "jet-washes."

"The lungs are literally given a kind of jet-wash," she said.

"The phlegm gets stuck in the airways.

"Niemann-Pick Disease has had a huge impact on my life.

"Initially, because the condition was so rare we had no idea what it was. Even the doctors had to look the condition up in the medical books.

"My husband and I can't make any plans because we just never know how I'll be feeling."

● Anyone wanting further information can visit www.niemann-pick.org.uk.

What is Niemann-Pick Disease?

NIEMANN-PICK Disease is actually a term for a group of diseases which are caused by specific genetic mutations.

These affect the metabolism and prevent suffers from using cholesterol properly. It requires a gene from each parent, which is what makes it so rare.

It is named after Germans Albert Niemann and Luddwick Pick who provided evidence of it. The three most commonly recognised forms of the disease are Types A, B and C.

Type A is the acute infantile form, Type B is a less common, chronic, non-neurological form, while Type C is a biochemically and genetically distinct form of the disease.

Type C usually affects children of school age, but the disease may strike at any time from early infancy to adulthood.

The condition is progressive.

The first symptoms include:

- Poor balance
- Deterioration in motor skills
- An inability to move the eyes

up or down quickly.

In children, symptoms include:

- Enlarged spleen or liver
- Jaundice following birth
- Unusual shortness of breath
- Repeated lung infections
- A cherry red spot inside the eye
- Progressive loss of early motor skills
- Feeding and swallowing difficulties
- Learning problems
- Sudden loss of muscle tone.

From the Swindon Advertiser.

And This My Child

Submitted by Lisa Chavez in honour of her daughter, Breann ~ age 2, NPC.

I have waited for your arrival sweet baby and now you are finally here. I hear your first cry and see your beautiful face with cheeks as soft as an angel's kiss. I count your tiny fingers and toes and realize that the Creator Himself has given me a precious gift. I hold you close and feel your warm breath against my neck and as I do I feel your tiny heart beat against my own. The waiting and longing for you is over. You have captivated my heart in the first few moments of your birth. My life has been made complete with your arrival. This my child is called LOVE.

I cannot believe how my life has changed since you came into our family. I watch you roll over and scoot across the floor. You call for your kitty and give her a hug. Your smile could melt the coldest of hearts and I believe that nothing could possibly make me happier than I am at this moment. I pick you up and dance

with you in my arms. I never knew that the song was so beautiful or that my arms had been so empty. Then I realize that I never had you to sing this song to and I know that this my child is called JOY.

I watch as you play. Should you be saying some words? Should you not be walking? I mentally brush away these thoughts whenever the questions surface. I can do that for only a short period of time. We see the doctors, they run their tests and I stand by feeling so helpless. What could be wrong? In time these issues will be resolved and you will do these things and more. On some days I want to know - I want the doctor to answer my questions. For a moment my heart is gripped by a new feeling which my child I admit is FEAR.

The doctor's call comes. I can hear her voice but certainly the words she is speaking are meant for

someone else. She tells me about a disease called Neimann-Pick. How could anything that sounds so sinister possibly be invading your body. The call must be for someone else - it cannot be meant for me. I am bombarded with information that my mind cannot absorb. It is not so - I can see you are learning and growing. I know they are wrong, everything will be OK. My heart is pounding and without even thinking I cover my ears. I need to shut out the voice and resolve that I will not speak the word for along with the diagnosis comes another feeling and that my child is DENIAL.

The clear blue sky and warm breezes call for us to come and play. I carry you to the sandbox - your legs too weak to walk on their own. We call for the ducks to come and be fed and watch as the fish jump in the pond. Your lips do not form the words but I know that inside you are speaking. Your eyes and smile say so much to me and I realize it's the language of the heart through which you speak for I can hear all your say. The bluffs loom in the background the pond reflects the shade trees that we sitting under. We lay next to each other on the hammock and I allow my mind to wander as aimlessly as the clouds that float overhead. Maybe today you will take a few steps on your own. Maybe this will be the day you say Mommy or Daddy. Maybe - maybe. This my child is called HOPE.

I want to stay in this position forever. Covers pulled tightly over my head, my body rigid I long to stay in this foetal position. I cannot move. I can hear the sounds of the early morning but my heart holds no song like the birds singing outside my window. The melody and song in my heart have disappeared. I can barely tell when a day begins or ends - they all seem so endless. How many nights do I lay awake, willing myself to sleep yet I am robbed of life being normal if only in dreams. I cannot imagine anyone else ever feeling like I do. I am depleted of energy. The pain that grips my inner most being pounds like angry waves hitting the shore. Your little hand reaches out and touches my face. The deep brown pools of your eyes and your sweet little giggle makes me realize I too am smiling. I see you are ready to begin your day even though I long to stay here forever. How can it be that one so young is teaching me life lessons? Alone as I may feel I know that other families are facing the same struggles I am. Perhaps they are at a far different stage than I but none the less they are struggling with broken dreams and unexpressed fears. Questions cloud their mind and anger may rise within their hearts like a volcano spewing forth it's hot lava. I recognize that what is affecting our family has moved beyond the walls of just our home. Our extended family, loving friends and caring therapists are all

players in this story of your life. With an unexplained heaviness in my soul I will myself to emerge from the cocoon I have built around myself. We will play and read books and take a ride in your wagon. The ducks will come to eat from your outstretched hand and we will watch as the butterflies dance about the yard like tiny little fairies. I know that in the face of insurmountable circumstances I will make it through this valley and I will come forth a different person than before. I will be far stronger than I was because I have pulled from within myself courage that I never before knew I had. I will have learned compassion for others for I will have walked in their very shoes. I will have learned that the greatest lessons in this life come with a high price tag. My faith may be shaken, my heart may be scarred and bruised, my dreams may for the moment seem illusive, discouragement may be part of my life yet within me remains the steadfast faith I know I need to survive this hideous disease. Suddenly, just as the first rays of light peek through my window I realize that this my child is called PERSEVERENCE.

This article appeared in the NNPDF Fall Newsletter, and has been reproduced with their kind permission.

Walk with me ... to all parents ... you're not alone ... lets walk

Walk in my shoes

for one single day

Then you'll see why

I need to pray.

Come live in my home

for a week or two

and then remember

I'm just like you.

I didn't ask for a thing I was given

I didn't choose this road I have taken

Walk a mile with me hand in hand

then perhaps you will understand.

I'm not really complaining about the stress in my life.

I know that we all have some toil, some strife.

But walk with me, walk with me, please just walk with me

understand my life

By Lisa Bushaway

Emily by Emily's Nana

*I have a special place where I keep my jewels
 There are eight of them in all
 Some of them are pretty big
 Some are very small
 They were taken from two lovely crowns
 and just given to me
 Some are very bright, others sparkle and shine
 Some are dark in colour, others light and fine
 But they're all mine
 One of these jewels has a flaw
 You can't see it, but it's there all the same
 But it sparkles and shines so brightly
 It dazzles your eyes with it's flame
 All of my jewels are precious
 They are with me wherever I go
 But that one little jewels with flaw
 Lifts my spirit and makes my heart glow.*

Special Child

To Emily

*Why are you looking at me
 Do you think I cannot see
 Why not come and have a talk
 I'd come to you but I cannot walk
 I notice that people sometimes stare
 I look right back, I don't care
 If you wonder why I'm poorly
 I could tell you quite a story
 I used to be just like you
 Running, jumping, shouting too
 Then along came NPC
 And decided that it wanted me
 But don't look sad, I'll tell you true
 I'm a special child and see more than you*

By Irene Bushaway



A Parent's Perspective

David and Sally Wray have produced a booklet called **"Caring for a Child with a Degenerative Disorder: A Parent's Perspective"**, which is a summary of the skills, knowledge and understanding needed to look after a child with a genetic, degenerative disorder (David and Sally's son Andrew had NPC).

David and Sally write:

'When a child is ill, the parents are the front line personnel who carry the main burden of care; they look after the child for the greater part of the 24 hour period, seven days a week, month after month. For parents whose child has a static condition, the task of understanding the illness and organising the support is a largely one-off exercise, given that there will be changes as the child matures.

For the parents of a child with a degenerative disorder, no sooner have they adjusted to the existing situation, then they have to start again, organising services, finding equipment, improving their understanding of the condition and discovering how to deal with new symptoms. A major problem for them is gaining access to information.

Much of the information they are given is provided in a haphazard manner, on a need-to-know basis and is therefore in a fragmented and limited form. This is not conducive to good learning and will not result in the desired outcome of a better understanding of the care processes.

Community Health Planning seems to assume a large professional training and care input. The reality is that input is limited and the parents carry much of burden relying on intuition, creativity and common sense.

From the perspective of a professional specialist, with their extensive field of knowledge, it may appear that the proportion of information required by parents to care for their child is quite limited. However, this overlooks the fact that the parent carers are generalists; they are hands-on practitioners with their own body of knowledge consisting of elements drawn from many different disciplines. This does not make it any less academically valid or less arduous to acquire; it is an essential body of knowledge without which they cannot function effectively.

Much of this information already exists in different forms. It simply requires putting together as one information source set within an agreed set of parameters. It is extraordinary that such a simple editing exercise, which would make such a huge

difference to the lives of carers and sick children, has not been done before! This paper is written in the hope that it might be a starting point; the first stage of a longer process in which the themes are expanded and developed by others to form a series of more comprehensive fact sheets and information packs'

This paper can be found on the Contact a Family Website at:

<http://www.cafamily.org.uk/CaringForaChildwithaDegenerativeDisorder.pdf>

David and Sally have also written **'Louise Wray – Sibling of Andrew'** describing the impact on siblings of having a brother or sister with a life-limiting illness, and looking at how they, as a family were able to deal with the everyday challenges it presents.

David and Sally write:

The impact on siblings of having a brother or sister with a life-limiting illness is frequently overlooked. This is largely because the focus of attention is on the ill child in an attempt to provide and maintain as good a quality of life for them as possible.

Many of the difficulties encountered by the parents will also have an impact on the children. Like them, the siblings also suffer a restriction of freedom, gradually "lose" a brother or sister with whom they still live, have to accept additional responsibilities, do not have a "normal", (as perceived by their friends) home and life-style and have to deal with the problem of the death of a family member.

For the siblings however, life still goes on. For the younger children particularly, they are at a formative age and there are future consequences to their current experiences. The way in which they are guided through the unfolding events will shape their reactions to future circumstances; opportunities taken or missed now will return later to influence the child's adult world.

It might also be regarded as a parents' tribute to Louise; recognition of her patience, trust, steadfastness and bravery.

If you would like a copy of either paper sent to you by post or email, please contact Toni Mathieson at the NPDG (UK) office, 0191 415 0693 or email niemann-pick@zetnet.co.uk

Dads' Stories.

Contact a Family has been commissioned to pull together a book of dads' personal experiences of parenting a disabled child. At present there are no books giving the perspective of UK dads of disabled children. If you are the father of a disabled child, they would really like to hear your story for inclusion in the book. They would be interested to hear anything about your family situation and how you dealt with it – how you found out about your child's condition, the process of diagnosis, the kind of support that you got, or didn't get. Top tips for other dads or for people who want to provide services to dads would be great.

Either:

- write your own story (anything from a few hundred words to 3,500 words is fine) and send it to Contact a Family (Dads) 209/211 City Road, EC1V 1JN e-mail: <mailto:dads@cafamily.org.uk> ;
- or if you prefer not to write but would be happy to chat in person or over the phone, they can arrange for an interview at a time to suit you, either e-mail: <mailto:dads@cafamily.org.uk> or call their dads worker, Matthew Henderson Tel: 0808 808 3555 Mon-Fri, (10am to 4pm) to arrange a time.

Lone Parent Guide

More than a quarter of lone parents have a disabled or ill child.

A guide called The Lone Parent Guide to Caring for a Child with Additional Needs has been produced by One Parent Families. This guide covers a range of topics including financial help, coping with caring for a child on your own, education, help from social

services and travel and holidays. It also tells you where you can get further help and information. The guide is available free to one parent families by calling 0800 018 5026 and is also available on their website in five community languages.

<http://www.oneparentfamilies.org.uk> .

Thank You!

Many thanks to the families with Niemann-Pick disease who have willingly given up their free time to talk to journalists, allowed television crews into their homes and who have spoken so openly about their own personal situations. This is a very valuable way of helping the NPDG (UK) to raise awareness and get

stories into the media. We do appreciate everyone's commitment and help.

If you would like to volunteer to speak to the media about your experience of Niemann-Pick disease, please contact Toni on the Helpline 0191 415 0693 or email niemann-pick@zetnet.co.uk

Child Bereavement Trust

The Child Bereavement Trust has launched its new redesigned website. The site has a forum for bereaved families as well as a discussion forum for professionals, and is at www.childbereavement.org.uk

In Loving Memory



We remember
Sam Roberts Age 4
David Rusby age 22
Leanne Colquhoun age 19



*Be still,
 Close your eyes,
 Breathe.
 Listen for my footfall in your heart.
 I am not gone but merely walk in you.*

~ Sibling Stories ~

Welcome to the very first 'Sibling Stories'!

Hello,

My name is Monica and I grew up with a disabled brother. When I was growing up, I really wanted to know about other brothers and sisters like me. I used to think that I was the only one. Now I work for Sibs, which is an organisation just for siblings (brothers and sisters) of people with disability or serious illness.

Siblings tell us that there are both good things and difficult things about their family life.

Many siblings say the hardest bit is not getting enough attention from their mum or dad. Parents have a lot to do taking care of their brother or sister – but it is still really important for siblings to get time with parents.

Some good things



- ☞ Having fun with your brother or sister
- ☞ Learning about medical things
- ☞ Getting to the front of a queue in a theme park
- ☞ Feeling proud of your brother or sister
- ☞ Being understanding about people who are different
- ☞ Helping your brother or sister with something.

Some difficult things



- ☞ Feeling lonely if your brother or sister can't play
- ☞ Not understanding Niemann Pick Disease
- ☞ Not enough attention from your parent
- ☞ Worry about the future
- ☞ People staring at your brother or sister
- ☞ Not being able to talk about your feelings

Some tips for getting attention



Ask your parent to spend some time with you each day – maybe when your brother or sister is in bed, or when someone else is looking after him or her



Make a list of things you would like to do with your parent and put it on the fridge – then when your parent has time you can do one of those activities together

Sometimes you might really want to talk to your parent when your brother or sister needs help with feeding or is crying – at these times write a short note to your parent about what you want to talk about. This will help you remember it and your parent will know it's important to talk about it later on.

Monica McCaffery is the founder and Director of Sibs, the only UK Charity specifically for siblings.

To contact Sibs - Email info@sibs.co.uk

Website www.sibs.org.uk

Tel 01535 645453

My precious sister

By Alicia Van Aarde

*The year was 1994 and it is now 2005.
 Time has passed and continues to pass!
 I can still see her lying in the hospital,
 I can still see her sweet face even though it was drained from the pain and of course the tears she
 cried,
 I can still see the drips through her nose and
 I can still see the anguish in her eyes and
 I can still hear the beating of my heart as it races with sadness and regret!
 I can still hear the doctors talking though I don't know what they're saying,
 Talk, talk, talk that's all I hear!
 Yes my precious sister is still fresh in my mind.*

*Jade, precious is what you are!
 You are never far, no never far from my heart!
 I keep you close ever so close.
 You are my precious sister and I love you!*

*My eyes are filled with green, showing my envy.
 My heart is filled with black, showing my lament.
 Why lament a state, which we should envy?
 Precious you were and precious you are!
 My precious sister!*

*In my heart and mind you are,
 Day in and day out!
 I long to talk to you and
 Tell you that I love you and
 Tell you that you're my precious sister!*

THIS POEM IS DEDICATED TO MY BELOVED SISTER JADE!



Special Brothers and Sisters

Stories and Tips for Siblings of Children with a Disability or Serious Illness

Edited by Annette Hames and Monica McCaffrey

Illustrated by Brendan McCaffrey

2005 96 pages ISBN 1 84310 383 4 £10.99

Special Brothers and Sisters is a collection of real-life accounts from the brothers and sisters of children with special needs, disability or serious illness, ranging in age from 3 to 18 years. They explain, in their own words, what it's like to live with their siblings.

There is a lot of advice available for parents of a child with a disability or illness, but very little about the important issue of educating their siblings about how they feel, and why they may behave differently from other children. These stories - from 40 different families - come with related tips to help siblings deal with some of the things that happen in their family lives. The book also provides a helpful glossary to explain, in child friendly language, the disabilities and medical conditions mentioned, including: ADHD, autism, cerebral palsy, cystic fibrosis, Down syndrome.

Special Brothers and Sisters is an engaging and educational collection that will enable young people and adults to share in the extraordinary experience of being a sibling of a child with special needs, a disability or serious illness.

Annette Hames is a Consultant Clinical Psychologist who has worked with children with learning disabilities for over 20 years. She currently works with families with children with learning disabilities for Northgate and Prudhoe NHS Trust, in Newcastle-upon-Tyne. Monica McCaffrey is the director of Sibs, the UK organisation for siblings. One of her brothers has learning difficulties and another brother died in childhood from a life-limiting condition. Monica lives in Yorkshire.

CONTENTS:

1. A note the brothers and sisters.
2. A note to parents.
3. Stories and tips.
4. Where you can get help.
5. Dictionary.

If you would like a copy of Special Brothers and Sisters, please contact Jessica Kingsley Publishers, 116 Pentonville Rd, London, N1 NJB. Tel 0207 837 2917

*If you would like your story or poem to appear in
'Sibling Stories', please contact*

*Toni on 0191 415 0693 or email niemann-
pick@zetnet.co.uk*

National Development Manager Report

As I write this I am surrounded by boxes containing the Niemann-Pick office. I have now been in post for almost two months, and I am just beginning to get organised! My main concern so far has been the set up of the office, and the establishment of the processes that will enable it to run smoothly and efficiently. I have enjoyed getting to know Michelle, and we have discovered the joys (and the hiccups!) of 'distance working'. Without the technical support of Michelle's very helpful husband, Dave, I doubt we would have got this far! I have also had fantastic support from the Board of Trustees, Jackie Imrie and my husband, Stewart.



I am still on a very steep learning curve, but the good news is that I am enjoying all of the challenges that my work brings. I have skipped from one demanding project to another, from completing my first grant application to preparing this Newsletter, which I do hope you find to be helpful and informative.

I have travelled to Scotland to meet with Jim and Susan Green, spending the whole day enthralled as I listened to the history of the Group - and eating too much shortbread! The following week I collected 'the office' from Richard Brooks, and we were joined by Bill

Owen and Michelle. I then went on to meet with Tanya Collin-Histed, who very kindly agreed to pass on some of her vast knowledge and experience. I have also visited Jackie Imrie at the Willink, and look forward to attending the Clinic Days in the New Year.

In the very near future I will be meeting with Bill Owen and Clive Roberts to talk about updating the Group's website, I hope to tell you more about this in my next report. I am also working with Richard Rogerson to promote The Peter Carlton Jones Memorial Award, which you will have seen advertised in this Newsletter.



I have very much enjoyed talking to some of you over the phone, please feel free to contact me for any reason. I hope to meet a lot more of you at the 2006 Family Conference, full details of which, will be available soon.

Over the next few months I will be developing a work plan to assist me in furthering the objectives of the Group and continuing to explore the work of other disease groups and organisations. With the help of the Group's Fundraising Co-ordinator, Janice Brooks, I will be developing new fundraising initiatives and assisting Zoe Meade-Wynne in organising the Conference. It is going to be a very busy time, but I am certainly looking forward to it!

Jackie's Journal

December 2005

Hello everyone.

By the time you get this you will all hopefully be recovering from Christmas. I hope you all had a good one and aren't too exhausted.

This year seems to have flown by and has been as busy as ever.

The most recent outing was to Liverpool where I joined with Colin Cosgrove from the Alzheimer's Society and Susan Green to raise awareness about Dementia in young people and children. In Alzheimer terms young people are classed as under 60 but as we are all aware we have young children that do suffer from Dementia as part of NPC and we need to make provision for them and be able to convince teachers in schools that children are not just "being naughty"! As you can see from the photo Colin survived the day, despite doing the organising of what was an excellent conference. You know conferences are good when 250 delegates arrive in the morning and most are still there for the closing comments.

On the subject of conferences, our next one (June 24th and 25th) is well into the planning stages with many speakers already confirmed. We do need to know what you want to ensure a really good meeting for all, so please do tell us. Should you wish to attend and need support, whether that be hands on, transport, financial or anything do call me and I will assist where I can.

We are hoping several new families will be coming this year and we know how daunting it can be to meet other families for the first time. If you are one of these families please let me know beforehand and we will support you as much as you wish. For those families who have been before and know how



that first time felt, please could you let me know if you would be willing to befriend a new family prior to the conference, so that not everyone will be a stranger.

Talking of new families, we have had many new children and adults diagnosed in the last year and several have asked to be put in touch with other families. If chatting to new parents is something you feel you would be happy to do, please could you let me know.

For now I will go out and face the snow that is falling heavily otherwise I may be here in the office overnight, and although I love my work, I don't want to sleep here.

If there is anything at all I can help with, please don't hesitate to call

Jackie

Jackie Imrie
Clinical Nurse Specialist
Niemann Pick Disease
Phone/fax: 0161 922 2414
Mobile: 07932 737417
Email: Jackie.imrie@cmmc.nhs.uk

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 28th February 2005

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