

Niemann-Pick News

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

Spring 2008

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Chairman's Chat



“Care Matters”, the theme of this year’s conference, which is to be held on 28th and 29th June at the Hilton Hotel East Midlands Airport, is at the heart of everything we do as a Charity.

Where would any of us be if “care” didn’t matter? In the middle of our busy lives, driven on by a whole range of different priorities, Niemann-Pick reminds us, most forcefully, that care does matter. Care for each other, care for those who have the disease, care for the carers, care for the clinicians and care for the scientists.

The core objectives of our Group are Care and Support, Information and Research – the Clinical Nurse Specialist, Jackie Imrie, who is funded by the Group, is a key member of our support programme; the Clinical Research Nurse, Liz Jacklin, also funded by the Group, is involved in direct clinical research work; Toni Mathieson and Sue Lowe, our Development team, work hard to provide up to date accurate information (such as this newsletter and many other publications). These people, along with the Trustees and many other volunteers are there because “Care Matters” and because so many people throughout the country, and beyond, donate money, time and energy to making a difference.

I hope you all enjoy the newsletter and reap some of the results of the fact that care certainly does matter

Jim Green

Front cover picture: Clacton Fire Brigade's Car Wash, in aid of *Hope for Hollie*.
See page 12 for full story.

Research News

Research Nurse Update



The post of Clinical Research Nurse for Niemann-Pick Disease was created in 2006 and is fully funded by the NPDG (UK), initially for a two year period. The good news - for me at least (and I hope for everybody else!), is that the NPDG (UK) has been able to extend my contract for a further two years. As well as continuing to concentrate on research in the field of Niemann-Pick diseases, my role will now be to assist the Clinical Nurse Specialist, Jackie Imrie, in providing an increased level of family support.

Research is an exciting process, but at the same time it can often be slow, frustrating and disappointing. I think that during the past two years we have all, at times, experienced these emotions. We want answers and treatments as quickly as possible – but these are held back for a variety of reasons.

As you may be aware, the clinical trials of Miglustat (registered name Zavesca, previously known as OGT 918) have recently come to end. Whilst we knew this was going to happen, for those who have been involved with the study for such a long period of time, the last few months have been a difficult and uncertain time. The good news is that all the patients involved in the study who wish to continue taking the drug will be able to do so. We will not know the future

of the drug (whether it will be licensed) for a few weeks yet.

The Phase 1 clinical study of rhASM for Niemann-Pick Type B, continues in the USA. The study, conducted by Genzyme Corporation, is still recruiting adults with NP-B to participate in a single dose study of lab-created enzyme rhASM to be administered at Mt, Sinai hospital in New York. An update on the progress of this trial can be found on page ?? of this newsletter and we hope to once again welcome Dr Margaret McGovern (lead clinician for the trial) to our Annual Family Conference in June. The NPC Natural History Study (“Evaluation of Biochemical Markers and Clinical Investigation of Niemann-Pick type C”), being run by Dr Porter at the National Institutes of Health in Bethesda, Maryland, also continues to recruit. If you would like to participate in either of these studies, or would like more information, then please feel free to contact me. Jackie Imrie or Toni Mathieson would also be happy to help.

Exciting news has just been announced regarding a possible new treatment approach for Niemann-Pick Type C – please see Chemical Chaperone article on page 5 of this newsletter.

It was really good to meet all the families and children who came along to the Toddler Clinic at the Willink on 1st February. This was the first time I had had the opportunity to meet them and I found it a very beneficial experience. From the comments and feedback received from the families, it seemed they enjoyed it too. There are great benefits to be had from meeting and talking to others who are going through the same experiences - this is something very positive I have seen at our Annual Family Conference.

I am once again co-ordinating the Children and Young Adult’s Programme for our Annual Family Conference. This year the Conference is to be held at the Hilton Hotel, East Midlands Airport on 28th/29th June. We will be taking the Children to “CONKERS”, which is described as “a hands on experience at the heart of the National Forest”. Please see their website www.visitconkers.com for more information. If you do not have access to the internet, I will be happy to send you more details by post. It looks like it will be a really great day out and I am very much looking forward to it! Once again I will be ably assisted by Scout Leaders but CONKERS also provides Rangers,

who will help to supervise the children during the course of the day, meaning that more adults will be able to take part in the conference and leave their kids to us!! To book your place on the programme please contact Toni Mathieson on 0191 415 0693 or email niemann-pick@zetnet.co.uk

Finally, I was invited to speak at the WORLD symposium on Lysosomal Storage Disorders, in Las Vegas in February. This was a very big meeting attended by many influential people in the field of

Niemann-Pick diseases. I will look forward to telling you more about my experiences at this meeting in the Autumn 2008 issue of Niemann-Pick News.

Liz

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Press Release from Actelion

Miglustat (Zavesca) in Niemann-Pick disease type C

In February 2008, Actelion took the decision to withdraw the application for the extension of indication on the use of miglustat (Zavesca®) for the treatment of progressive neurological manifestations in patients with Niemann-Pick type C disease (NP-C). This decision was taken following an oral explanation with the European Committee for Medicinal Products for Human Use (CHMP) and Actelion is currently evaluating its options to resubmit an application at a later stage. At that point in time, new, relevant data from an ongoing survey of NP-C patients treated with

miglustat could be provided in order to support an extension of indication.

Liz Jacklin, Clinical Research Nurse for the NPDG (UK) comments:

“At the present time this information does not have any implications for patients currently receiving Zavesca®. If you have any questions regarding the press release, please contact me on 0161 922 2967 or email Elizabeth.Jacklin@CMMC.nhs.uk. Alternatively, contact Jackie Imrie on 0161 922 2414 or email Jackie.Imrie@CMMC.nhs.uk”



Building Important Therapeutic Collaborations

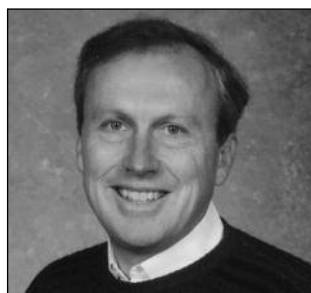
Submitted by Glen Shepherd, Executive Director, Ara Parseghian Medical Research Foundation

Written by: Loni Nannini, APMRF Volunteer

In its commitment to finding a treatment and cure for NP-C, the APMRF is building partnerships and developing collaborations with implications for all genetic neurodegenerative diseases and for the development of disease therapies.

One such APMRF-funded project is the team comprised of Frederick Maxfield, Ph.D., Chairman of the Department of Biochemistry at Weill Medical College of Cornell University in New York, Paul Helquist, Ph.D. and Olaf Wiest, Ph.D., Professors in the Department of Chemistry at University of Notre Dame, and Steve Walkley, D.V.M., Ph.D., Professor of Neuroscience at the Albert Einstein College of Medicine.

The APMRF began funding Maxfield, who has a longstanding research interest in cholesterol, in 2001 after he became aware that his work might be directly applicable to understanding NP-C.



Maxfield has discovered several different classes of compounds with promising effects on NP-C cells in culture, and is now pursuing further steps and testing the most promising compounds for toxicity in mice. He is also determining the cellular mechanisms by which several of these compounds work to help prioritize which groups of compounds to pursue most vigorously.

A unique opportunity to take Maxfield's findings to the next level presented itself several years ago at an annual NPC Scientific Conference attended by Helquist and Wiest. The Notre Dame chemists proposed they form a collaboration to provide the

medicinal chemistry to take Maxfield's compounds toward possible drug development.

Helquist's chemistry expertise allows the team to create additional compounds related to the first "hits" and as the molecular targets of the compounds are identified, Weist's proficiency in computer modeling illustrates the method in which chemicals bind to these targets.

This process leads to ideas for improved compounds that can be formulated in Helquist's laboratory, tested by Maxfield and eventually passed along for testing in NP-C mice with APMRF-funded researcher Steve Walkley of Albert Einstein College of Medicine.

"So with the screening and follow-up cellular testing in my laboratory, the chemistry and computer modeling at Notre Dame and the animal testing at Albert Einstein, we now have the key parts of an interdisciplinary team for the early stages of drug discovery," said Maxfield.

While he is realistic about the fact that many promising therapies fail at some stage and that momentous diligence, effort and patience is needed to test compounds in animal models and possibly patients, Maxfield is optimistic about the potential new classes of molecules that may lead to new therapies.

He is also excited about his role within the team of scientists working thoughtfully towards optimizing treatment for NP-C and possibly other cholesterol-related diseases.

"Until the past few years my laboratory research was entirely focused on basic science. I had hoped that others would take our discoveries in fundamental science and use these to develop therapies. It has become increasingly clear that basic scientists need to think about ways to translate their discoveries into medicines themselves. It is very exciting to think about being involved in the early stages of developing new treatments," said Maxfield



Ara Parseghian Medical Research Foundation

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Chemical Chaperones – the way forward for NPC?

In a recently published article from Washington University in St. Louis, researchers propose a new treatment approach for NPC.

The article talks about a discovery that has led researchers to suggest that chemical compounds could potentially "chaperone" mutant protein molecules through cells. The chemical compounds, which act as chaperones, allow the mutant protein to pass through the cells quality control system, and do its job of moving cholesterol out of cells.

Whilst this is very exciting news and may lead the way to potential new treatments for NPC, it is unlikely that these treatments would be available in the short term.

As with all these things it is not possible to give an exact time scale. Suitable compounds would need to be identified and then these compounds would have to be developed by a pharmaceutical company. Before a drug could be used on patients, it would need to go through animal testing and then Phase I studies, which is the first stage of use in humans. This would initially be done using healthy volunteers. This is a very

important part of the process and it can take quite a long time to ensure that the compound is safe and well tolerated. The correct dosage would also need to be ascertained. Following this, a clinical trial would have to be developed and patients recruited. When a trial is finished and the data collected, then the pharmaceutical company would need to apply for a licence to market the drug. All this can take quite a long time!

The news of chemical chaperone therapy as a possible treatment of NPC is, however, very positive and exciting news for all of us who are hoping and praying for an effective therapy for this disease.

You can be sure that the Niemann-Pick Disease Group (UK) will continue to keep up to date with developments in this area and will give you feedback on any information we have.

You can read the article at

<http://mednews.wustl.edu/news/page/normal/10988.html>

Analysis of Family Health Histories of Patients with Niemann-Pick type C by Karen Quandt RN, BSN, MSN

Karen Quandt is a University of Washington Graduate Nursing Student and, most importantly, Mom of Ty, 10 years old with NPC. Families and members of the NPDG (UK), along with those of The National Niemann-Pick Disease Foundation in the USA, recently assisted Karen with a survey that she conducted as part of her graduate work in the Nursing Program at the University of Washington. Karen's research was designed to help develop a better understanding of whether and how the gene for Niemann-Pick Type C may be associated with genes for other diseases. The goal of the research was to determine whether other diseases occur at a higher frequency in NPC families than in families from the general population.

Karen has kindly provided us with a summary of the survey results. She writes:

"I have now completed my research titled 'Analysis of Family Health Histories of Patients with Niemann-Pick type C'. I was privileged to have 25 families willing to complete a lengthy family health history survey. Each of the families had at least one family member affected by the disease Niemann-Pick type C. There were 299 family members across three generations (child, parents, aunts and uncles and grandparents). There were 153 females and 146 males. There were 31 individuals with Niemann-Pick type C, 18 males and 13 females. The average age at diagnosis of Niemann-Pick type C was 10.5 years.

The prevalence of the diseases and conditions listed by the families in the study was compared to the prevalence of the same diseases and conditions in the general population. The families in this study had a prevalence of 10.3% of autoimmune diseases, compared to 5%-8% prevalence of autoimmune diseases in the general population. There were 31 subjects with at least one autoimmune disease, and three subjects had two autoimmune diseases each. The autoimmune diseases found in the families were asthma, hypothyroidism, psoriasis, Grave's disease, anklosing spondylitis, rheumatoid arthritis, Crohn's disease, psoriatic arthritis, rheumatic fever, eosinophilic fasciitis, Meniere's disease and multiple sclerosis. Asthma was included as an autoimmune disease because the most common type of asthma is allergic asthma, which is an autoimmune disease. Hypothyroidism was included because the most common cause of hypothyroidism is Hashimoto's disease, which is an autoimmune disease. Over 50% of the families had at least one autoimmune disease and 24% had three or more autoimmune diseases.



Hypothyroid disorder had a prevalence of 2.3% in this study compared to 0.4% population prevalence in a study by Canaris, Manowitz, Mayor and Ridgeway, 2000. Grave's disease had a prevalence of 0.7% in this study compared to a population prevalence of 0.1% in the study by Canaris and colleagues.

There were three cases of Parkinson disease in the surveyed families. The prevalence of Parkinson disease in the general population is 1%-2%. The prevalence in this study was 1%. There were two cases of Alzheimer's disease in this study. The prevalence of Alzheimer's disease in the general population is 13% of individuals over the age of 65 years. The prevalence in this study was 9% of the individuals over 65 years. There was one case of Hodgkin's lymphoma, one case of non-Hodgkin's lymphoma, one case of thyroid cancer, and two cases of thyroid cysts. The non-autoimmune diseases such as heart disease, cancer, high blood pressure, all types of arthritis, diabetes type II, depression and high cholesterol did not show a higher prevalence in the families than the general population.

There are numerous studies looking at the relationship between autoimmune disease and multiple sclerosis and autoimmune disease and autism. In two recent studies, both multiple sclerosis patients and autism patients had a prevalence of hypothyroidism in their family histories as well."

If you have any questions about the survey results, you can contact Karen directly by email at kquandt@u.washington.edu

Update from Genzyme

Clinical Trial Update

The Phase 1 clinical study of recombinant human acid sphingomyelinase (rhASM) for treating ASM Deficiency (Niemann-Pick Disease, Type B) is currently ongoing. Seven patients have completed the study to date, with additional patients being screened and scheduled for treatment and assessment visits.

The main purpose of this Phase 1 study is to evaluate the safety of rhASM, an investigational enzyme replacement therapy, given as a single dose to adults with ASM deficiency. During the study, known as a sequential dose escalation study, successive groups of patients are receiving increasingly higher doses of enzyme. The study includes five total dose groups; two have completed the study and the third is in progress. Administration of rhASM has been well-tolerated in all patients to date.

More patients are needed to complete the trial. Based on their medical history, potentially eligible individuals for the remaining two cohorts are being contacted by the study staff. The study is taking place at Mt. Sinai School of Medicine (MSSM) and is open to eligible patients worldwide. Participation in the trial requires up to four visits to MSSM. Travel expenses and study-related medical treatments are being paid for by Genzyme Corporation, which is sponsoring the study. Anyone interested in participating may visit www.clinicaltrials.gov/ct/show/NCT00410566?order=2 for more information.

Enrolment in the Phase 1 study is expected to be completed this year, with data collection and analysis to follow in 2009. Appreciation goes to all the patients who have participated in the study thus far, as well as to Drs. McGovern, Wasserstein, Schuchman, and Desnick at MSSM for their ongoing work on the study.

Plans for a multi-national Phase 2 study are currently in development. Findings from the Phase 1 study will be used to help design the Phase 2 trial. We anticipate that the Phase 2 study will be the first opportunity (of at least two required for regulatory approval) to evaluate the effect of repeat dosing on various disease symptoms over several months.

Staffing Transitions

Betsy Bogard recently assumed responsibility for the Niemann-Pick development program at Genzyme. Betsy has a master's degree in health policy and management from the Harvard School of Public Health and has 11 years of experience in the



biotechnology industry in areas that include clinical trials management, health economics research, post-marketing studies, disease registries, and program management. She has been at Genzyme for over four years, working primarily on Gauchers disease. She is also a member of the rare disease community in her own right—her younger brother has fibrodysplasia ossificans progressiva, an extremely rare genetic disease that causes bone to form in muscles.

Betsy is very excited to be part of Genzyme's Niemann-Pick development program. She is continuing the work started by Paul Kaplan, who led the program for the past six years and successfully brought it into the clinical trial phase. Paul will not be far away; he is focusing his attention on Parkinson's disease research at Genzyme.

Betsy Bogard

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Research Report by Bill Owen, Research Co-ordinator

General Outlook

2008 seems to have come around very quickly and we have continued to see new cases of all the Niemann-Pick variants being diagnosed in the past year. Sadly, we have also witnessed the loss of others. This pattern will continue into the foreseeable future, although we continue in the hope that a breakthrough will shortly happen. Solving the problems of the science associated with cellular lipid transport is clearly a major challenge to the scientific community, even with the support of Nobel Prize winners. I understand that Drs Brown and Goldstein, who won the prize in 1985 for their work on cholesterol transport, will be speaking at the Ara Parseghian Medical Research Foundation Annual Scientific Conference on Niemann-Pick Type C in June.

Looking through some recent research publications, I was reminded of the problems faced and also, how the research is becoming more focussed on what happens in the brain. Preventing cell death in the brain would be a major contribution to survival of patients although there may be other problems to resolve in attempting to deliver full recovery. I fear that none of this is just around the corner, but who can say?

Prospects for this year

We still await the results of the miglustat trial and, most hopefully, the Type B trial will yield benefits in the next few years. We can watch and wait, always a difficult thing to do, but we can begin to take our own actions aimed at disease prevention in future generations. I have previously written about prevention, or risk reduction and we are now developing some initial feasibility work I carried out last year, into a proposal against which we intend to seek funding to set up a carrier testing facility for known carriers and their partners. To do this we will need to establish partnerships with academic scientists, others in healthcare and possibly, industry. Most importantly, the willingness of families to undertake self help will be crucial. I believe that the availability of testing, once established and, with suitable education and counselling will provide a route to reduction of these diseases within our community. My nephew has taken

a first step towards this goal and has written an article about his experience and feelings of taking a carrier test. Thanks Stuart. This is a start. I think that we are going to be in for a very busy year.

Attempts to interest MPs and the Department of Health (DH) in taking an active interest in rare genetic diseases in general, have met with little success to date. I believe that we need to show the way. One brighter note concerns the activities undertaken by the umbrella charity, Eurordis, of which we are members. After some years of work with the health organisation of the European Union a major success has been achieved for rare genetic diseases. Lesley Greene from CLIMB was very much involved in this. The European Commission has issued a consultation document entitled Rare Diseases: Europe's Challenge. We, along with many other charities, are in the process of responding to this document which appears to recognise the serious neglect of families with rare diseases and is attempting to address the management issues as a starting point. We are interested to determine what part our own healthcare providers are playing in this EC initiative. I have yet to interpret the reply from the Department of Health to my letter on this question.

To advance this EC initiative, the umbrella charity Genetic Interest Group (GIG) in which we also have membership, is holding a reception at the House of Commons late in February to mark the occasion of the 1st European Rare Disease Day. They have requested that we, the rare disease charities, invite as many MPs and Lords to attend as we can manage. It is planned to attract media attention by using family experiences in order to get the message on rare genetic conditions to a wider audience. I have collected press cuttings over many years so I may be able to assist. Some of these have been brought to my attention by the staff at our corner shop where we also have a collection box. Perhaps awareness is improving but it seems to take forever. Let us hope that the Consultation Paper and the meeting at the Commons is the start of some meaningful programme of work for rare genetic diseases.

Summer Concert to Benefit the NPDG (UK)

Tony Jellings, former Trustee and Board member continues to do so much to support the fundraising efforts of the Group. He has once again organised a charity concert in aid of the NPDG (UK), which will take place on Thursday the 14th August at 7pm at St George's Church, Kemp Town, Brighton, featuring the award winning musicians Guy Johnston (cello), who is also one of our esteemed Patrons, and Simon Lane (piano). Thank you Tony, your efforts are very much appreciated!

Niemann-Pick Type C – Carrier Testing

Stuart Owen 17 January 2008

For about fifteen years, I saw my cousin, Caroline's, health deteriorate very slowly. She had always been a lively, talkative and very friendly person. I didn't really understand what was happening to her, or why. One thing I'll always remember was her iron grip whenever you held her hand!

At some stage, it had been identified that my paternal grandfather was a carrier of the Niemann-Pick Type-C P1007A genetic mutation and that my uncle Bill (Caroline's father) was considering asking his sisters and brother (my father) to be tested. As well as for personal reasons, this would help document its history in our family for future generations. Around the time of my grandfather's death, I became interested in genealogy. When Caroline passed away of the disease in 2005, at the age of 30, I also wanted to find out more about Niemann-Pick and how it related to the Owen family.

In June 2007, my wife Yvonne and I underwent IVF treatment at Hammersmith Hospital, London, which proved successful. To satisfy our curiosity, we both agreed that I should arrange for a genetics test. I had a lengthy telephone conversation with Bill; He's become quite an expert in his capacity as the NPDG (UK) Research Co-ordinator. My Consultant Geneticist & Counsellor was also very helpful; she drew up a family tree (another one!) and took some blood samples. I gave her a copy of the 'Establishment of Carrier Status' letter that Bill had sent me.

A few months later, I rearranged my follow up appointment for a different date to the one I was initially given. Due to an administrative error, instead of receiving my results in person at the hospital, I was

sent a letter prior to my supposed appointment asking me if there was a reason why I had not attended. My consultant went on to mention that, although it wasn't essential to have the results face to face, it would be more beneficial. This was especially relevant in my case as my results were positive and I was a carrier of the Niemann-Pick Type-C mutation that had been identified in my family.

My initial reaction was irritation over the way in which I had received this important news, rather than what it actually meant. Then it began to sink in, with Yvonne and I feeling stunned for the rest of the day. I got in touch with my consultant, explained the situation and made another appointment to discuss the results with her.

The outcome of this session provided us with some helpful answers, such as the risk of us having a child affected by Type-C was low – at 1 in 800. Put in context, everyone has a 2-3% chance of having a child with some sort of genetic disorder. Even so, it's frustrating that there's nothing more we could do about the diagnosis. It's like doing a jigsaw with half the pieces missing - I'm a carrier, but Yvonne cannot be tested in the same way, because there is no known history in her family.

Within a few weeks of this news, we found out on the day of her twelve week scan that Yvonne had miscarried. Obviously, this was a severe blow. In the long term, it did help put things into perspective. Yes, I am a carrier of a genetic disorder. Yes, I still want children. No, it's not going to change our plans, why should it? Perhaps our more positive outlook helped us later in the year to conceive naturally. In June 2008, we are expecting our first addition to the family tree.

NPDG (UK) Annual Family Conference 2008

Care Matters

27th/ 28th / 29th, June 2008, Hilton Hotel, East Midlands Airport

This year's Conference will focus on providing families with practical advice and solutions to assist them in dealing with the day to day challenges presented by the Niemann-Pick diseases.

We are grateful to all of those who provided us with suggestions as to how we could tailor the Conference to meet your needs. All feedback from Conference

2007 was taken into account as we began to plan for this year and I hope that you will find the programme helpful and interesting.

We have made a few changes to the programme – the main one being the inclusion of a Welcome Meeting on the Friday evening. Following comments received after previous conferences it became apparent that those of you arriving on the Friday evening would like the opportunity to meet and get to know the Board of



Sally Wray and family

Trustees' and other attendees informally. As you will see from the programme enclosed with this newsletter, the meeting will take place from 7.30pm until 9.30pm, with light snacks and refreshments being served.

Although the Conference will not focus on scientific research this year, there will be important updates on the progress of the clinical trials of Miglustat (*Zavesca*) for NPC and the Phase 1 study of rhASM for NPB. Dr's Marc Patterson and Margaret McGovern will once again be joining us from the USA, along with Nicole Yanjanin, who will tell us all about the NPC Natural History Study taking place at the National Institutes of Health in Washington DC. From France, we once again welcome Dr Marie Vanier, who will provide our keynote speech.

Peter Limbrick, Chair of the Handsel Trust, and editor of the 'Interconnections' e-bulletin will share his experiences with us, as will Louise Derbryshire from Contact a Family and, back by popular demand, Lois Race will update us on changes to the benefits system.

There will also be presentations from some of the many care professionals that you may come into contact with; they will each explain how their roles can assist you in ensuring that your family receives the optimum level of care and support.

Conference 2008 aims to include something for

everyone, as you will see from the enclosed programme. There will be the popular and informative Breakout sessions, Family Voices and as always, plenty of opportunity to ask our Speakers any questions you may have.

The Children and Young Adult's Programme is looking as exciting as usual. Liz Jacklin, our own Clinical Research Nurse, is running the programme again this year with help from a team of experienced volunteers. This year there will be a trip to "CONKERS", which is described as "a hands on experience at the heart of the National Forest" (Please see their website for more information www.visitconkers.com), plus fairground games, crafts and many other fun activities. It looks like it will be a really great weekend and I am sure that all participants will have lots of fun!

To book your place for Conference 2008 simply complete the enclosed form and return it to the office at 11 Greenwood Close, Fatfield, Washington Tyne and Wear, NE38 8LR. If you have any questions about the Conference contact Toni Mathieson on 0191 415 0693 or Jackie Imrie on 0161 922 2414, they will be happy to help. Limited funds are available for those families needing help with expenses, please contact the office for more details: all enquiries will be treated in confidence.

We do hope you will be able to join us for this exciting weekend, as well as hearing the latest important information. This weekend is all about sharing information and strengthening our family support network, so there will be plenty of opportunity to meet other families, make new friends and speak directly to the professionals involved.

Finally, whether you are a regular attendee or joining us at Conference for the first time, we want you to have a positive experience and hope that Conference 2008 will meet all of your needs and expectations.

See you in June!

Janice

Janice Brooks
Conference Chair

COMMENTS RECEIVED AFTER CONFERENCE 2007:

'This was our first NPDG (UK) meeting and it was very interesting to hear about the diverse aspects of the Disease. It was very good to put faces to names and meet the staff.'

'The children, again, enjoyed themselves and the fact that they get looked after helps us relax and enjoy the Conference which we found most helpful.'

'A very good overall mix of families, medics and scientists – a similar programme next year would be excellent.'

'It was especially good speaking to other families who have been affected by NP. They made us realise we are not alone in everything we are feeling and going through.'

Fundraising News

Thank you to each and every one of you who has been busily fundraising for the NPDG (UK) recently, your efforts make a huge difference and are very much appreciated – so please keep up the good work!! If there is any thing we can do to support you in organising an event or if you would like more information on fundraising, or a fundraising pack, please contact us at the office on 0191 415 0693 or email niemann-pick@zetnet.co.uk .

UIA (Insurance) is a keen supporter of local and small charities, and when looking at ways to raise money, staff members decided to hold monthly dress down days, where a small donation of a £1 each was made for the privilege of wearing casual clothes. In order to make a more noticeable contribution, it was agreed that one nominated charity would benefit for a period of a whole year. Since summer 2007, UIA has been supporting the Niemann-Pick Disease Group (UK), due, not only to its status as a worthy cause, but also because it is a cause close to one staff member's heart. So far UIA staff members have raised over £600,



but just as importantly have raised awareness of the disease and the Charity's good work within the company. UIA will continue to support Niemann-Pick during the first half of 2008.

Picture shows staff members Nikki Creasey and Donna Beckles raising money on UIA's dress down day in January.

BG Group, an international oil and gas operator, sent a donation of £1,400, which they raised by operating a "Lost Time Incident" free charity donation incentive, which involved their main contracting companies donating a set amount of money to charity for every day completed with out an LTI. Each week the workforce selected a charity to benefit from the money. During October 2007, NPDG (UK) was one of the lucky charities selected to benefit.

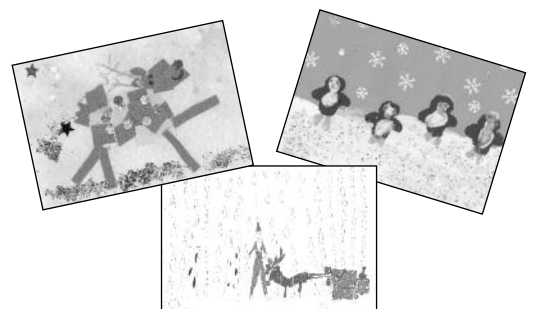
Kait and Norman Pyne (*right*), from Clyst St Mary in Exeter, held a bonfire party raising £100 whilst Clyst St Mary Primary School raised £210.70 from a collection held during their nativity play. The total amount for Norman's Land's End to John O'Groats Bike ride now stands at £7,619.00 – Wow!!



2007 Christmas Card Competition

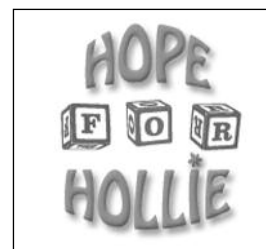


Thank you to all who generously supported the Group by purchasing our 2007 Christmas cards, which were once again very popular thanks to the wonderful designs of our budding young artists; Annie Pyne and Beth Lingard, plus sisters Alice and Ellen Byrne. The total amount raised was an amazing £2,397.50!



'Hope for Hollie' Campaign

The Hope for Hollie Campaign, started by parents Pete and Helen Carter following their daughter Hollie's diagnosis with NPC (see page 18 for Hollie's story), has so far raised £2,419.00, with all proceeds kindly donated to the NPDG (UK). Events have included:



Clacton Fire Brigade's Car Wash



After seeing an article about Hollie and her family in the local press, Steve Moseley and his Green Watch colleagues at Clacton Fire Station decided to hold a 'Car Wash' event in aid of the 'Hope for Hollie' Campaign. The weather actually behaved and as you can see from the photos all involved enjoyed the day, which successfully raised £522.00!

MKDSA Juniors vs Downs Barn Juniors – All for Charity!

On the 8th December the MK Dons Supporters Association (MKDSA) Juniors took on Downs Barn Juniors in a charity football match in aid of the "Hope for Hollie" Campaign. The weather wasn't kind, it poured down with rain for the duration of the game! The final score was 2-2. Grateful thanks go to all the players and the organisers for arranging this event which raised £325.00 for NPDG (UK). What a fantastic amount!

MK Dons Bucket Collection



With the kind consent of the MK Dons Football Club, the MK Dons Supporters Association (MKDSA) held a bucket collection as part of the 'Hope for Hollie' campaign in aid of NPDG (UK), at the home game against Barnet on Saturday 12 January 2008. The event was an overwhelming success thanks to all the bucket collectors who worked so hard on the day and to all those MK Dons fans and Barnet supporters who kindly donated. The fantastic sum of £1,202.00 was raised on the day!!

Casino Royale Charity Ball

Helen and Pete Carter will be hosting the first 'Hope For Hollie' Charity Ball, in aid of the Niemann Pick Disease Group (UK), at the Stadium:MK Ballroom on Saturday 5th April 2008, the ball will have a 007



theme. The night will start with a Champagne Reception and will feature live music, a charity auction, a disco and light show plus a three course meal including wine. All proceeds from this event will go directly to support the work of the Niemann Pick Disease Group (UK). This event will help to raise awareness of NPD as well as funds to aid research into a treatment/cure. Tickets are still available, please contact the Central Office on 0191 415 0693 or email niemann-pick@zetnet.co.uk to find out more.

Running Wild...

Thank you to the following very energetic people who managed to raise funds and keep fit at the same time:

- Sharon Hunter ran in the Folkestone Half Marathon and raised £300.00
- Liz Wright ran in the 13 Mile Robin Hood Marathon and raised £200.
- Stewart Mathieson and his colleagues at Ernst & Young raised £2798.00 when they once again took on the challenge of the Great North Run
- Marghanita Rutson got her whole family involved, they ran in two separate London events raising £100.00
- Look out for Nigel Hopwood as he trains for the Bristol 10k run, which will take place on the 4th May this year. Nigel has been persuaded to run in aid of the NPDG (UK) by Coral MacLean and family – we wish him lots of luck!! If you would like to sponsor Nigel, you can visit his fundraising page on <http://www.justgiving.com/nigelhopwood>

Heartfelt thanks also go to:

- ▷ Northumbria Police, who held a dress down day and raised £76.91.
- ▷ Deborah and Cathy Murphy from Belfast raised £230.00 from the sale of wristbands and collection boxes.
- ▷ Mrs Clarke held a coffee morning – raising £245.75.
- ▷ 1st St John's Brownies raised £43.00.
- ▷ Val Ridley and Rose Thompson, with help from their many friends at Astral house in Sunderland raised over £500 from various events including coffee mornings, table top sales, a pie and pea supper and raffles.
- ▷ Ernst & Young for raising a total of £2468 at their Christmas part by way of a raffle and tombola.
- ▷ Mary and Phil Winetroube, of Warner of Wingate, for their kind donation of £250.00.
- ▷ John Imrie and his work colleagues at Stockport Borough Council raised £63.00
- ▷ Jannick Dance raised £150.00 by organising a dance show.
- ▷ The staff of the Cheltenham and Gloucester Nuffield Hospital raised £42.00. The money was raised by staff purchasing a gift tag for their Christmas tree to wish Happy Christmas to their colleagues in lieu of sending cards.

Awareness Week 2008

July 13th – 19th 2008

We are hoping that, as our 5th Annual Awareness Week approaches, the weather will be at least a bit better than last year – although most of you still managed to hold successful events, even with torrential rain and floods to contend with! We hope that once again, those of you who have joined in the fun in previous years will help to raise awareness of Niemann-Pick diseases and raise funds to support the work of the Group.

This year, we once again invite you to organise an event such as a 'Niemann-Pick-Nic', a balloon race or a garden party. Perhaps you could involve your local school or community group? Our aim is to have many events all over the country, attracting the attention of the media. After all it is all about awareness!

Remember any event, whether it is large or small is welcome. We can provide you with advice on how to

organise an event and supply you with a fundraising pack, containing; a t-shirt, stickers, a poster, an ideas sheet, collection boxes, information leaflets and a sponsor form. We also have other items you can use to help raise funds, such as wristbands and balloons.

If you would like some ideas or a fundraising pack, please get in touch with us at the office on 0191 415 0693 or by email niemann-pick@zetnet.co.uk

Remember, Awareness Week is your chance to make a difference and your chance to help in the fight against Niemann-Pick disease – so please get involved – we need your help to make Awareness Week 2008 a success!

Not free during Awareness Week?

Don't worry – you can organise your event on a day suitable for you – the most important thing is to get involved!

Order your NPDG (UK) fundraising pack:

Many items in the pack can be downloaded from our website www.niemannpick.org.uk or you can order a full pack to be sent in the post by calling 0191 415 0693 or email niemann-pick@zetnet.co.uk

Let us know what you've got planned!

We would love to know what you are planning and will offer all the support we can to help make it a success! We can also promote events open to the public on our website. Email niemann-pick@zetnet.co.uk also - don't forget to take some photos – our members love to read about the events in our newsletter!

Why not set up your own fundraising page with justgiving.com?

Create your own fundraising page on justgiving.com, personalise it with a photo and a message, and then email it to all your friends and family. They can then donate securely on line with a credit or debit card. No more dealing with cash or cheques, or having to chase people up after the event! Plus, on line donations from UK taxpayers collect a 28% bonus! So you do less, but the charity gets more! This service is free and should take around 5 minutes to set up — that's it! Easy!

To create your own fundraising page visit:

<http://www.justgiving.com/niemannpick/raisemoney>

Shop on line and raise funds without leaving the house!

We've teamed up with [easyfundraising](http://easyfundraising.org.uk), an online shopping directory listing over 400 of your favourite stores, including Amazon, NEXT, Debenhams, John Lewis, Tesco, DELL and many more. Every time you shop online with any one of them, we'll receive up to 15% of every purchase – at no extra cost to you! It's completely FREE to register and use and it doesn't cost a penny extra to shop and raise funds in this way.

All you need to do is log on to the [easyfundraising](http://easyfundraising.org.uk) site at

<http://www.easyfundraising.org.uk/niemannpickdiseasigroupuk> - then register and start shopping!

justgiving
fundraising made easy

easyfundraising
.org.uk

Benefits News

Now let's talk money

Trying to cope with money worries alone can be hugely stressful. A new national Freephone helpline has now been launched by the Department for Work and Pensions to encourage people on low incomes to ask for help. If you would like to know where to go for affordable credit, information about banking, or how you can access free face-to-face money and debt advice, the number to call is: 0800 012 1656 or visit their website: <http://www.nowletstalkmoney.com> Everything you discuss will be held in complete confidence.

Do you qualify for a reduction on your Council Tax?

It was recently reported that very few carers are aware that they may be able to claim a reduction on their Council Tax. If the carer, or another resident of the house is 'substantially and permanently disabled' – this could apply to either an adult or child of any age, whether they are related to you or not – then you may be eligible to claim a disability reduction. To qualify, you must meet at least one of the following criteria:

- You have an additional bathroom or kitchen that is needed by the disabled person.
- You have a room (other than the bathroom, kitchen or toilet) that is mainly used by the disabled person.
- Your house is spacious enough to enable the use of a wheelchair indoors.

For more information contact your local council office or visit the Direct Gov website: www.direct.gov.uk/en/DisabledPeople/FinancialSupport

The Independent Living Fund (ILF) – new rules from April 2008

As a result of increased demand for help, the ILF has announced that it will introduce some rule changes which will reduce the numbers of people they can help.

The ILF gives money to people aged 16 or over, who qualify for Disability Living Allowance (DLA) care component at the highest rate and who need an extensive package of care in order to live independently. This money must be used to buy extra care to top up the help they get from social services.

The changes include:

- You will only receive help from the ILF if social services agree to fund the first £320 per week (up from £200) of your care package.
- The ILF will no longer be able to provide help to everyone who meets their criteria. Instead they are likely to have to start prioritising requests for help with highest priority given to applications from disabled people working 16 hours or more a week.

The new rules will apply to anyone in England, Scotland or Wales applying to the ILF after April 2008. If you already get ILF payments you will not be affected, unless you ask for an increase in help after that date. For more information call the Contact a Family free helpline on 0808 808 3555.

VAT Relief for Disabled People

The HM Revenue and Customs Booklet 707/1 outlines the rules on claiming VAT relief on home alterations and items of equipment. If you would like a copy call 0845 010 9000

Changes to the Blue Badge Scheme

In October 2007, The Department of Transport introduced some changes to the Blue Badge Scheme. This scheme was extended to include children under the age of two who need to travel with bulky medical equipment; may need to be near a vehicle for emergency treatment; or have hip dysplasia. The scheme is still under review and the Department of Transport may announce further changes which will take effect in spring 2008. For more information contact your local council office or visit the Direct Gov website: www.direct.gov.uk/en/DisabledPeople

The Directgov Blue Badge Parking Map

The Blue Badge parking map, which provides information on blue badge parking bays, is being significantly improved from February 2008. There will be a large increase in the number of locations featured as well as additional information on accessible toilets, train stations, Shopmobility sites and much more. For further information visit

http://www.direct.gov.uk/en/DisabledPeople/MotoringAndTransport/DG_10038295

Every Disabled Child Matters Campaign - Fuel poverty hits families with disabled children hardest

The Every Disabled Child Matters campaign has added its voice to national charity Contact a Family's call on government to give families with disabled children a winter fuel allowance, which would match that currently paid to pensioners.

Two thirds of families with disabled children struggle to pay their fuel bills in wintertime. And 10 per cent have had their gas or electricity supply cut off – either by their supplier or because they couldn't afford to top up a prepayment meter.

Research shows that UK families of disabled children are among the poorest. It costs three times as much to bring up a disabled child compared with other children, and childcare costs for disabled children are up to five times as much.

They are both 50 per cent more likely to be in debt than other families, and 50 per cent less likely to be able to afford essentials like new clothes or school outings.

Higher energy bills in the winter are yet another cost that hits families with disabled children disproportionately hard. Surveys show their yearly gas bill is nearly £600, compared to £350 for other families. Many conditions are made worse by the cold, or children may be more prone to picking up infections, so the heating needs to be on for longer.

Steve Broach, Campaign Manager for EDCM, comments: 'The government is committed to halving child poverty by 2010 on the way to ending child poverty by 2020. Reducing poverty amongst families with disabled children must be part of this, and extending the winter fuel allowance to them will help achieve this target.'

For more information and to find out how you can help visit <http://www.edcm.org.uk>.

Direct Payments

Local Authorities can give payments, instead of services, to allow disabled people and their carers to buy in the services they have been assessed as needing. Direct Payments are seen to promote the independence of parents and their disabled children who would like to manage their own social care needs. If you are unhappy or frustrated with the services you currently receive, Direct Payments may be the answer.

If you have a child who is under 16, Direct Payments will usually be made to you as their parent. When a child becomes 16 she or he can receive payments in their own right to allow them to buy in the services they need. Direct Payments give you the flexibility to employ a carer or the money can be used to buy services from a private care agency, a nursery or a voluntary organisation.

Whilst using Direct Payments could give you more control over the services you receive, it could also mean more work for you, as you will need to arrange and manage the care yourself.

In the past you could not insist on Direct Payments, however, in England, a request should now be refused only in very limited circumstances. The amount you receive should be enough to allow you to meet all the costs involved; including tax and National Insurance as well as the fee for a police check (should you employ help directly). Further information on Direct Payments can be found on the Department of Health website: www.doh.gov.uk/directpayments

Contact a Family can also provide advice on Direct Payments, call their free Helpline on 0808 808 3555 or visit their website www.cafamily.org.uk

Help and Advice

Action for Kids - support for disabled children.

Action for Kids is a small, national charity which supports disabled children, young people, their parents and carers. They provide three main services:

- Mobility equipment provision service. They provide a range of equipment including trikes, walkers, communications aids and sophisticated powered wheelchairs. They also offer a wheelchair

maintenance and repair service, for children and young people aged 2 to 26 across the UK.

- Work related learning services for young people aged 14 to 26 which offer work experience in 17 London boroughs and in Hertfordshire.
- Family support services for disabled young people and their families.

For more details please visit their website: <http://www.actionforkids.org> or contact their helpline on 0845 300 0237

Need Advice? Call Scope Response.

Are you confused by the wealth of information available on disability issues? Scope Response is a new free and confidential helpline that can provide callers in England and Wales with advice on a wide range of disability issues. Their Helpline staff aim to answer your enquiry clearly and quickly and will point you in the direction of other relevant services if necessary. Call the Scope Response Helpline 0808 800 3333 or visit their website www.scope.org

Transition

What is Transition?

Transition takes place between the ages of 14-25, and is the time when young people have to make important decisions about their future. Like all teenagers and young adults, those with disabilities will inevitably experience the ups and downs of adolescence; however these experiences may be more problematic than their non-disabled peers, due to additional health problems and the fact that they are often involved with a number of additional support agencies.

As research has shown that transition can be a difficult time, and in order to ensure the young person's needs are fully met, it is wise to start planning well in advance. Young people may find the decisions they are making about further education, independent living or relationships exciting, but equally they may find the whole experience daunting. The purpose of planning is to guarantee continuity of care, and to ensure that the young person has access to information and support to assist them in making informed decisions. It is imperative to keep the young person and their family at the centre of the planning and to consider their feelings and wishes about throughout the process. Forward thinking will help to reduce feelings of stress and anxiety and will enable the creation of a clear plan to assist all involved in the transition process.

Resources and Further Information

The Transition Information Network (TIN) www.after16.org.uk

The Transition Information Network (TIN) is a website for parents, carers and people who work with and for disabled young people in transition to adulthood. TIN is an alliance of organisations and individuals who come together with a common aim: to improve the experience of disabled young people's transition to adulthood. TIN is a source of information and good practice for disabled young people, families and professionals.

A transition guide, funded by the Department for Children, Schools and Families (DCSF) and produced by the Council for Disabled Children with a reference group, has now been published. It is available to download from www.after16.org.uk Hard copies can be obtained from the DCSF's publications department, you can contact them on 0845 60 222 60, textphone 0845 60 555 60 or email at dcsf@prolog.uk.com

Contact a Family has an excellent factsheet which can be downloaded from their website <http://www.cafamily.org.uk/transition.html>

Useful publications and websites

Understanding Niemann-Pick disease Type C – a new booklet

Published by Blackwell Publishing in conjunction with Actelion and written by Dr Ed Wraith and Jackie Imrie, this booklet aims to highlight the social and healthcare impact of this NPC.

Defining Children's Palliative Care

ACT has published two new resources for anyone using or working with children's palliative care services. These are a 'Children's Palliative Care Definitions' guide and a factsheet explaining the differences between children's and adults palliative care. You can download both of these factsheets from the ACT website www.act.org.uk

Useful Websites

Living Life with Greater Challenges is a website for young people, with chronic or long term conditions, to share their stories and insights. The issues explored include symptoms and diagnosis, staying in hospital, information needs, feelings and emotions, life style, school, university and work, friends and relationships. www.youthhealthtalk.org

The International Children's Palliative Care Network has a new website with information about good practice in palliative care, and a directory that includes hospices and other organisations in the UK. www.icpcn.org.uk

Network 81 is a national network of parents working towards properly resourced education for children with Special Educational Needs. They aim to support groups and individual parents of children with special educational needs, to raise awareness and publicise good practice in inclusive education and, to promote parent-professional partnerships. www.network81.org

The **Transition Information Network (TIN)** is for disabled young people in transition to adulthood,

their families and people they work with. TIN is an alliance of organisations and individuals who come together with a common aim: to improve the experience of disabled young people's transition to adulthood. TIN is a source of information and good practice for disabled young people, families and professionals. www.after16.org.uk

The National Society for Epilepsy provides information and support to professionals and people affected by epilepsy. They also provide care for people with epilepsy through medical, residential and rehabilitation services. Their website contains a range of information on epilepsy, treatment and many aspects of living with epilepsy. www.epilepsynse.org.uk

Special Educational Needs and Disability – Removing the Barriers to Achievement

Department of Education and Skills - Special Educational Needs section (SEN): www.dfes.gov.uk/sen

It is the ambition of the SEN and disability division that every child with special educational needs reaches their full potential in school. To promote the welfare and interests of disabled children and to improve the support they receive, this website provides a wide range of SEN and disability advice and materials for teachers, parents and others working with children with SEN in England. For advice on SEN in other areas of the UK see below or contact the SEN team at the Department for Children, School and Families by email: sen.queries@dcsf.gsi.gov.uk.

Scotland: www.scotland.gov.uk/about/strategic-board

Wales:

www.learning.wales.gov.uk/scripts/fe/news_list_archive_bysubject.asp?CatID=23

Northern Ireland: www.deni.gov.uk/index.htm

The Foyle Foundation – A Thank You

The NPDG (UK) would like to thank The Foyle Foundation for their generous offer of £20,000 in grant funding, in support of our work in 2008. The grant is specifically to support the posts of Clinical Nurse Specialist, National Development Manager and Administration Assistant. This is an important contribution to the funding of the above posts and the support of The Foyle Foundation is greatly appreciated.

The Foyle Foundation is an independent grantmaking trust that distributes grants to UK charities whose core work is in the areas of Learning, the Arts and Health. Since it became operational in November 2001 The Foundation has disbursed over £26.2m in grants.



THE
FOYLE FOUNDATION

Family Focus

Hollie's Story



We are relative newcomers to Niemann Pick disease. Until July last year neither we nor any of our family and friends had ever heard of the disease.

Hollie (*above*) is our second child. We already have a son, Joshua (*pictured above, right*) who is aged 6. Thinking back my pregnancy with Hollie had always felt different to when I was carrying Joshua. I would regularly visit my midwife and question whether my “bump” was growing as it should. I felt extremely small in comparison to when I carried Josh, certainly in the latter stages of pregnancy. At about 32 weeks my midwife measured my stomach with a tape measure and told me I was measuring approximately two weeks behind but reassured me it was unlikely to be anything to worry about, but still it continued to play on my mind. By 37 weeks I was measuring 4 weeks behind and my midwife immediately referred me to our local hospital for a scan as she was worried that the baby was failing to grow. This resulted in my pregnancy being induced at 38 weeks and Hollie was safely delivered on the 11 June 2005 weighting a tiny 5lb 12oz. Even when she was born and handed to me I remember saying to the midwife “she is so tiny, something must be wrong with her” but the midwife replied “she is small but perfectly formed”.

We returned home with Hollie the following day. Joshua was over the moon with his new baby sister. Straight away we noticed Hollie was quite “tanned” looking but we knew it was not uncommon for newborns to have jaundice for the first few days so never gave it much more thought. As the early days passed Hollie’s jaundice seemed to subside but her stomach seemed a strange shape. Again, we were not overly concerned as we put it down to the fact that

Hollie was small but feeding well and her stomach was just swollen from all the milk she was drinking. But then we noticed that Hollie seemed to become more and more unsettled at night, she would scream constantly and if she wasn’t screaming she was feeding contin-



uously. Nothing seemed to settle her at night and it began to concern us that this was more than just a simple case of colic. I began to notice how jaundice Hollie looked first thing in the morning when I picked her up from her moses basket. The midwife however appeared happy with Hollie as she was growing well and her jaundice would appear to fade as the day went on. It wasn’t until our Health Visitor did a home visit at 8 weeks that things changed. I undressed Hollie so she could be weighed and straight away my Health Visitor raised concerns about the shape of Hollie’s tummy and the fact that she still appeared jaundice. We were immediately referred to our GP. I knew something was wrong when the GP asked me to leave the room whilst she spoke to the hospital and then came out and told me to take Hollie to the hospital straight away.

By the time I reached Milton Keynes Hospital I was frantic with worry, numerous doctors were tapping Hollie’s stomach and talking amongst themselves. I then had to face the dreaded blood tests for the first time, a procedure which is now second nature to Hollie! I can recall standing in the corridor in floods of tears from hearing Hollie’s screams as three doctors tried to get blood from her. From Milton Keynes we were referred to Kings College Hospital in London where we were told that Hollie’s liver functions were extremely poor and she was very unwell.

From this point to diagnosis took a further two years. During the early weeks Hollie underwent extremely invasive testing for numerous illnesses and liver related diseases. All tests drew a blank but Hollie’s liver functions were extremely poor and doctors were seriously concerned and baffled. Then strangely at

around ten weeks old Hollie's liver functions started to return to near normal and we were allowed to take her home. She continued to develop normally apart from her enlarged spleen. Hollie was classed as having "undiagnosed liver disease" and put under six monthly review. Life returned to normal and we felt like we were extremely lucky to have escaped anything more serious. The only sign that Hollie ever had a problem was her distended tummy. We had expected this to subside with time but it never did. Every time Hollie had a review at Kings College Hospital the ultrasound would show that her spleen was enlarged although her liver seemed to be stable. Every doctor we came across told us not to worry and her spleen was just enlarged due to the liver disease.

It wasn't until we met a doctor on a routine liver review at the beginning of 2007 who had come across Niemann Pick disease before that we were put on the road to a devastating diagnosis. By this time Hollie's spleen measured a huge 14.5cm on the scan and he felt a fresh investigation was required. Hollie underwent further invasive testing, a second liver and bone marrow biopsy and a skin biopsy. No one could have prepared me for the news I was to receive over the telephone at work shortly after from a Registrar who appeared to know little about the disease. I was told that tests indicated that Hollie could have Niemann Pick C and that she would start to decline any time after the age of two and lose all the motor skills already developed. He said he was extremely sorry especially as Hollie had been "doing so well". I was told to go and look on the internet to find out more and come back in six to eight weeks when the results were ready. I went straight to my desk and typed into an internet search "Niemann Pick C". What faced me was horrific. I remember my eyes just skipping from one sentence to

"Hope for Hollie"

Following on from Hollie's diagnosis we decided that we needed to be doing something positive to enable us to cope. The one thing that struck us immediately was the lack of knowledge of this disease. We therefore decided to launch a campaign called "Hope for Hollie" the aim of which was to raise awareness of the disease not only in the community but across the medical profession and to raise funds to help NPDG (UK) to continue to support families and aid research into a treatment/cure.

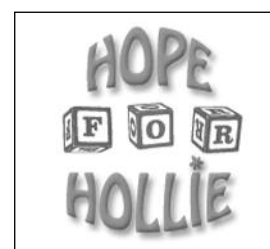
So far the support we have received towards this campaign has been amazing. We are hosting our first charity ball on the 5th April at the ballroom of our newly built MK Dons football stadium. The theme will be 007 and we hope to make this a fantastic night not only for the general public but also for the local medical

the next looking for a positive but I just couldn't see anything. At that point I broke down in tears and had to be physically helped from the office.

The next ten weeks were the worst of our life. I took it upon myself to search out more information and found the contact details for NPDG (UK). Toni kindly provided me with the support I needed and referred me to Jackie at the Willink who gave my husband and I a clearer picture of the disease and testing process. The definitive results came through in July but I think we had already accepted the fact it was NPC long before that. We knew that the liver biopsy and bone marrow biopsy all pointed strongly in that direction and although we tried to remain positive deep down we knew it was now just a case of rubber stamping the results.

People constantly ask us how we cope and comment about how brave we are but I don't think we are brave. We have no choice but to accept that NPC is part of our lives and the fact that we may never know for sure when the disease will start to show itself fully. The uncertainty that this carries is extremely hard. Each day Hollie learns something new and she is the most happy little girl, bubbly and full of fun. She is a very active child and is never happier than when she is dancing to music in the lounge or playing on the slide and climbing frame in the garden. The thought of Hollie losing the ability to do these things is still unbearable. The immense fear that sometimes engulfs me about the future is terrible. We are determined to try to keep this hidden from Hollie and Josh and want to give Hollie as normal a life for as long as we possibly can not just for Hollie but also for her brother. He is too young to understand what the future holds for his sister and for the rest of our family - grandparents, aunts and uncles who all find the diagnosis difficult to come to terms with.

profession to come along and learn a bit about Niemann Pick Disease. We have been amazed by the early response to this ball and we have already sold 200 tickets. If you want to know more about the campaign or the ball you can visit our website www.hopeforhollie.co.uk.



As for the future, we will take each day as it comes and hope that when the time comes we will be strong enough to cope with what we will have to face.

Helen Carter

Mum to Hollie aged 2 (NPC) and Joshua aged 6

In Loving Memory of Lucy Catherine Mathieson, 27/05/03 - 29/09/07



When Lucy was diagnosed with NPC at just five weeks old, the news had a devastating effect on our whole family, destroying our hopes and dreams for her future. Gradually, we came to accept that our longed-for child would not live a full life, and devoted ourselves to making sure that her precious time here with her family, and especially her big brother Jake, was as happy, enjoyable and comfortable as possible.

Lucy was the centre of our life, her huge blue eyes demanded your attention and her sunny personality shone through, even as the disease progressed and she could no longer manage to smile - her eyes smiled for her. She was calm, patient and inspiring; bringing so much love and laughter into our world.

When she was younger her cheeky smile and that naughty, infectious giggle were never too far away... and she had a wicked sense of humour...which belonged to someone many years older. She was seldom separated from her 'Peppa Pig' toy; we spent many hours pressing Peppa's tummy to hear her 'Oink' - and to hear Lucy giggle.

For someone so small she touched the hearts of so many people and no one knows what she may have achieved given the opportunity. Those of you who were fortunate to know and love her will have realised that she was a fighter. She suffered many symptoms of the disease during her short life, such as developmental delay, seizures, a lack of mobility and swallowing problems that resulted in her being tube-

fed. Throughout it all she remained a serene and happy child who was beautiful, both inside and out

The time since that Saturday night when she finally left us, after such a brave fight, has been the most terrible of all, the pain and the sense of emptiness - just simply knowing we will never see her again - it is just so hard to contemplate. We are comforted by the thought that she is now free from NPD and happily doing all those little girly things that this disease prevented her from enjoying - singing, dancing, skipping and generally doing what little girls do.

We learned so much from Lucy; she changed our lives in a very positive way and helped us to discover skills we did not know we had. She gave us strength and courage, patience and hope; she taught us how to smile when all we wanted to do was cry. Through her we met so many good people, who have since become great friends. You could say she brought out the best in us...made us appreciate what life is all about and gave us the ability to cherish each day.

In memory of Lucy, and to help others affected by this cruel disease, we, her family and friends, continue to do all we can to raise awareness of Niemann-Pick diseases and raise funds to support the work of the NPDG (UK). We want to bring hope to future families and believe that there will one day be light at the end of this long dark tunnel. By the way, Lucy's name means 'light' and she truly was -and always will be - the light of our lives.

Let Me Go

*We've known lots of pleasure,
At times endured pain;
We've lived in the sunshine
And walked in the rain.
But now we're separated
And for a time apart,
But you are not alone -
I am forever in your heart.
There may be times you miss me,
I sort of hope you do,
But smile when you think of me,
For I'll be waiting for you.
Now there's many things for you to do,
And lots of ways to grow,
So get busy and be happy,
Miss me, but let me go.*

Anon



Lucy

In Loving Memory

We remember

Carl Symes Age 20

Rachel Lewis Age 11

Lucy Catherine Mathieson Age 4

I'm Free

Don't grieve for me, for now I'm free
I'm following the path God has laid for me.
I took His hand when I heard him call
I turned my back and left it all.
If my parting has left a void
Then fill it with remembered joy.
Be not burdened with times of sorrow

I wish you the sunshine of tomorrow.
Perhaps my time seemed all too brief
Don't lengthen it now with undue grief.
Lift up your hearts and peace to thee
God wanted me now; He set me free.

Author unknown

Sibs

Sibs is the UK charity for people who grow up with a disabled brother or sister. They support siblings who are growing up with, or who have grown up with, a brother or sister with any disability, long term chronic illness, or life limiting condition. There are over half a million young siblings and over a million adult siblings in the UK. Siblings have specific needs that require attention at different stages of their lives, including relief of isolation, information, and strategies for coping with the situations they find themselves in. Their aim is to enhance the lives of siblings by providing them with information and support, and by influencing service provision for siblings throughout the UK. Their long term vision is that each local authority in the UK will have a dedicated sibling service, providing sibling groups and one to one support for children who are siblings.

Sibs aims to enhance the lives of siblings by providing them with information and support, and by influencing service provision throughout the UK.

They believe that siblings have a right:

- To be valued for who they are and what they do
- To be seen as individuals with specific needs
- To access reliable sources of advice and information, and support services
- To influence the services and policies that affect them
- To reach their full potential



Sibs can help in the following ways:

Young Siblings – are offered advice and ideas for coping with difficult situations and the opportunity to learn about the experiences of other siblings

Adult Siblings – can get information on planning for the future and on dealing with issues related to childhood

Parents – can find out about sibling issues and get tips on supporting siblings at home

Professionals – can learn more about sibling issues and how to support siblings through their work with families

Sibling Group Leaders can get information on running sibling groups and on developing their sibling support service

To find out more about Sibs visit their website www.sibs.org.uk or telephone 01535 645453

The Roald Dahl Foundation

The NPDG (UK) would like to thank The Roald Dahl Foundation for their generous offer of grant funding support in 2008/9. This will be the second time that the Group has received grant funding from The Roald Dahl Foundation and we are very grateful for their continued support. The grant is specifically to support the travel costs of our Clinical Nurse Specialist, totalling £5000 over two years.

The Roald Dahl Foundation is a UK-based registered charity offering a programme of grant-giving to charities, hospitals, and individuals in the UK only. They support many varied projects, in exactly the same way Roald Dahl did when he was alive, offering practical assistance to children and families in three areas: neurology, haematology and literacy. Since their creation in 1991, they have donated over £5.5 million in grants



National Development Manager Report



On September 29th 2007, our beautiful daughter Lucy, aged just four years old, lost her battle with Niemann-Pick Type C. The years since Lucy's birth have flown over in a whirlwind of emotion – joy, sadness, fear and frustration – ending finally in our acceptance of the disease and its devastating process. Acceptance does not make this journey any easier; it just allows you to focus on enjoying every treasured moment with your loved one.

Lucy was everything I had ever wished for and I am so very grateful for the time we had with her and the many positive lessons she taught us. Lucy made a lasting impression on everyone she met, managing to beguile them with her huge blue eyes and usually to inspire them into wanting to help in some small way. Losing her has only strengthened my resolve to help others dealing with this devastating group of diseases

and I believe we will one day find solutions to the many challenges they present.

My work for the NPDG (UK) continues to be challenging but it has helped me to have something to focus on during this difficult time in our lives. Please do not be afraid to contact me here at the office if there is anything you feel I could help with – I would be only too pleased to talk to you – especially if you were to ask about Lucy as talking about her will always be one of my favourite pastimes!

Toni

Toni Mathieson

National Development Manager

Telephone: 0191 415 0693

Email niemann-pick@zetnet.co.uk

Jackie's Journal

I would say Happy New year but as we are already in February, and you won't be reading this until March, it seems a bit late!

We have started the year well with what used to be called our "Toddler Clinic Day", but as the oldest is now 7 and the youngest 13 months, we have quite a mixture now. It is reassuring for the parents of the tiny tots to see the school age children running around and looking well. I must admit that Dr Wraith looked a bit wary when I told him that 17 children were attending, but after seeing the 14 that were able to attend he did survive! As usual I accompanied the families on their appointment with Dr Wraith, whilst Liz Jacklin (our Clinical Research Nurse) spent time getting to know everyone and answering any queries. Also invited to attend were a doctor and nurse from the London metabolic team; it was good to meet them and to show them how well the Clinics work. Lunch was provided and it must have been OK as not a sandwich or crisp was available when we went scavenging at the end of the day! Unfortunately, the weather did prevent a few from joining us, so we may decide to hold Clinic a bit later in the year in 2009. I am looking forward to hearing the thoughts and suggestions of those who did attend, as this will help us to further improve things for next year.

Following the Board of Trustees' recent decision to extend the contract of the Clinical Research Nurse, I am now looking forward to working with Liz Jacklin for a further two years. Most of you will be aware that both my post and that of the Clinical Research Nurse are funded by the NPDG (UK), with part funding for my post kindly provided by BBC Children in Need, The Foyle Foundation and The Roald Dahl Foundation. We are very grateful to these organisations for supporting the work of the Group



and are especially appreciative of the fundraising efforts of all our families and friends who, through their hard work, continue to ensure the continuation of these posts.

Plans are now well underway for our Annual Family Conference 2008. If you have been before and would like to make suggestions as to how we can tailor the

Conference to meet your needs, it is not too late, we would appreciate this. If you have never been before, call us and we can tell you what happens during the weekend or perhaps put you in touch with another family who will be happy to share their experiences. We hope to start this year with a get together over light snacks on Friday evening, enabling people to meet the Board of Trustees and staff, to put names to faces and to catch up with old friends and new.

This year we are veering away from science to concentrate more on caring so do tell us what subjects you would like to know more about and we will make sure your questions/concerns are answered.

This does not just apply to the Conference – I am around every day to answer any queries or concerns – either in the office or on the mobile. Even if you are unsure if your question is NPD related I am happy for you to call.

Looking forward to the phone ringing or to your E mails arriving.

Jackie

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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 Autumn Newsletter by 15th August 2008

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