



Niemann-Pick News

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



Men's Health Survival of the Fittest 2009 – Survivors Paul Bayliss, Jimmy Cowan, Kev Hallinan and Martin Ashurst

CONTENTS

Chairman's Chat	2	Help and Advice	25
Research News	3	Family Focus	28
Family Conference 2010	15	Update from the Central office	31
Fundraising News	16	Jackie's Journal	32

Registered Charity
Number 1061881

NP Clinical Nurse Specialist 0161 701 2414
NPDG(UK) 24 Hour contact number 0191 415 0693

Niemann-Pick Disease Group (UK)**Central Office**

Toni Mathieson

*Executive Director*Sue Lowe, *Administration Officer*

11 Greenwood Close

The Pastures, Fatfield, Washington

NE38 8LR

Tel/Fax: 0191 415 0693

Email: niemann-pick@zetnet.co.uk

Chairman:

Jim Green

Tel: 01592 713409

Email jimgee@zetnet.co.uk

Secretary:

Bill Owen

Tel: 01225 865 273

Email: Bill_owen43hotmail.com

Treasurer:

Richard Brooks

Tel: 01249 811311

Email: richard@ketta.com

Management Committee

Janice Brooks

Tom Brooks

David Roberts

Helen Carter

Patrons

The Rt. Hon. The Earl Cairns CVO CBE

The Rt. Rev. Dominic Walker OGS

Bishop of Monmouth

Dora Bryan OBE

Rt. Hon. The Lord Bassam of

Brighton

Prof. Martin Rossor MA MD FRCP

Guy Johnston

Nicholas Mathias ARAM

Denise Nolan

Medical Adviser

J.E. Wraith MB ChB FRCP

Genetic Medicine

Manchester Academic Health

Science Centre

Central Manchester University

Hospitals

6th Floor, St Mary's Hospital

Oxford Road

Manchester, M13 9WL

Clinical Nurse Specialist

Jackie Imrie RCN

(address as Medical Adviser)

Tel/Fax: 0161 701 2414

Email Jacqueline.Imrie@cmft.nhs.uk

Clinical Research Nurse

Elizabeth Jacklin RGN

(Address as Medical Adviser)

Tel/Fax: 0161 701 2967

Email: Elizabeth.Jacklin@cmft.nhs.uk

Chairman's Chat



Ah, spring at last, (well almost!) - a time of growth and renewal. Our Charity is also heading for a period of growth, thanks to the work of many people. We welcomed, at the end of last year, the award of a grant from the Big Lottery to help extend and develop our family support service. This service is made possible by the fund raising efforts of many and the award of the grant to help boost the service is a mark of the value placed on it.

This boost to the service is much needed as we find the calls on Jackie Imrie, our Clinical Nurse Specialist, and the other members of staff are increasing as the number of patients diagnosed increases. It is a core part of the work we do and we are delighted to be able to extend and develop this service which many families have benefited from.

Details of the development of the service are being finalised at present and we will announce them in the next newsletter.

Also in this edition there is news of the work going on in research into the causes and treatment of these diseases, activities in awareness raising and fund raising. On the latter

front it is just amazing what people are achieving in these times of financial difficulties. People are showing endless energy and creativity in tackling fund raising with very positive results.

It's not just the family support service that is growing and developing. News in this issue of a new date for our Family Conference 10-12th September 2010 and a development in the programme to include a professional's research and awareness raising day. This is something that has been talked about and requested for a number of years and now I'm delighted to say that we are going to manage to do it.

These developments and many others in the field represent progress and only happen because people are working so hard. If you feel you would like to join in please let us know. The more we do the more we can achieve.

I hope you enjoy this issue and that spring is warm and that growth is strong.

Best wishes

Jim Green

Research News

Research Report by Bill Owen, Research Co-ordinator



I am aware that there is no particular synchronism between our Newsletters being published and advances in research into Niemann-Pick diseases. As a consequence I often miss important study reports and repeat information already reported in earlier Newsletters. I also realise the problems faced by newly diagnosed families in attempting to find out what has gone before. To keep an ongoing narrative would be very difficult and would probably not mean a great deal without considerable editing. The information published in our Newsletters over the years however, is held in electronic format for the most part and, may be made available to those with an interest. This also applies to Research Reports that I prepare for the Board of Trustees meetings. Just mail me to discuss.

Research Progress

I suppose that the bad news is that the Ara Parseghian research conference in Arizona has been put on hold for the present but no reason has been given which leads to speculation. As I have previously stated this is the main event on the NPC research calendar so I am keeping my fingers crossed. On a brighter note, at home, we have seen the launch of an International Niemann-Pick Organisation at a meeting in Edinburgh which took place in October. It was very moving to see at first hand, the enthusiasm of delegates to cooperate in whatever ways help each organisation to fulfil their role in their own country and, to promote awareness and research internationally. We also held our own research meeting at the new St Mary's Hospital, Manchester in November and although the agenda proved too much for the time available, the opportunity for families to hear the issues surrounding research, whether cell/genetic or clinical proved to be a great success. It is hoped to repeat such sessions taking into account lessons learned from that first meeting.

Elsewhere, Rare Diseases UK, which is a organisation funded by the pharmaceutical industry and backed by

an all party parliamentary MPs, continues in its task of gathering information in support of production of a report which will be submitted to Department of Health early in 2011. This seeks to assist patients with rare genetic diseases in receiving the appropriate attention within the UK healthcare system.

The National Institutes of Health PubMed website continues to list abstracts of basic science and clinical research into the Niemann-Pick diseases. Each of these studies provides another brick in the wall of understanding. Unfortunately we have no idea how big the wall is!

Those who follow research into disorders affecting the brain will be aware of the difficulty of delivering therapeutic medicine to that organ. The blood supply would seem ideal in many ways as the blood vessels deliver nutrients to every cell in addition to removing waste or toxic products. However, the brain is very well protected from substances other than the small molecules eg oxygen, sugar, salt, and water, needed for cell nourishment. Other molecules are unable to cross the walls of the capillary vessels as these are lined with a continuous layer of endothelial cells, one cell thick. These cells are connected to each of their neighbours by their cell walls which contain tight junctions. This forms an effective barrier to most substances carried in the blood, and is called the blood-brain barrier (BBB). The brain is also protected by other similar barriers such as exist in the meninges membrane, the blood-CSF barrier, and also in the nasal epithelium. Whilst these barriers are generally effective against unwanted intruders, they are equally effective at repelling therapeutic medicines. A consortium of scientist meeting under the epithet Brains for Brains (B4B) are focussing their efforts on ways to design vectors which are able to cross these barriers. This work, in conjunction with small molecule therapy will be of great importance in treating neurological disorders.

Prevention: A Developing Landscape

Genetic testing for carrier status is nothing new and in populations known to be at risk, eg the Ashkenazi Jewish population or even the Niemann-Pick type C community in Nova Scotia, Canada, the problem of inherited disease has been addressed with considerable success. It may be argued that this is because of the high carrier frequency in relatively isolated communities. In the former case the isolation is religious and in the latter,

geographical, but what about the rest of us? Are we to accept that we are simply victims of chance or is there something we can do? Families affected by rare genetic conditions are only too aware of the impact of disease and, it is this, rather than gene frequency, that should be the focus of healthcare considerations. The technology is available and is affordable. The priority has not yet been recognised by healthcare services but the situation is changing.

I find the number of articles dealing with the identification of defective genes and testing in broader population groups is on the increase. In addition to what I have previously reported a number of interesting stories have recently provided an insight into new initiatives being taken.

In the USA more states are introducing newborn screening and expanding the range of genes to be tested which have the potential to cause disease. This is useful in some cases but will not cater for prenatal onset or rapidly aggressive conditions. In another report, a college professor is offering students tests for a panel of rare conditions on campus. These include Niemann-Pick (type not specified) and less rare conditions such as cystic fibrosis. Perhaps the most dramatic news is that a US company called Counsyl is offering home based tests for a panel of over 100 rare genetic conditions. An article recently appeared in BioNews on this subject, you can access the article by following this link:

http://bionews.org.uk/page_54617.asp

I am aware that there are concerns amongst healthcare professionals about this initiative, to the extent that it is on the agenda for discussion in the professional genetic testing community.

Other changes relate to the way in which genetic testing is managed within the UK at present. A proposal has been made to make the testing process an integral part of NHS mainstream specialisations. It is early days and I do not understand the implications for very rare disease conditions. It is clear though, that a forecast for the growth of genetic medicine has been recognised and it will alter the way that the NHS does business. I believe we will hear more on this in the near future.

An intriguing topic called genetic genealogy was reported in the press. This involves going back through historical records to identify disease causing genes in families and then to track forward into the present to find long lost relatives who may be at risk. A researcher at Utah University is a pioneer of this approach and has conducted a 10 year study of modern populations with a specific form of bowel cancer using family histories and genealogy databases. It has been possible to track back 16 generations to trace the origin to a migrant family in the UK in 1640 (a typical founder effect situation).

Developing the initial family records through time has resulted in many branches of the family, over 5000 individuals, being identified. These individuals are now aware of their carrier status and can be monitored for early signs of polyp growth.

There are other examples of this approach and no doubt there are many limitations for application to rare conditions as I am finding out by attempting to trace my father's line back beyond a few generations. It would be helpful to have a research team on hand to do this and to make contact with living relatives in order to warn them of the potential risk. Being able to do this raises other issues; would these relatives welcome the warning or would they prefer not to know? Perhaps some of these have already experience the disease but would be unaware of their relationship to me? It is all very difficult and working alone has its drawbacks.

Genetic Testing and Research

In order that a prevention programme can achieve an acceptable level risk reduction, it is important to understand the range of disease causing mutation in the population. To date we do not have an effective way of testing the mature protein product for functionality and, this results in complex problems relating to gene sequence interpretation.

For example finding a mutation in the first instance is technically difficult. About 40% of NPC patients have one mutation unidentified although disease will have been diagnosed by clinical presentation, histology and biochemistry.

Even where the gene sequence predicts a functional amino acid sequence, changes called silent mutations can result in protein production which is incorrect (exon skipping), untimely (non standard RNA folding patterns), or lacking in quantity (insufficient dose). In addition, so called cryptic mutations may be harboured within intron sequences causing truncated protein. These introns are not sequenced in a genetic test. There are other sources which may contribute to disease such as the cell's quality control system, transport of a synthesised protein to its site of operation, post translational modification where, say, sugars are added and interaction with other, unknown proteins in a metabolic pathway. Problems may also arise in the chromosomal arrangement in the post fertilised egg cell and subsequent cell divisions (mitosis). I suspect there are many other issues of which I am completely unaware, all of which, require research attention.

The above may seem a bit 'techie' but it highlights the need for a better way of testing genes and their protein products. In NPC a library of cellular defects already exists and this continues to grow with biomarkers, storage and trafficking defects being identified. These

defects can be reversed or normalised, by the introduction of 'wild type' protein or can be modified by therapeutic agents eg miglustat. Surely this points to a new research approach to be considered for the near future which will remove many of the uncertainties of the present methods.

Family Matters

In my own family and that of my wife, we are slowly making progress in carrier testing and for most of those tested to date it has been good news in that no family mutation has been found which means that they are free from that particular, but specific burden. Where a mutation has been found, then those family members are now aware of the risk involved to their own children and future generations. The knowledge of carrier status now resides with those members and that it where it ought to be.

The process of accessing a genetic test is variable even within regions as is the time it takes to get the initial stages of discussions with GPs and genetic counsellors out of the way. I have mentioned before that the target time to conduct a test in the laboratory is defined

nationally and so far post test briefing has been simple requiring a letter advising on carrier status. I suspect that the next phase where partners of known carriers request a test will prove altogether more problematic. There are to the best of my knowledge, only three laboratories in the UK that offer NPC testing and I am not aware of any offering type A/B tests.

One thing I have learned is that if you have any documentation relating to a genetic test for any form of Niemann-Pick, make sure that you don't lose it. Although the lab that carried out the test will have records, I am not clear whether these are included in medical records held by your GP. Concerns have been raised about including genetic information on medical records, eg because insurance companies are able to access these and issues may arise if a claim is made; this is especially relevant in the USA, but how much quicker could diagnosis of rare conditions such as Niemann-Pick be if this data was known to the GP? Life is full of compromise and the ultimate decision should rest with individuals and families. If official records do not do the job then the families may, if they so wish.

BioNews

BioNews is the free weekly news digest of the top stories in assisted conception, genetics, embryo/stem cell research and related areas, published by the Progress Educational Trust. Sent to registered subscribers each week, BioNews by email is aimed at informing debate in these areas by providing balanced and timely summaries of the week's news and developments alongside comment, reviews and recommendations of selected topical conferences, events and more.



Visit the BioNews website at www.bionews.org.uk, where you can subscribe for free to receive BioNews by email in one of three formats, plus view more news, comment, reviews and job advertisements and search the full archive.

NPDG (UK) Research Nurse Update, Elizabeth Jacklin RGN



World Lysosomal Storage Disease Symposium 2010

*February 10th -12th
Miami, Florida, USA*

I recently attended the 6th WORLD (We're Organising Research into Lysosomal storage Disorders) Symposium held in Miami, USA. My attendance at this meeting was sponsored by Actelion Pharmaceuticals

I went along primarily to present a poster entitled 'Long Term Clinical Outcome of 12 Patients with Niemann-Pick Type C treated with Miglustat'. There was a lot of very positive interest in the poster, which you can view on the NPDG (UK) website, and it is now planned to extend this work into a paper for publication. We also hope to look further at the use of severity scoring systems.

In addition to the meeting itself, which included many interesting and relevant presentations, a selection of which will soon be available to view on our website, and the poster presentation, this trip was a very useful opportunity for networking, raising the profile of the NPDG (UK) and my role as Research Nurse.

NPC Research Meeting

Following on from the success of the NPC Research Meeting which was held at the Royal Manchester Children's Hospital on 1st December 2009, we would like to hold another meeting of this kind. Thank you to all those who contributed to the meeting, we have received some useful feedback and this will assist us in organising future events. We now need to decide on a suitable time and venue for the next meeting. Please let us know when and where you would prefer the meeting to be held – for instance, would midweek or the weekend be more suitable for you? Is Manchester a convenient location, or would you prefer a different location? Your suggestions will be most welcome and helpful. We are hoping the meeting will take place in May/June.

Zavesca – FDA Advisory Committee Meets in the USA

On January 12th 2010, an FDA Advisory Committee meeting was held in Maryland, Washington, USA. The Committee was convened to discuss the application for Zavesca (Miglustat) to be used for Niemann-Pick type C patients in the USA. You may recall that the drug was approved by the European Medicines Agency, for use in Europe, in January 2009. Actelion Pharmaceuticals, the company who developed Zavesca, asked that I attend the hearing on behalf of the NPDG (UK) as an 'expert' witness. The Committee were required to vote on a number of points throughout the day, the most critical being "In the light of safety and efficacy data presented in this application, does the risk/benefit of Zavesca support its approval for treatment of NPC?"

Following a tense day of presentations and questions, I am delighted to report that the Advisory Committee voted 10 to 3 in support of approval. Whilst this does not guarantee a positive outcome, the FDA will meet again in March this year to make a final decision, the Advisory Committee's recommendation may mean that the application is looked upon more favourably. The success of this meeting was due in no small part to the presentations made by, and on behalf of, NPC families; Barb Vorpahl, Chair of the Niemann-Pick Disease Foundation, Cindy Parseghian, President of the Ara Parseghian Medical Research Foundation and Phil Marella, Trustee of Dana's Angels Research Trust, all gave tremendous presentations. I was given the opportunity to speak on behalf of the NPDG (UK) and our UK families on several occasions. I am delighted that the NPDG (UK) was able to be involved in this process.

Please note: I regret to inform you that since writing the above article, we have heard via the NNPDF that the FDA has chosen NOT to approve Zavesca (miglustat) for the treatment of NPC in the USA at this time. The FDA has requested further data to support this application. We will keep you informed of further developments.

Clinical Studies Group for Inherited Metabolic Diseases

Research into rare inherited disorders is poorly funded and often regarded as not scientifically significant due to the small amount of patients involved. To get larger numbers it is important to collaborate on studies

The UK Lysosomal Disorders (LSD) Patient Organisation Collaboration has worked together with the Medicines for Children Research Network, to form an Inherited Metabolic Disorders (IMD) Clinical Studies Group (CSG). The objectives of the group are to improve the research profile for patients with rare diseases. The vision is to develop a comprehensive and collaborative research programme in IMD, utilising the expertise of the clinicians working in IMD, the support of the UK LSD Collaboration, other charities for IMD and the MCRN, together with others involved in IMD research. This will permit the investigation of important diseases affecting children and adults, and the testing of both new and existing interventions based on a nationally agreed and scientifically robust research agenda. In achieving this, there will be an improvement in both knowledge and quality of care.

The CSG for IMD consists of the following members: 4 paediatric consultants in IMD, 1 adult consultant in IMD; 1 clinical psychologist, 1 physiotherapist, 1 pharmacist, 1 consultant from the devolved nations (Scotland, Ireland and Wales) and a representative from the UK LSD collaboration.

I have been nominated to represent the UK LSD Collaboration. Other members from London, Birmingham, Manchester, Liverpool and Glasgow will be representative of the community caring for those affected by Inherited Metabolic Disorders.

The Board also requires two LSD family members to represent the whole LSD community from a patient/family perspective. If you feel that this might be something you are interested in being involved in then please contact me – my contact details are given below. It is hoped that the formation of this group will encourage the development of research protocols into Inherited Metabolic Diseases, and enable a greater number of patients to access and participate in such studies.

If you are interested in learning more about the Inherited Metabolic Disorders Clinical Studies Group please do contact me.

As always, I would be happy to talk to you about any research issue or question you may have, you can contact me by telephone, 0161 922 2967, or email Elizabeth.Jacklin@cmft.nhs.uk

Liz

NPDG (UK) Clinical Nurse Specialist invited to Speak at Actelion Meeting in Lausanne Switzerland, 10th January 2010

Every winter Actelion Global arrange for as many representatives from Actelion Worldwide to gather together in Switzerland for team building, brain storming and working together on International projects. They are there all week spending many hours in the meetings room.

One of their major projects revolves around improving International networks to improve diagnosis and ultimately offer treatment to more patients with NPC. They are initially concentrating on targeting those hospitals that have good facilities, both clinical and scientific for NPC and then those hospitals that feed in to those centres.

But why was I there?

Actelion Global feel that as far as networking goes, the UK is very much a leader. We have excellent centres that care for families with NPC and with 79 patients currently

diagnosed have a huge cohort compared to most other European countries so they feel we must be doing something right. My talk aimed to show how the centres worked together, how the Niemann-Pick Disease Group UK is a vital part of that network and where my role fits in. They all asked many questions and very important contacts were made. It was a long way to go for a short time but feedback from several countries that were represented shows it was certainly worthwhile.

Jackie

Jackie Imrie RCN
Clinical Nurse Specialist
Niemann Pick Disease

Phone/fax: 0161 701 2414

Mobile: 07932 737417

E-mail: Jacqueline.Imrie@cmft.nhs.uk

First International Meeting of Non-profit Niemann-Pick Organisations



The First International Meeting of Non-profit Niemann-Pick Organisations took place in October 2009. The meeting was held in Edinburgh and was jointly hosted by the NPDG (UK) and the National Niemann-Pick Disease Foundation in the USA.

The objectives of the meeting were:

- To establish links between all non-profit Niemann-Pick support groups and to raise awareness of the nature and operations of these different organisations.
- To hear updates on the most recent developments in science and therapeutic opportunities.
- To investigate ways of maximising communication,

economising on the use of resources and providing mutual support for each other.

- To discuss future opportunities for collaboration and accelerating progress.

Each organisation was asked to send three representatives: including two senior representatives of the organisation plus a clinician from that country, or associated with that organisation, who had a special interest or expertise in the Niemann-Pick diseases. Although this was not a scientific meeting, it was attended by clinicians, scientists and representatives of pharmaceutical companies who are currently leaders in the field of NPD. In addition, there was a representative of the Gaucher's Association, their Executive Director, Tanya Collin-Histed, who was able to provide an overview of their own experiences in forming an international network.

The meeting programme was designed to provide opportunities to network, to update and inform, and to explore ways in which organisations may be able to support each other in order to facilitate progress. It was a mixture of meetings, presentations, and social opportunities to meet and share experiences and friendship.

In all, 46 delegates attended, from nine organisations located in Argentina, Canada, France, Germany, Holland, Italy, Spain, the UK and the USA.

The meeting achieved much for individuals and organisations. There was unanimous agreement on the following:

- The formation of an international alliance of NPD non-profit organisations. The "INPDA" "International Niemann-Pick Disease Alliance"
- A co-ordinator was appointed for an initial period of two years (J Green – Chairman of NPDG (UK) was elected as the first co-ordinator).
- Goals were set for the first two years
- Longer term goals were discussed and a possible series of long term outcomes set down

The Next Steps

Each organisation will contact the Co-ordinator to confirm that they wish to become members of the INPDA and will appoint a representative.

The first tasks for the Co-ordinator will be to establish a website, and once nominations have been received, organise a first meeting.

To read the full report of the meeting please visit the NPDG (UK) Website:

http://www.niemannpick.org.uk/what_is_npd/inpda.html

The NPDG (UK) and the NNPDF would like to acknowledge the support of all those who helped to make this meeting a success:

- Actelion Pharmaceuticals and Genzyme Corporation for their grant funding for this meeting
- The steering committee for their tireless efforts and hard work in organising the meeting
- The guest speakers who contributed so much time and expertise
- All attendees who helped make the meeting such a success



SOAR – NPC

Support of Accelerated Research for Niemann-Pick Disease Type C

In the Spring 2009 issue of Niemann-Pick News Newsletter, we provided information about SOAR - NPC (Support of Accelerated Research for Niemann-Pick Disease Type C), a collaborative research project which was formed in 2007 and is funded by Dana's Angels Research Trust, The Addi & Cassi Fund, The Hadley Hope Fund and The Hide & Seek Foundation for Lysosomal Disease Research. The goal of SOAR is to accelerate therapy development for Niemann-Pick Disease Type C, a neurodegenerative disorder in the lysosomal disease family.

Recently, our Clinical Nurse Specialist, Jackie Imrie attended the Brains for Brain Workshop in Frankfurt, Germany, where she was pleased to hear Dr Steve Walkley give an overview of SOAR's work. Dr Walkley has kindly given permission for us to reproduce the following extract of his presentation in our Newsletter for your interest:

Support Of Accelerated Research for Niemann-Pick type C disease

- The SOAR-NPC was established in 2008 as an academic research-family partnership designed to accelerate development and clinical testing of therapies for NPC disease.
- The SOAR Collaborative consists of an international group of scientists and physicians with expertise in basic and clinical NPC disease research
- Is the only consortium in the world dedicated to the study of this disease.
- The SOAR-NPC has close ties to the NIH (ongoing natural history study and clinical trial; high throughput testing of potential compounds -NCGC).
- Additional investigators and their laboratories are recruited to assist the SOAR Collaborative in its discovery process as the need arises.
- A key feature of the Collaborative is urgency: To develop a therapy that could significantly ameliorate clinical disease progression within 5 years .

These are the 4 investigators who, with the main aim of fulfilling the last point above, have teleconferences every 2 weeks.

- Yiannis Ioannou, PhD (Mt Sinai School of Medicine, NY), widely recognized for his contributions to understanding the molecular function of the NPC1 protein.
- Daniel Ory, MD (Washington University School of Medicine, St. Louis, MO), important contributions to our understanding of the role of the NPC proteins in regulation of cellular cholesterol homeostasis.
- Frances Platt, PhD (University of Oxford, UK), developed miglustat for the treatment of type 1 Gaucher disease and showed its utility in mouse models of the gangliosidoses.
- Steven Walkley, DVM, PhD (Albert Einstein College of Medicine, Bronx, NY), expanded understanding of the role of gangliosides in NPC disease and pioneered the use of substrate reduction therapy (miglustat) as a therapeutic approach for this disorder.

This team is supported by additional investigators

- Forbes Porter, MD, PhD and Nicole Yanjanin, R.N., M.S.N. (NIH-NICHD). Dr. Porter is the Principal Investigator of the NPC Natural History Study at the NIH, an ongoing protocol to improve our understanding of this disease and to serve as a resource for providing clinical samples to SOAR-NPC investigators.
- Charles Vite, DVM, PhD (University of Pennsylvania, Philadelphia, PA) has a long-standing interest in neurodegeneration in feline and canine models of genetic disease, with specific focus on feline NPC disease.

And additional collaborators

- Chris Austin, MD and Wei Zheng, PhD are, respectively, the Centre Director and the Group Leader for Cellular Signaling Assay Technologies at the National Chemical Genomics Centre (NCGC) at NIH. Broad experience in small molecule high throughput testing and development for treatment of disease.
- David Begley, PhD (Kings College London, UK) is carrying out studies on the ability of cyclodextrin to penetrate the blood-brain-barrier in normal mice and mice with NPC disease.

Also, a very important component is the Research Advisory Committee (RAC)

The RAC is an independent body, whose members include leading experts in rare disease and lysosomal storage disease research, and in drug development.

- John Curd, MD, President and Chief Medical Officer of Threshold Pharmaceuticals; formerly with Genentech, Maxygen and VaxGen. Expertise in

design and execution of clinical trials to test safety and efficacy of new drug.

- William Gahl, MD, PhD, Head Section on Human Biochemical Genetics and Clinical Director NHGRI at NIH.
- Elizabeth Neufeld, PhD, Professor Emeritus in Biochemistry at UCLA. Dr Neufeld is an internationally recognized expert in lysosomal storage diseases
- Marc Patterson, MD, Chairman of Pediatric Neurology at the Mayo Clinic. Leading authority on diagnosis and care of NPC disease
- William Sly, MD, Chairman of Biochemistry and Molecular Biology at St. Louis University. Dr. Sly is a pioneer in the study of lysosomal storage diseases.

Twice a year, ALL members of the collaborative, investigators, scientific advisory board, families and co-ordinators meet face to face.

SOAR have very specific aims

- Perform high-throughput cell-based screens for identification of small molecules with NPC disease-modifying potential.
- Evaluate in NPC animal models candidate compounds that emerge from small molecule screens, as well as candidate FDA-approved agents and compounds from natural libraries.
- Provide continued support for the NPC natural history study at NIH to acquire and curate human sample collections for biomarker discovery.
- Identify NPC disease biomarkers using cell-based assays and mass spectrometry-based proteomic and lipidomic approaches, and validate markers in animal models of NPC disease and in tissue/fluid samples from NPC subjects.
- Evaluate utility of serum-based biomarkers for early detection of NPC disease.
- Validate biomarkers in human NPC clinical trials.
- Develop a drug cocktail that successfully ameliorates or eliminates progression of neurological disease in NPC patients.

And these are their achievements so far -

- Developed in vitro screening techniques to facilitate high throughput testing of novel compounds.
- Completed in vivo screens on 15 high priority drugs and nutraceuticals in NPC mouse models.
- Performed in depth analysis of most promising compound (cyclodextrin) identified to date in NPC animal models.
- Identified potential biomarkers for NPC disease progression.

- Helped facilitate the initiation of a clinical trial for NPC disease at the NIH using N-acetylcysteine (NAC).

And finally Dr Walkley explained why all of this work is so important, not only for families dealing with NPC:

- Focuses a great deal of talent on finding a viable treatment for NPC disease – and to do so as quickly as possible.

- Could serve as a model system for development of similar research collaboratives focused on other lysosomal diseases (e.g., Sanfilippo, MLIV disease, CLN3 disease, etc.), or on other types of rare diseases where there are patient advocacy groups or foundations and identifiable labs with an interest in their disorder.

With grateful thanks to Dr Steve Walkley for allowing us to reproduce this information

Testing Times

By Helen Carter, Mum to Joshua, age 8, Hollie, age 4 (NPC) and Lucas, age 7 months

When we received Hollie's NPC diagnosis in 2007 we were devastated. Our hopes and dreams for Hollie had been destroyed and we were terrified of what the future would hold for her and our family.

It is so difficult to come to terms with a diagnosis of Niemann-Pick but then realisation also begins to dawn that this is genetic condition and not only has your child been afflicted with a cruel terminal illness but also that any future children we may have planned could also be at serious risk of being struck by the disease. What was once a relatively easy decision for us, as we had always planned more children, had turned into a frightening one. Could we and should we have more children?

All these questions filled my mind, should we just forget the idea of another child, would people judge us badly, would they think we were in some way trying to replace Hollie, were we being selfish to even consider having another baby, were we doing right by Hollie. More and more questions, some completely irrational but I guess all questions that other parents in our position have asked in the past.

The decision to have another baby didn't come lightly. Firstly we looked into the options open to us. We knew that in order to test accurately and quickly we needed to know both of the disease making mutations that we had passed down to Hollie. The biggest hurdle for us was waiting for the labs to find Hollie's second mutation. They had found the first which my Husband had passed down but the second was proving more difficult.

Having researched the options before my husband and I were both in agreement on one thing, if we were to have another baby we would have pre-natal testing whether this be by Chorionic villus sampling ("CVS") or Pre-implantation Genetic Diagnosis ("PGD"). We felt we owed it to a future child and to the rest of our family to make an informed choice and not to gamble with that child's future. I know this is a very personal decision and therefore I am sure some reading this may have differing



opinions on whether to test or not but whatever decision a family comes to one thing will be for sure, it won't be made lightly.

Once we had made the decision to try for another baby we looked into the options available to us. PGD was a relatively cutting edge procedure for diagnosis of NPC, which involved a cycle of IVF. Although this was a real possibility, we felt that funding on the NHS may be an issue especially as we had an older son, who although untested appears to be clear from the disease, and this may reflect their decision whether or not to offer funding. It would be a waiting game and we may have to fight. We didn't want to risk the emotional stress of fighting for funding and the risk of being turned down many months later. In the end we decided to opt for testing via CVS which is normally carried out around 13 weeks of pregnancy. At that point we knew it was imperative to find that second elusive mutation. I put in a quick call to Jackie Imrie, the NPDG (UK) Clinical Nurse Specialist, to explain our decision and to see whether she could get the lab to prioritise looking for the second mutation. However there was no need, five minutes later we got the call we so wanted. Jackie had checked with the lab and by coincidence they had already found the mutation that very same week. We now had the information we needed and we could start trying for a baby.

I still remember vividly the day the blue line came up on the pregnancy test to confirm I was pregnant. The fear immediately struck me as did the enormity of the situation. From that moment onwards we knew our baby's fate had already been decided and it was just a waiting game to find out whether or not the odds were in our favour. I burst into tears, partly tears of joy and partly tears of fear.

At that point in time I was unclear of what the next step was, who should I contact and what did I need to do? I therefore contacted Jackie once more and asked her to help. Jackie was able to contact our local geneticist at Oxford John Radcliffe Hospital and I was contacted immediately by the nurse who was assisting the geneticist. She made an appointment for us to meet with the geneticist at our local hospital at the next available clinic and she also talked me through the process once I had seen her. In the meantime, I was to visit my GP and ask him to book me in for an early dating scan, as they needed to establish the exact number of weeks of pregnancy, which would enable them to carry out the CVS at the safest opportunity. A CVS cannot be carried out any earlier than 13 weeks of pregnancy due to the risk to the baby. I was given a choice of hospitals and we opted to go back to Kings College Hospital in London as this was a hospital we knew well because of Hollie's liver disease. We had also researched the unit and it was one of the top in the country for carrying out such procedures with a lower percentage rate for miscarriage.

When we met with the geneticist she took our family history, explained the 1 in 4 chance of having an affected child with NPC and that there was a 25% chance the child would be unaffected, a 50% chance of being a carrier, and 25% chance of having NPC. Although technically the odds were stacked in our favour at the time it really felt more like Russian roulette. At our request she referred us to Kings College for the CVS procedure. This was carried out at 13.5 weeks but by then it felt far longer and the fear of the unknown was just as daunting as waiting for the skin biopsy when Hollie was going through the process of diagnosis.

The CVS is a relatively straight forward procedure. Whilst an ultrasound scan is carried out a needle is placed through the uterus and a small piece of the placenta is removed for testing. It's an uncomfortable procedure but is over before you know it. There is a 1% risk of miscarriage after the procedure but they did reassure us that normally if there is a problem it is pretty immediate and apparent from the follow up ultrasound carried out after the procedure. Our scan thankfully showed a healthy heartbeat. That's not to say that the next few days I didn't spend most of my time worrying myself silly. Luckily I had a midwife appointment a few

days later and the sound of the baby's heartbeat was enough to finally reassure me all was well.

After the CVS, the now very familiar waiting procedure begins. The CVS was transferred from Kings College to the Willink in Manchester and the transfer was the longest bit and most stressful which I am sure Jackie, and Liz, NPDG (UK)'s Research Nurse, can vouch for! Kings were not used to testing for Niemann-Pick. CVS is usually carried out to test for Down's Syndrome and other chromosomal abnormalities, where tests are carried out in their own labs at Kings, so on this occasion they were not entirely sure what was required and where it needed to be sent! Again Liz and Jackie came to the rescue and it was finally delivered safely to the Willink labs in Manchester. Once at the Willink I knew it wouldn't be long to wait but time still went by so slowly. It felt like I was never off the phone chasing and that's probably because I wasn't!!! I cannot thank the Willink enough though. After all the delay in getting the CVS to the Willink they pulled out all the stops to make sure I didn't have to wait much longer.

There are two phone calls which will remain in my thoughts forever, one was the call I received about Hollie having Niemann Pick and the second was the call I received from Jackie one afternoon whilst sat by the computer. I knew the call was coming as I had been told it was likely the results would be in that day. I promised myself I would be brave and hold it together but as soon as I heard Jackie's voice and she asked whether I was alone or whether my husband was with me I just crumbled. All the fear had built up inside and I was expecting the worst news. Then I heard the words "it's all ok, the baby isn't affected, you can enjoy the rest of your pregnancy". After that, I think I was unable to speak and I just burst into tears. Jackie had to call back later once I had calmed down! At that split second I felt like the luckiest person alive! The first call after that was to tell my husband and the second was to Kings to find out what the sex of the baby was! They had asked me a few days earlier if I wanted to know if it was a boy or a girl but I didn't, not until I knew everything was going to be ok.

I would like to say the rest of the pregnancy was relaxed but I would be lying. I felt like I was carrying such precious cargo. If I could have stayed in bed and wrapped myself in cotton wool I probably would have!! My biggest fear was whether he would be jaundiced when he was born. I knew he didn't have NPC but you can't help being a little bit irrational about these things.

Our little boy wasn't going to hang around though, he knew when he was expected to arrive and he made sure he did. On his due date, the 5 August 2009 we welcomed our new addition, our beautiful precious little boy Lucas Max Carter into the world weighing 8lb 1oz.

Was he jaundice, yes a few days later taking him for his first walk round the park' we commented that he looked a little bit yellow but that soon disappeared the next day and so did our fears.

Josh and Hollie have a gorgeous baby brother who they absolutely dote on. He is now seven months and I can't believe how quickly the time has passed. We couldn't be happier and we feel extremely blessed. Everyday he gets an extra special squeeze x

IVF with Pre-implantation Genetic Diagnosis for NPC – Hope for our Family

When our first son was diagnosed in 2007 with NPC at 4 months old, part of the horror was the impact NPC would have on our future family. We'd always imagined a large family. S was conceived quickly and we enjoyed a straightforward, care free pregnancy. When he became unwell with liver problems, at 5 weeks, we had no concept of what lay in store.

Options

Quite soon after the NPC diagnosis, we started to look into what options were available for further children. These were:

1. To decide not to have any more children. This felt unbearable. The idea of losing him was unthinkable, to be left with nothing to live for.
2. To conceive naturally and have prenatal diagnosis (PND) during the pregnancy (involving chorion villus sampling). At this point, we didn't feel that we would be able to terminate a child knowing that it was affected with NPC, or run the risk of losing a healthy child through miscarriage because of invasive prenatal testing that carries risks in itself.
3. To have a pregnancy without prenatal testing, taking the decision to live with the risks and consequences of another child affected by NPC. A one in four chance is high by anyone's standards and it didn't feel like good odds.
4. To adopt a child. We briefly looked into this, and it seemed that it may not be possible, because we would have an existing child with a fatal and progressive illness. Would we have the physical and mental energy to meet the demands of the adoption process and the challenges that come with bringing a child, who perhaps has their own troubles and difficult past experiences, into our family? If S was unwell and needed us, surely he would become our priority? Quite rightly no adoption panel would approve us if we were unsure ourselves.
5. To try Preimplantation Genetic Diagnosis (PGD) using in vitro fertilisation (IVF). PGD is a technology which allows genetic testing of an embryo prior to implantation and before pregnancy occurs. It is used in conjunction with IVF and allows only those embryos diagnosed as being free of a specific genetic disorder to be transferred into a woman to try to achieve a pregnancy.

Choosing PGD

This last option held some hope for us. I don't know what we would have done in terms of risking a natural conception, because PGD offered an alternative - an option that doesn't force an impossible decision about a pregnancy, and also doesn't enter us into the roulette of taking our chances. IVF was something we'd heard of and knew of other couples who had had it. It seemed like it could work. It gave us a chance of having an NPC free child, without invasive testing during pregnancy, and the possibility of aborting a foetus. However it didn't, and still doesn't, sit entirely comfortably. In carrying out IVF, embryos are created. They may be only 3 days old, and made of just 8 cells, but they still have the potential for life. And I still struggle with that.

The cost of PGD/IVF is extremely expensive, over £10,000 for the first cycle including development of the testing probe (which must be developed individually for each NPC family as it is dependent on the individual mutations in the gene), and around £5,000 for any subsequent cycle.

In our case, both our mutations in the NPC gene have been identified. If only one mutation is known, PGD/IVF is still a possibility, although the chances of success are lowered.

To arrange funding, we requested that our GP apply on our behalf to our local Primary Care Trust (PCT). It is down to individual PCTs how they fund services. However, the Department of Health provides guidelines relevant to the funding of PGD*:

- Each case should be considered individually.
- Risk of child affected by the disorder is greater than 10%.
- Eligibility criteria for IVF are usually strict, for example, not funding couples with existing children. Applying these criteria may not be appropriate for couples requesting PGD as their circumstances are different to infertile couples.
- Consideration should be given to the family structure, with greater priority given to people with no living or no healthy children.
- A limit to the number of cycles funded would be appropriate. The first cycle is the most costly due to the development of a genetic probe; a further 1-2 cycles would increase the chance of a pregnancy and make best use of the probe.

**See Department of Health publication from the Genetics Commissioning Advisory Group:*

http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4118934

We felt we ticked every box, and our PCT agreed, and initially approved 1 cycle of treatment. They replied to us within a few weeks. It's worth noting that the funding guidelines for PGD/IVF are different to straight IVF for fertility problems. While your PCT may not fund IVF, it may still fund PGD/IVF.

We also attended an appointment with a Genetic Counsellor, to ensure that we understood fully all the options and associated risks.

With funding in place, we were able to start planning treatment. We chose Care Nottingham, a private fertility clinic that also treats NHS funded patients, one of a half dozen or so centres in the UK licensed by the Human Fertilisation and Embryology Authority (HFEA) to conduct PGD. The HFEA is the UK's independent regulator overseeing fertility treatment, and approves the conditions that PGD may be applied to.

Treatment

I spoke to the PGD embryologist at Care Nottingham, who was incredibly helpful, understanding and knowledgeable. We had an initial consultation, which explained what the treatment entailed, the risks and possibility of success – 22% at the start of treatment. However, PGD/IVF isn't widely used, and each clinic has its own data and predicted success rates.

PGD/IVF basically follows the standard IVF procedure, with an extra embryo testing step added in. The idea is to get your ovaries to produce the highest number of good quality eggs.

Fertility drugs are used to stimulate the ovaries, and each individual has a specific drug regime, or "Protocol", which is self-administered. I followed the standard Long Protocol. This meant daily hormone injections over a few weeks to "down regulate" my system, shutting my ovaries down. Then daily injections of stimulating drugs to kick start egg production, with ultrasound monitoring, for a couple of weeks. Then eggs were recovered using a vaginal ultrasound procedure. W provided a sperm sample at the same time, and a single sperm was injected directly into the centre of each egg (Intra-Cytoplasmic Sperm Injection – ICSI) to achieve fertilisation.

The embryologist then telephoned us daily to provide an update. Typically, if, say 14 eggs are recovered, 12 may be mature (and ready to be fertilised), 9 may fertilise, 7 may make it to Day 3 (3 days post egg collection), etc. In other words, there is an expected tailing off in numbers of embryos remaining. Unfortunately, we only recovered 4 eggs in the first cycle. All 4 fertilised successfully. On Day

3, each embryo consists of 6-8 cells. One cell is taken from each embryo and tested for NPC using the specially developed testing probe. On Day 4, we received the results: 2 were affected, and 2 were paternal carriers – just like W, so despite carrying one copy of the affected gene, would not develop NPC. On Day 5, the 2 paternal carriers were transferred into my womb. The transfer specialist used a speculum (like the one used for a smear test) and the embryologist loaded the embryos into a catheter to be placed in to the womb, the process taking around 15 minutes.

We then had to simply wait. I took it easy for a few days, but carried on largely as normal. We were due to take a pregnancy test 14 days following embryo transfer. However, 8 days later I started to bleed, and despite several days of bed rest, we knew it hadn't worked.

That it didn't work came as a real surprise. I think we naively thought that because we'd conceived naturally with S, IVF would be an inconvenience, but it would work. No one knows why those first embryos didn't implant, it just sometimes doesn't happen. By the time you get to embryo transfer stage, you have around a 60% chance of success – good, but no guarantee.

So we had a review appointment with the IVF specialist. We requested further funding with our PCT, and they agreed to fund a further two cycles. We had to wait three months before we could try again, to allow my body to return to normal. In the second cycle, the level of stimulating drugs was increased, to try to get my body to produce more eggs. So, injections again. I didn't enjoy self-injecting but it was manageable. You have to give them at the same time each day so I got quite efficient and could do it in restaurant toilets, on the train, in the park! It's hard to distinguish physical side effects of the hormones from the emotional. I definitely felt that my body was not in my control and I didn't like the feeling. But it was a means to an end. The biggest inconvenience was really the travel to and from Nottingham, and the requisite time off work. However IVF is definitely not a straight forward or easy option, and your body can feel assaulted by the effects of the hormones. I think I got off relatively lightly, but I know of other women who are completely floored by the process and need to take most of the 6 weeks treatment window off work.

The second cycle was less successful than the first. Again, 4 eggs were retrieved, just 2 fertilised, but both tested positive for NPC, which meant there were no embryos to transfer back. We were devastated. Another review appointment was held with the IVF specialist, with the suggestion that, because I wasn't responding to the stimulating drugs sufficiently, producing only 4 eggs as opposed to 12-15 eggs, I had an underlying fertility issue. Fertility problems have all kinds of horrible

negative terms, like “dysfunctional maturity”, “poor responder”, “reduced ovarian reserve”. It was even suggested that S’s conception may have just been lucky. We really didn’t feel lucky, and this was a very low point. Not only did I pass on a defective gene, I couldn’t even produce enough eggs to give us a chance to fix it.

We didn’t know what to do next. Producing just 4 eggs isn’t enough to give PGD a chance of success. Given that it’s expected that all won’t fertilise, that not all will divide, and some will be affected, it’s really important that the maximum number of eggs can be recovered. We had an appointment with another local fertility specialist, really just to get a second opinion. They agreed with Care Nottingham, and it looked unlikely that PGD was going to work for us.

But we had one further funded cycle, so we decided to try it again. It seemed we had nothing to lose. I was given the maximum dose of stimulating drugs, and I also had some acupuncture sessions (in for a penny, in for a pound!). On scan day, where an ultrasound is performed to see how many eggs are developing, we were not hopeful. However, we each prayed to God that although we wanted it to work we put it in His hands, and prayed that whatever happened, we would be able to deal with it and understand that He may have a plan for our lives that we don’t yet know or appreciate. You could have knocked me over with a catheter when they told me there were lots of eggs developing very nicely!

12 eggs were recovered, which gave the embryologists so much more to work with. We had two embryos transferred back, and some unaffected embryos were suitable for freezing – this means they are in storage and have potential for life in the future. It’s a hard concept to get your head around.

Following transfer, we again had to wait two weeks before taking a pregnancy test. I became an avid knicker-checker, constantly worried that I would bleed and it would all be over. I rested as much as possible, and had further acupuncture to try to relax and convince the embryos to hang on in there.

And now, I’m 19 weeks pregnant with twins. I still can’t believe it, life travels on a narrow track with joy on one side and devastation on the other, and there’s just a fraction of a moment dividing the two. I wouldn’t say we’re jubilant. We’re incredibly pleased, but cautious until they arrive safely – because although we know they are NPC free, the PGD process does not rule out other conditions. We feel incredibly blessed, because we know

there are other families who never had this option and who won’t ever have the chance. It doesn’t take away the devastation of NPC. It doesn’t make everything right, and it doesn’t make S OK which is truly the only wish I have. It’s incredibly hard to reconcile in my mind that I’m carrying two babies who won’t have NPC, but I have S, my boy, who does. And that I can’t fix it for him. And I feel so very sad that it was us, our combination of crappy genes, that made this happen to him. I don’t know how I’ll feel when the babies come. I don’t know how we’ll cope if S gets unwell and we have two other children to care for. Sometimes I feel so very selfish to have embarked on this at all. Surely I should just dedicate my life to caring for him. I couldn’t want for more, he is everything I could ever dream of, so I feel guilty that I’m asking for more, and that these children could distract me from him.

But then I see S playing with other children, and I know it’s right that he should be a big brother. That he should have siblings who care for and love him, and who he loves back. That there should be other people in this world that love him and cherish him the way that W and I do. And we need some happiness. It really is selfish I suppose, but we really do. And I know that I will love them with the same passion and overwhelming intensity that I do S, and that once they are here I won’t be able to imagine life without them, and that their presence in our lives will bring us joy that we can’t begin to envisage now. And I know we’ll cope with whatever happens because we can, because even with our crappy gene combination we’re stronger together than anything we will face.

If you want to try PGD:

1. We’d be happy to talk to you directly about our experience, contact the NPDG office to arrange
2. The HFEA website is a good source of information on PGD, see <http://www.hfea.gov.uk/preimplantation-genetic-diagnosis.html>
3. Speak with a genetic counsellor to fully understand the risks, implications, etc, get referral from Dr Wraith / your local consultant / or GP
4. Search for a clinic licensed for PGD, and call them to discuss. The HFEA website has a search function, go to <http://guide.hfea.gov.uk/guide/> . Most clinics list a PGD coordinator on their website. Consider using a clinic that has experience of NPC or similar genetic conditions, not just one that is geographically close
5. Visit your GP and, if appropriate, request funding from your PCT

“Worry does not empty tomorrow of its sorrow, it empties today of its strength”

Corrie Ten Boom

NPDG (UK) Annual Family Conference 2010

10th – 12th September 2010

The Robinson Executive Centre, Wyboston Lakes, Bedfordshire

“Pathway to Progress”

“A journey of a 1000 miles starts with a single step”, Lao Tzu

The NPDG (UK) Annual Family Conference and AGM has moved to September this year, over the weekend of 10th – 12th, when we hope to welcome many familiar and friendly faces, as well as those attending for the first time.

The programme for Conference 2010 is currently being finalised, and will once again feature Speakers from across the world, including clinicians, researchers, scientists and our own NPDG (UK) families and staff members. We have taken into account feedback received following last year’s event to ensure that we include something for everyone, including the ever popular Breakout Sessions and Family Voices, as well as the usual – and very important – updates regarding research and clinical care.

This year’s programme will also feature a comprehensive programme for ASMD Niemann-Pick Type A & B families, including discussions and presentations regarding the clinical trial of recombinant human acid sphingomyelinase (rhASM) as a potential treatment for ASM Deficiency.

The Children and Young Adult’s Activity Programme will be as exciting as ever! Liz Jacklin, our own Clinical Research Nurse, will once again Co-ordinate the programme this year, along with Alison Hitchens and Ged Rowles – of course they couldn’t do it without the help of their team of experienced volunteers! For more information about the

Programme, please contact Toni or Sue at the Central Office on 0191 415 0693 or email niemann-pick@zetnet.co.uk

Although plans for Conference 2010 are well underway, it is not too late to send us your thoughts and ideas as to how we can tailor the conference to meet your needs. So, if you are thinking about attending and would like to see a particular speaker or subject covered, please email your comments to sue.npdg@tiscali.co.uk we would be glad to receive them.

Further information regarding this event will be available soon. If you would like to register your interest beforehand, or if you have any questions about the Conference, please contact the NPDG (UK) Central Office on 0191 415 0693 or email niemann-pick@zetnet.co.uk 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.

As well as hearing the latest important information, this exciting weekend is all about sharing experiences and strengthening our family support network, we do hope that you will be able to join us for our 17th Annual Family Conference.

Janice

Janice Brooks, Conference Chair



The Big Lottery Reaching Communities – Success!!

Family Care and Interactive Support Service

The Niemann-Pick Disease Group (UK) is delighted to announce that our work is to be supported by a grant from the Big Lottery Fund Reaching Communities Programme over the next three years. We would like to thank the Big Lottery Fund for choosing to support our organisation with the grant, for the amount of £172,601, which has given us the fantastic opportunity to enhance our current support service with a new project, entitled the 'Family Care and Interactive Support Service'.

The overall aim of the project is to improve outcomes for families with Niemann-Pick diseases, through the use of interactive technology that will enhance access to support and information services. The symptoms of NPD can often appear, or change, suddenly, as the disease progresses; in consultation with families and healthcare providers, the NPDG (UK) learnt that, although families do get to see, and feel supported by, their local GP or Consultant when they have concerns, it is not always possible to describe or replicate the symptoms or behaviour that was displayed in the home environment. Using appropriate interactive technology, such as webcams and media links, families will have increased access to expert care and advice provided by NPDG (UK)'s Clinical Nurse Specialist for Niemann-Pick Disease, Jackie Imrie, enabling them to achieve a more timely response to their questions and concerns.

This project will provide specialist care and support that will assist families in coping with the challenging symptoms of the disease, enable them to communicate more effectively with their health care team, and will



LOTTERY FUNDED

tackle the issues of isolation and despair, that so often accompany a diagnosis of a rare disease, by creating links with other affected families.

The project will start in May 2010 with a pilot scheme lasting six months. We will shortly be seeking five families who would be willing to participate in the pilot scheme, which will help us to determine the potential benefits of this enhanced support service, plus the most effective methods of communication and appropriate equipment. Following the pilot scheme we aim to make this project accessible to all those who wish to participate.

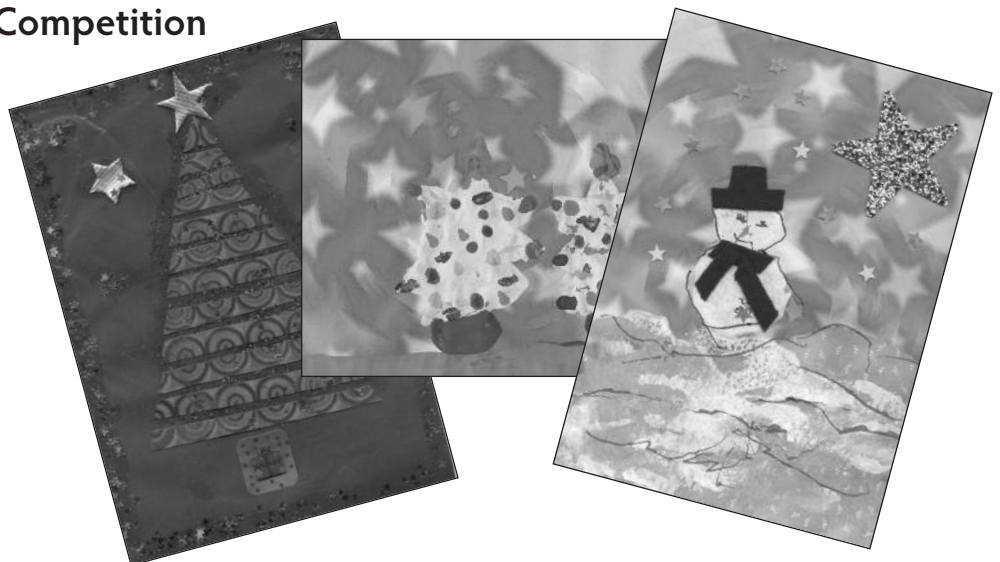
It is our hope that the innovative use of technology at the heart of this project will enable more effective and efficient communication between families and our Clinical Nurse Specialist, and will lead to an improved quality of life for those affected by Niemann-Pick Disease and their families.

If you would like to know more about this project, or if you would be interested in participating, please contact Toni Mathieson at the NPDG (UK) Central Office by telephone 0191 415 0693 or email niemann-pick@tiscali.co.uk

Fundraising News

2009 Christmas Card Competition

Thank you to all who generously supported the Group by purchasing our 2009 Christmas cards, which were once again very popular thanks to the wonderful designs of our budding young artists; Annie Pyne and sisters Alice and Ellen Byrne. The total amount raised was an amazing £1,900!



Nissan Motor Manufacturing (UK) Ltd – A Thank You

The NPDG (UK) would like to acknowledge the generous donation of £2000 received from the Nissan Employee Christmas Saving Fund. The NPDG (UK) was nominated to receive this donation by employee Will Woodhouse, of Washington Tyne and Wear. We would like to thank Will and all of his colleagues at Nissan for their support.



Gala Concert at Champ's Hill

Tony Jellings, former Trustee and Board member continues to do much to support the fundraising efforts of the Group. Tony writes:

On 5th December 2009 a concert, sponsored by Sotheby's of London, was held at the home of Mary and David Bowerman, in their splendid Music Room at Champ's Hill, Sussex, to raise funds for the Niemann-Pick Disease Group (UK).

To add to the interest of the occasion, David Bowerman, our host, had arranged an exhibition of paintings from his collection of 20th Century painters to hang on the walls of the music room. Before the Concert started, these were briefly discussed by Sotheby's Head of Modern Paintings.

The concert was given by one of our patrons, international cellist Guy Johnston, his brother, violinist Magnus, pianist Tom Poster and viola player Tom Hankey.

The musicians played a medley of Mozart, Mendelssohn, Chopin and Brahms with great brilliance and received rapturous applause from the audience of 150 guests.

During the interval Tim Ingles, Head of Sotheby's Music Department, showed the audience the "Ex Hamma"



Stradivari violin of circa 1717, valued at \$2,000,000, which he had brought along with him. Magnus Johnston gave a demonstration of the instruments superb quality.

Our Chairman, Jim Green, who had travelled down to Sussex from Scotland to attend the Concert, addressed the audience briefly before William Lucy, a Senior Director of Sotheby's, auctioned a lunch for eight persons, donated by the company and to be held at the Sotheby's Board Room in London. This was sold to a successful bidder for £1000.

The evening raised a grand total of just under £7,000.

I wish to thank Mary and David Bowerman for their hospitality and great generosity, Sotheby's for their sponsorship and in particular Jenny Denman, Sotheby's representative on our planning committee, all the members of my family who helped to make and serve the delicious refreshments, all those who supported the event, including Richard and Marion Rogerson, who travelled down from Birmingham, and finally, but by no means least, the four brilliant and inspirational young men who once again gave of their time and very considerable talents to support the Niemann-Pick Disease Group (UK). THANK YOU!!



School Supports NPDG (UK) Throughout 2010



Denton West End Primary School, Manchester, has chosen the NPDG (UK) as their adopted Charity for 2010, following a presentation made by Alex Jacklin, age 7. Alex, a pupil in year 3, decided to give a presentation about the NPDG (UK) to his classmates when the opportunity arose for pupils to decide which Charity the School would support throughout 2010. Alex's presentation was chosen from four others in year 3 to go forward to be presented to the whole school in a special assembly. Following all six presentations, the children then voted for the Charity they most wanted to support, and thanks to Alex, the NPDG (UK) won! So far, the pupils and staff have raised almost £1000 through their efforts – the NPDG (UK) would like to thank all who have contributed so generously, and also say a special thank you to Alex, for his hard work!!

Hope for Hannah

The Men's Health Survival of the Fittest

10th October 2009 in Nottingham

We must have been mad.....!! To volunteer for a gruelling 12km run which included a number of obstacles such as running up and down the stairs at Nottingham Forest football club, swimming across a river, and tackling an assault course....!!

What a fantastic day had by all....the weather was excellent, the atmosphere was superb. Already we are considering going through the pain again next year!!

The survivors are as follows:

Paul BAYLISS, Jimmy COWAN, Kev HALLINAN, Martin ASHURST

We would like to thank everyone who supported us in raising £400 to support Hope for Hannah and the NPDG (UK)..

Paul Bayliss



The 'Hope for Hollie' Campaign

Maisy supports the 'Hope for Hollie' Campaign



Maisy Dyer and her friends raised almost £130 for the 'Hope for Hollie' Campaign when they organised and ran a stall at their School Christmas Fayre selling cakes and gifts. Well done girls!



The Milton Keynes Baby & Toddler Fair 2010

The Hope for Hollie Campaign has been selected by the Milton Keynes Baby & Toddler Fair as their featured Charity in 2010. Helen and Pete Cater were delighted to learn that the Baby & Toddler Fair will be supporting the Hope for Hollie Campaign and the Niemann-Pick Disease Group (UK), after the Campaign was chosen from many other worthy causes. The show will house around 60 exhibitors, and will feature a fantastic mix of classes, activities and entertainment. Look out for more information coming soon!



Hollie's Halloween Ball!!

October 2010

Saturday 23rd October 2010, StadiumMK, Milton Keynes

Watch the Hope for Hollie Campaign Website for further information coming soon!!

www.hopeforhollie.co.uk



If you would like more details about the 'Hope for Hollie' campaign, visit their website www.hopeforhollie.co.uk

Campaign for Calum

Who wants to be a Millionaire?

On Sunday 20th December Colleen and Maureen Nolan appeared on 'Who wants to be a Millionaire' on ITV 1. Maureen opted to play for two charities, one of them being the Niemann-Pick Disease Group (UK). Maureen raised the fantastic amount of £12,500 for the Group, and the sisters also helped two other worthy causes.

The Nolan sisters have been a great support to the Group over the past eighteen months, with Denise Nolan becoming a Patron of the Group in Summer 2009, following her appearance at the 'Campaign for Calum' Charity Variety Show held in January 2009. Denise first met Calum Burdon, who has Niemann-Pick Disease Type C, then age 4, and his parents, Carl and Emma, at the Variety Show. Since then, Denise and her family have helped to raise awareness of the disease and funds for the Group by holding several events, including a Christmas Carol Concert.



Sue French recounts the story of a spectacular fundraising effort by her friends...

"On Sunday, 27th September our great friends from university Jeremy and Helen (aka Uncle Jeffa and Auntie Hells Bels) ran the New Forest Marathon to raise funds for the Niemann Pick Disease Group in the UK. Dave and I along with William and Rebecca went down to support them and look after their three children Martha, Louis and Seth. It was a beautiful sunny day (in fact a little too hot for running a marathon) and the 800 odd runners got away just after 10am. Our job was to hand out the jelly babies at various points along the way and provide encouragement. First stop was 5 miles. Despite having the luxury of driving, we missed Hells at this point as she was already setting a great pace but just managed to catch Jeff.

9 miles was the next planned stop, but having taken a wrong turn we had a scenic tour of the countryside instead and met a wild boar to boot. Back on track we found the 15 mile marker, this time managing to cheer Hells on up the hill. Jeff wasn't far behind but the jelly babies were proving popular with the kids, so with too few in hand, Dave had to run up the hill after Jeff to hand more over! We decided to try and get to the 19 mile marker, but found Hells already at 21 miles and decided we needed to get back to see her finish. They were setting a great pace!

Hells finished 26th overall, the 4th woman home only missing out on a top 3 finish by 1 minute. Her time was a staggering 3 hours 14 mins. Jeff wasn't far behind,

finishing in 202nd, completing the marathon in a very respectable 3 hours 56 minutes.

Before the Marathon, Jeff had set up a Justgiving web page to raise some funds and we were overwhelmed with the support that we received from our friends and families, in particular the friends of Jeff and Hells who supported us so generously despite not knowing our family personally and who had previously never heard of the Niemann Pick Disease before. As I write this article, the total amount raised, including gift aid, is over £15,000 which is an amazing total. We are looking forward to working with the NPDG to put this money to



good use to help set up trials in the UK and finance research. We would love this to be the starting point for a Research/Trial fund to be set up within the society that we can target fund raising towards and hope that we can help find a treatment for this disease that devastates our lives. It would be great if UK families had the option of participating in future trials in the UK without having to travel to the US.

William had a fantastic day with his friends and wrote a lovely thank you to his Aunty Hels Bels and Uncle Jeffa. I couldn't put it better than he did when he said: "Thank

you very much for helping us by running the race. You were awesome!"

We are so grateful to Jeff and Hels for dedicating their time and effort to the Niemann Pick Disease Group (UK), and Dave and I would love to follow in their footsteps next year - whilst a marathon would be a little ambitious, we hope a 10K may be possible. Anyone up to join us?

*David, Sue, William & Rebecca
French*

So You Want to Run a Marathon?

NPDG (UK) has secured one place in the 2011 London Marathon. If you, or anyone you know, are interested in running the 2011 Marathon on behalf of the Group, please contact either Toni or Sue at the Central Office to register your interest before 30th May 2010. We will be holding a ballot on June 1st to decide who the lucky person will be!

Just in case you are interested, Sue Lowe has put together the following information for potential Marathon Runners... the full article can be found on our website: www.niemann-pick.org.uk

The definition of a marathon is a race that is 26.2 miles. To put it into perspective – if you were to cover the distance by car at 60mph it would take you half an hour drive, let alone to run it! But don't despair – it isn't as difficult as it sounds! You just need to approach it with the right attitude, one step at a time; and if a marathon feels like a step too far, why not start with a half marathon?

"Jenny and I were really nervous and wondering why we'd put ourselves forward for this event. However everyone was really friendly and it was lovely seeing all the different charities represented." *Susie and Jenny Elliott – ASICS British 10k London Run July 2009*



Prepare Yourself!

Before you begin training for a marathon, a half marathon or if you have just decided to take up running to get fit, it is recommended that you pay your doctor a visit for a once over.

Don't worry about how far you can or can't run to start with you just need to get your body used to running. So, if you cannot run for at least 30 minutes without stopping it is important start gently with combinations of run/walks; not only will this ease your body into exercise but it also minimizes the chance of experiencing a running injury. It's also a good idea at this point to select an event and get signed up!

"First was London in 2007, 2008 New York, and in 2009 I competed in the Dublin Marathon"

Jon Beaumont



There are many marathons and other running events worldwide. You will need to decide whether you want to run in a large or small event, and if you want to travel to a fun location or stay close to home. You can find listings and reviews of marathons all over the UK at <http://www.marathonrunnersdiary.com/races/uk-marathon-list.php> Details of the main worldwide marathons are listed here

<http://www.myworldmarathons.com/index.html>



"Once again the run was a fantastic day, the weather was perfect for both runners and spectators and I smashed my time from last year by over 5 minutes" Rob Unsworth – BUPA Great Manchester Run May 2007

Get Down to Business...

- The most important thing you should invest your time and money in is a good pair of running shoes, good trainers are essential.
- When running in cold weather, wear layers. A t-shirt under a zip-up sweatshirt and a simple pair of running pants should be sufficient.
- In hot weather, wear light coloured clothing and a light coloured hat to protect you from the sun. Also, don't forget the sunscreen!

Make sure you eat right:

- Eat meals that are high in carbohydrates and low in fat and don't forget the lean proteins (chicken, fish, pulses).
- Eat fruits and vegetables, but be sure not to have too many the day before the race
- Drink plenty of water, even on days that you're not running.
- It's a good idea to eat a small snack and have a glass of water about a half-hour before you run. Carbohydrates are usually the best choice, with dairy being the worst.

Train... Train... Train...

These steps apply equally to half marathons - 10K races - 5K races or any other form of running including just increasing your recreational running:

- Achieve a basic fitness level – you should be able to walk, run or bike or any other aerobic activity for a period of 30 minutes or more.
- Pick your training plan: There are a number of great training plans online: You will want to find one that fits you and your schedule.

"I hadn't run more than a mile since leaving school so with nothing more than some old trainers we started to build up the miles. Eventually after 6 months of calf strains, blisters and swelling knees



we had convinced ourselves 26 and a bit miles could almost be achievable". Carl Hitchens – New York Marathon 2008

Motivation

Building mental stamina is essential. It's one thing to be motivated to begin training. It's another to stay motivated every day...

- Motivate yourself - even going out for a shorter run is better than not going out for a run at all.
- Stretch. After a 5 -10 minute warm-up jog to get your muscles ready, do a thorough 10 - 15 minute stretch of all the muscle groups in your legs
- Keep a slow pace - Many people make the mistake of running too fast and then burning out;

Preparing for the Race

A few weeks before the race....

- Organise your running gear and everything else you will need before, during or after the race
- Find out how far apart the refreshment stations will be located and plan how often you will need to stop.
- Plan your journey to and from the race venue in advance, allowing extra time in case there's heavy traffic or delays.
- Ease up your exercise programme for a few weeks before the race. If your body is overworked before you even begin the race, you're more likely to encounter problems.
- Make an extra effort to eat healthily in the weeks before the race. It's also important to stay hydrated.

The day before the race...

- Go for a gentle run the day before the race. A two or three mile run won't use up all your energy before the big day, but it will help relieve tension.
- Eat a healthy, balanced meal the evening before the race. It should include protein, carbohydrate and some fat. Treat yourself to a dessert too. It's best to

eat a tried and tested meal you know will agree with your body.

- Read a good book or watch your favourite film on the evening before race – it will take your mind off the day ahead and help you relax.

The morning of the race...

- Get up early to give yourself time to prepare for the day ahead.
- Eat a light breakfast
- Have a shower with plenty of soap on the morning of the race, it will remove oils from the body, which will help you stay cool and prevent you dehydrating too early on in the race.
- Apply surgical tape or grease (for example Vaseline) to sensitive parts of the body before you run, especially if you're prone to rubbing or blistering.
- Take some warm clothing with you because it may be cold while you're waiting to start the race.

"When the race finally started, the adrenalin soon kicked in and with an amazing crowd we were soon buzzing with excitement. The local children were keen to give us "high fives" and the crowd shouted my name, printed on my Niemann-Pick T-shirt."
Helen Roberts - New York Marathon 2008

"It was a beautiful day, a little too hot, but the atmosphere was very good – I ran with the Niemann-Pick logo on my back and am pleased to say several runners asked about the Group"
Amanda Martin - Swindon Half Marathon 2008



During the race...

- Warm up at the start line, but be careful not to overdo it – be gentle!
- Make sure you don't wear any new clothing or footwear for the first time on race day - it's important you feel comfortable and secure in your running gear.
- Split the race up into 5 mile or 5 km sections as you make your way around the course. It'll feel less daunting than counting down mile by mile, and as

you complete each section you'll really feel you're making progress.

- Stick to an even pace you can keep up throughout the race. If you rush off at the start, you'll find yourself struggling to keep up further down the line.
- Sip water at every drinks station, but not in excess.
- Feel confident that you'll make it to the finish line.

Know how to pace yourself – Remember, you have trained long and trained hard for this marathon and you owe it to yourself to enjoy it – this Is Your Day!

"It was a great day and many of my family were there to watch" *Dave Buckingham – London Marathon 2007*

"The marathon was an amazing experience. The crowds spur you on and keep you going when you feel like your legs have nothing else to offer." *Leigh Price - London Marathon 2009*



Following the race...

it is important to replenish lost nutrients and don't forget to stretch as it will stop you feeling so sore the days following your run.

"Completing the New York Marathon was one of my favourite achievements and a truly amazing experience". *Helen Roberts – New York Marathon 2008*



For further information contact Sue Lowe at the NPDG (UK) Central Office on 0191 415 0693 or email sue.npdg@tiscali.co.uk

Thank You Freckleton Sports and Social Club!

The Campaign for Calum and the NPDG (UK) would like to thank the staff and customers of Freckleton Sports and Social Club for their amazing fundraising efforts over the past year. Throughout the year, and through many different events, their efforts raised the fantastic amount of £7140.96, which includes a generous donation of £250 from West End Football Club. We are very grateful to all who contributed so generously!



Niemann-Pick Awareness Week 2010: 19th – 25th July 2010



Our 7th Annual Awareness Week is fast approaching! We very much hope that those of you who have joined in the fun in previous years will once again help to raise awareness of Niemann-Pick diseases and/or funds to support the work of the Group by holding an event to mark Niemann-Pick Awareness Week 2010.

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!

Alternatively, you could choose to raise awareness amongst the medical and social care professionals in your local area – we can provide you with information to hand out, or if you prefer, we can send these out on your behalf. Just let us know who you would like us to contact. 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, teddy bears 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 Many items in the pack can also be downloaded from our website www.niemannpick.org.uk

Awareness Week is your chance to make a difference in the fight against Niemann-Pick disease – so please do get involved – we need your help to make Awareness Week 2010 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!

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 and 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. 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>

Sue

Sue Lowe, NPDG (UK) Administrative Officer
sue.npdg@tiscali.co.uk

Many thanks 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 about fundraising, or a fundraising pack, please contact us at the office on 0191 415 0693 or email niemann-pick@zetnet.co.uk .

Help and Advice

In case of emergency – 'ICE'

East Anglian Ambulance Service have launched a national "In case of Emergency (ICE)" campaign, with the support of Falklands war hero Simon Weston and in association with Vodafone's annual life savers award.

The idea is that you store the word "ICE" in your mobile phone address book, and against it enter the number of the person you would want to be contacted "In Case of Emergency".

In an emergency situation, ambulance and hospital staff will then be able to quickly find out who your next of kin are and be able to contact them. It's as simple as that, and for more than one contact name you can use ICE1, ICE2, ICE3 etc. Please pass this information on to your family and friends.

Home Access – Get on Line

Home Access is a Government scheme that will help low-income families who have children age three to nine to get access to a computer and/or broadband internet connection at home

You may be eligible for a grant if you are in receipt of one of the following benefits:

- Income based Jobseeker's Allowance
- Income support
- Child Tax Credit but not Working Tax Credit and an income of less than £16,040
- Guaranteed Pension Credit (not Savings Credit)
- Free school meals for your child

- Income-based Employment Support Allowance

Carers and foster parents of looked after children in years 1 to 13 may also be eligible for this grant.

The grant will allow eligible applicants to buy one of the following packages:

1. A computer, one year's internet access, service and support.
2. A computer with service and support only.
3. One year's internet access only.

For more information about the Home Access Grant Scheme call their helpline on 0333 200 1004 or visit their website www.homeaccess.org.uk

Useful Resources

IPSEA – Independent Parental Special Needs Advice



IPSEA is a voice for children with special edu-

cational needs. They are an independent charity, started 25 years ago, to help parents to get the special education that their children need. The SEN system can be very complex and it can be difficult for disadvantaged families to ensure that their child's needs are met. The majority of parents will need some help with the overwhelming volume of paperwork that securing their child's support entails. Lots of parents can find dealing with the Local Authority intimidating. With IPSEA's help, thousands of children have maximised their potential by receiving the special education that they are entitled to.

IPSEA have a small team of staff and volunteers to support and advise parents. They provide a unique service – free, expert, independent advice for every

child – no matter what their SEN and / or disability.

You can visit their website for:

- Model letters
- Support sheets
- Guidance on SEN Law
- SEN news

www.ipsea.org.uk

You can call their advice line, which is covered by a fully trained IPSEA volunteer adviser five days a week, including some evenings. Their volunteers can support parents in the following ways:

- Improving the way that schools meet children's needs
- Requesting an assessment of a child's needs
- Planning for an annual review
- Dealing with the exclusion of a child with SEN

IPSEA's advice line number is 0800 018 4016

Fledglings

Fledglings is a national charity which aims to assist parents and carers of disabled children, or those with additional needs of any kind, by identifying, sourcing and supplying practical, affordable products to address every day issues.

Their approach is to focus on finding solutions to difficulties which arise as a consequence of the disability, rather than on the disability itself.



Visit their website to view the wide range of items available for children with additional needs www.fledglings.org.uk or telephone 0845 458 1124.

Useful Websites

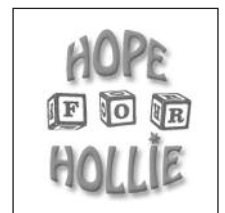
Have you joined the 'Hope for Hollie' Forum?

The 'Hope for Hollie' Campaign e-forum enables families affected by NPD to share their thoughts and experiences of living with Niemann-Pick disease. All aspects of the disease can be discussed, with contributions from families of people affected, carers, experts, and also discussion about fundraising activities.

We will shortly be introducing a new Research Blog onto

the forum – Liz Jacklin, NPDG (UK) Clinical Research Nurse, will provide a regular update of recent research news and will answer any queries or questions you may have in this area.

To join, visit www.hopeforhollie.co.uk or follow the link on the NPDG (UK) Website – www.niemannpick.org.uk.



Benefits News

Missing Millions

This year, local authorities are getting the final part of an extra £34m that families all over the UK campaigned for, to be spent on transforming services and support for disabled children and young people. Every council is getting a share of the remaining £11.3m and for Scotland's Disabled Children (fSDC) coalition has launched a campaign, We want our Missing Millions, to persuade local councils to spend it on families with disabled children in their area. But the coalition needs your help to succeed. So if you are a

parent/carer, disabled child or young person, or professional/practitioner and want your share of the Missing Millions, get involved! Visit the website <http://www.fsdco.org.uk/how-can-you-help/find-the-missing-millions> gives more info about the campaign, how to find local councilors /MSPs, details of each council's share of the £11.3m and downloadable campaign materials. There are also campaign postcards – order them at info@fsdc.org.uk or on 0131 659 2939.

Extra Child Trust Fund payments for disabled children

From April 2010 the government will start to make extra payments into the Child Trust Fund accounts of disabled children. The extra payment will be £100 per year, or £200 per year if a child is on the care component of Disability Living Allowance at the highest rate.

Will all disabled children receive these extra payments?

No. In order to qualify for an additional payment from the government your child must have been in receipt of Disability Living Allowance (DLA) at some point in

the previous year. In addition, only children born on or after 1st September 2002 have Child Trust Fund accounts.

My child gets DLA. What steps do I need to take to ensure my child receives these extra payments into their account?

The government expects to automatically identify those children who have both DLA and a child trust fund and will then make a payment directly into each child's account. Parents will receive a letter telling them once a payment has been made.

When will my child be able to get the money in their account?

A child must normally wait until they reach 18 years of age to access the money in their account. However if your child has a terminal illness and their death could be

reasonably expected within six months, you can get early access to buy things that your child needs.

Phone the Contact a Family helpline for further information (freephone 0808 808 3555).

Carer's Allowance earnings limit to increase from April

Carer's Allowance is the only state benefit specifically aimed at carers. However in order to get Carer's Allowance one of the rules is that your earnings must be no more than £95 per week. This earnings limit has been unchanged since October 2007. However from 6th April 2010 the government is to increase this figure to £100 per week.

How are my earnings calculated for Carer's Allowance?

In working out your weekly earnings certain deductions can be made from your gross wages. For instance any tax and national insurance you pay is deducted, alongside half of any pension contributions you make.

What about if I have to pay someone to look after my children while I am at work?

If because of your work you have to pay someone else to care for the person you look after, or to look after your children, you may also be able to deduct these costs from your earnings. However the maximum amount that you can deduct for alternative care costs is 50% of what would otherwise have been your earnings. No

deduction is allowed if the person you pay is a close relative.

These rules may allow some carers to qualify for Carer's Allowance even though they are earning slightly more than £100 per week.

What are the other Carers Allowance rules?

You must be at least 16 to claim and you can only get Carer's Allowance if the person you look after is in receipt of the care component of Disability Living Allowance at the middle or highest rate or Attendance Allowance (a benefit for elderly people). You cannot claim if you are a student involved in 21 hours or more supervised study.

If you are looking after a disabled adult then in certain circumstances an award of Carer's Allowance could lead to a reduction in that disabled person's benefits.

Parents with disabled children who are working and who want to know if the change in the earnings rule will help them claim Carer's Allowance should call Contact a Family's free Helpline on 0808 808 3555.

New benefits rules – child maintenance payments ignored from April 2010

From 12th April, child maintenance payments will no longer be treated as income when working out if you are entitled to means tested benefits such as income support, income based job seekers allowance or income related Employment and Support Allowance. These payments are already ignored as income for housing and council tax benefit and for tax credits.

If you receive child maintenance payments and were told in the past that your income was too high to receive a means tested benefit, you may find that these new rules allow you to qualify for the first time. To find out whether this applies to you seek further advice. If you care for a child with a disability or illness you can get detailed advice from the Contact a Family Helpline on 0808 808 3555.

Cash Counts: A new online service from Contact a Family

Families with disabled children face enormous financial challenges. They incur additional costs caring for their child – childcare, heating, transport, home adaptations and equipment. It can also be difficult for families to hold down a job due to the demands of caring. Despite this, many are not claiming the benefits they are entitled to.

Cash Counts is a new online service from Contact a Family designed to provide families with disabled children with the latest financial advice and information that is specific to them.

Cash Counts includes:

- An online benefits calculator – so families can work out how much they are entitled to
- A top tips guide on benefits written by the Contact a Family helpline people
- A 'frequently asked questions' page to help families through the complex benefits system.

Visit: <http://www.cafamily.org.uk/cashcounts>

Counting the Costs 2010 – a Contact a Family survey of families with disabled children's finances

Finances can be a major concern for families with disabled children.

In 2008 Contact a Family asked families how they were doing financially and the results were shocking, with one in six reporting that they were going without food and heating.

Two years on, Contact a Family wants to find out how families are coping in 2010, to demonstrate if, and how things have changed.

Srabani Sen, Chief Executive of Contact a Family, said: "In these turbulent economic times everyone is struggling. But it costs three times more to raise a child with a

disability and families with caring responsibilities have greater difficulty working, so are more likely to be living in poverty.

"There is a lot of talk that the economy has turned a corner and things are beginning to improve. But we would like to know what the experience of families with disabled children is.

"Please take the time to fill out our short online survey, which will take no longer than five minutes to complete."

Counting the Costs 2010 survey: <http://www.surveymonkey.com/s/T2S2ZZF>

The closing date for the survey is 30th April 2010.

Family Focus

Lois Peacock opens a New Supermarket in Loughborough

During a recent stay at Rainbow's Children's Hospice, Lois Peacock, age 12, NPC, was recently asked to open a new Tesco Supermarket near to their home in Loughborough. Lois went along with her family, mum

Amanda, dad Roger and brother Luke. They met the Store Manager and Lois cut the ribbon to officially open the store for business!



In loving memory of: Annette Hediger

Dear Annette

On the 6th of January 32 years ago you opened your shining eyes for the first time.

Together with your brother Markus you spent a joyful and light-hearted childhood. You enjoyed the kindergarten and primary school here in Rickenbach and you made many good friends. As a family we experienced many wonderful events and adventures on bike tours, holidays at the sea side and hiking in the mountains.

When you were eleven we realised that you didn't speak as fluently as before and we contacted a doctor. After that several checkups and examinations by doctors, and in the hospital, followed. In spite of all these inconveniences you remained a happy and lively girl.

You visited the Teeny Club and the Sunday School and Jesus Christ became very important in your life. We know and we are convinced that he carried you through all the difficulties and hard times you had to face in your life.

A friend invited you to join the Adonia Choirs. You participated in the singing camps with enthusiasm and excitement and the songs you learnt there accompanied and encouraged you until your last day. Music became very important in your life. Because you lost more and more control of your movements you had to give up playing the flute and the piano and you concentrated the longer the more on singing and listening to music.

You found many good friends in the girls and ladies gymnastic club, and you remained an excited member even after you couldn't follow their exercises.

After school you wanted to become a kindergarten teacher but you had to bury this wish and you joined a school for handicapped young people in Zürich. Travelling there became more and more difficult but never the less you enjoyed the two years there very much. Your brilliant acting in two theatre plays is unforgettable.

The school prepared you to work in an office and you wanted to learn typing. As usual you invested all your efforts until it became too much for you and you collapsed completely. It took five months until you could look for a new professional training. In a home for handicapped people you were trained for two years as a domestic help but after that it was not possible for you to be employed in such a job.

Fortunately another door opened for you. You joined the Beatusheim in the neighbouring village where you found dear and devoted people who cared for you for almost eleven years. As you liked to live at home you travelled to and fro every day. When the weather was

fine we went by tandem and you always helped to pedal as much as you could. You always liked to go to work and the Beatusheim became a second home for you.

The possibilities to work were cut down more and more but you accepted whatever you had to face in a wonderful and admirable way. Breathing and swallowing became difficult and mucus in the lungs made you cough. You never complained about your hard destiny and you never lost your shining and winning eyes.

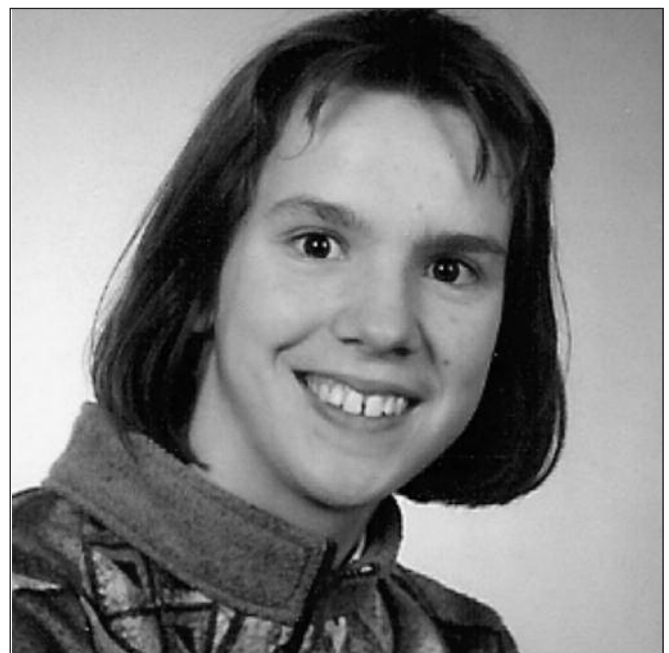
At last you couldn't speak with your lips and you tried to communicate more and more with your eyes but you never lost the ability of singing.

On Friday, 8th May we were called to come to the Beatusheim as you were not feeling well. But when we arrived you had already passed away. Most probably your breathing system was affected so that it couldn't do its work any longer. You had closed your shining eyes for ever. We couldn't tell you how much we loved you and we couldn't ask you to forgive us for what ever we had failed to do for you. You didn't open your wonderful eyes again.

We are very sad but also comforted to know that you left us in the arms of your beloved carers and that you could go quietly and peacefully to your Lord and Saviour. You are now well cared for in his arms.

Dear Annette we are very grateful and we thank God for the wonderful and blessed time we were given together with you.

*Your parents
Kathi and Andi
and your brother Markus*



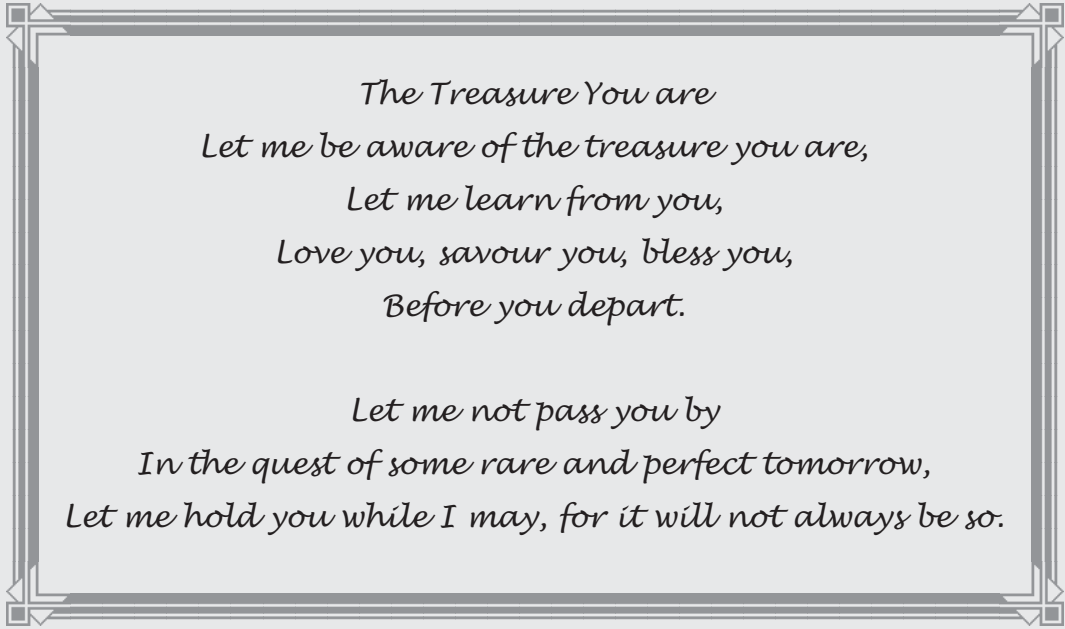


*In
Loving
Memory*

Annette Hediger - age 32



Ellie Sixsmith - age 12

Skye Davison - age 4



*The Treasure You are
Let me be aware of the treasure you are,
Let me learn from you,
Love you, savour you, bless you,
Before you depart.*

*Let me not pass you by
In the quest of some rare and perfect tomorrow,
Let me hold you while I may, for it will not always be so.*



Update from the Central Office

Over the past year, thanks to the awareness raising activities of our Trustees, staff, members and friends, the Central Office has been kept extremely busy – which is of course a good thing to be, but it also brings challenges for a small organisation such as ours. Interest in the work of the Group has increased, bringing more traffic to our website and many requests for information packs. Our relationship with the families and professional members to whom we provide support and information continues to grow, as does the need for our presence at meetings and conferences throughout the world.

The recently formed International Niemann-Pick Disease Alliance will present new opportunities to share information, raise awareness and create networks of support – generating a stronger voice for those affected by the Niemann-Pick Diseases and opening the door to progress.

You may also have read in this newsletter about our successful application to the Big Lottery Reaching Communities Programme – which is a great achievement for us as an organisation. Our focus now is to ensure that our project, the 'Family Care and Interactive Support Service', is delivered with positive impact and has lasting benefits for those we support.

Through our work with the Lysosomal Storage Disorders Patient Organisation Collaboration we will be lobbying for further recognition of rare diseases and their impact on family life, with a focus on maintaining LSD's current designation as a specialised service post 2012.

Our membership of Rare Disease UK the National Alliance created by GIG for people affected by rare diseases and all who support them, enables us to be involved in the campaign to make rare diseases a public health priority. The theme for the 2010 Rare Disease Day was "Patients and Researchers: Partners for life", with events taking place at the Welsh Assembly, the Scottish Parliament and the Northern Ireland Assembly. I attended the event at the Scottish Parliament along with Susan Green, co-founder of the NPDG (UK). Susan was a guest speaker at the event, conveying very deftly the serious nature of Niemann-Pick Disease Type C and her family's on-going experiences with it. Susan also explained the impact that research had had on her family's life and the important role it plays in the lives of all those affected by a rare disease. To find out more about Rare Disease day or Rare Disease UK visit their website www.raredisease.org.uk

We recently held our first meeting for families to get together and discuss NPC Research in Manchester. The day was well attended, and the presentations given were well received. Feedback from attendees has been positive even though the event had to end rather abruptly and there was still much left to cover. Liz Jacklin, NPDG (UK) Clinical



NPDG (UK) Staff, Trustees and Guest Speakers pictured at a meeting to discuss NPC Research Topics at Royal Manchester Children's Hospital in December 2010

research Nurse; will follow up the discussions held on the day with a new research topic that will shortly be available on the 'Hope for Hollie' e-forum. The forum has proved popular and successful so far, and has enabled families to join discussions, ask questions and develop mutual support networks. If you haven't already joined the forum, you can find out how by visiting our website www.niemannpick.org.uk and clicking on the 'Hope for Hollie' logo, which you will find on the homepage. We are always happy to discuss any areas of research, therapies or procedures that you may hear about; so please do contact us at any time with your questions or concerns.

Plans for our Annual Family Conference are well underway - we hope you will be able to join us on our new dates of 10th – 12th September 2010. The Conference Brochure and programme will be available very soon, however if you have any questions or ideas that you would like to share with us, please do get in touch – your feedback helps us greatly in shaping the Conference to meet your needs. The Conference weekend is just as much about networking and making new friends as it is about listening to speakers and attending break out sessions – and that kind of support can make a real difference.

As you can see, the Group continues to work in many areas in the hope of improving lives and outcomes for those affected by this group of diseases. We constantly need your feedback regarding our work to assist us in moving forward and ensuring we are meeting your needs, so please do contact us directly with your comments, we would be very pleased to hear from you.

I hope to see you at Conference in September. In the meantime, if you feel I can help in any way, please contact me at the office on 0191 415 0693 or email niemann-pick@zetnet.co.uk

Toni

Jackie's Journal



Jackie Imrie RCN

Clinical Nurse Specialist
Niemann Pick Disease

Phone/fax: 0161 922 2414

Mobile: 07932 737417

E-mail:
Jacqueline.Imrie@cmft.nhs.uk

Hello again from sunny but cold Manchester. I think we have had only a couple of frost free mornings since the middle of December. At least the snow has finally gone! Talking of snow - I was invited to speak at the Actelion Winter School Meeting in Switzerland recently. When I agreed to do it, I did say that I would have to fly in the afternoon, as I had a home visit in the morning. So 7am saw me driving across the pennines and approaching Bradford with the snow falling - not a lot but enough to worry about getting back to the airport by 12noon! Unlike some of my journeys this winter I managed it with no problem, arriving in Geneva at 6.30 pm. As the taxi set off from the airport to the hotel, the snow started falling, and falling. Once in Lausanne we headed up the steep hill to the hotel. I then had to walk from the hotel to a meeting with some of the team - which was only about 200 yards but guess who did not take boots! I was due to talk at midday the next day but blizzard conditions and about 8 inches of snow meant my talk was brought forward to allow plenty of time for questions before I had to be whisked back to the airport. Thinking about how a little snow stops play in the UK, I was a bit worried but it proved to be no problem at all, and I was safely home later that day. It was a long way to go for such a brief visit, but definitely worth it. Several

people from around the world have contacted me since with more questions about our experiences and how they might be of help their patients.

Back to the UK. We are doing a lot of work with Actelion to raise awareness about NPC. For example, we are producing a small easy to understand leaflet about what to look for, which will be accompanied by a DVD when we have filmed it (snow stopped play in January). One of the Actelion team had this leaflet at a recent neurology conference. One senior neurologist passed by and said he was not interested in stopping as he had no patients with NPC. Our friend from Actelion said "How do you know?" and handed him the leaflet. As he read it he exclaimed and mentioned 2 teenage brothers with vertical gaze palsy and ataxia and increasing neurological problems with the oldest now being in a wheel chair.

The question is "How many patients do we really have in the UK with NPC?" And as people so often ask "How many world wide?"

Over the last few years we have had an ever increasing number of adults diagnosed, the latest being 24 and 49 years old respectfully. We will soon be testing a gentleman aged 70 who may get an answer to his problems. The German team have found a moderately high percentage of cases under the care of psychiatrists. This is why we need to keep raising awareness. Well done to all the families that are helping to do this by telling their stories, raising funds and just talking about this disease. We can all help.

As always do not hesitate to call any time.

Jackie

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 1st September 2010

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.