

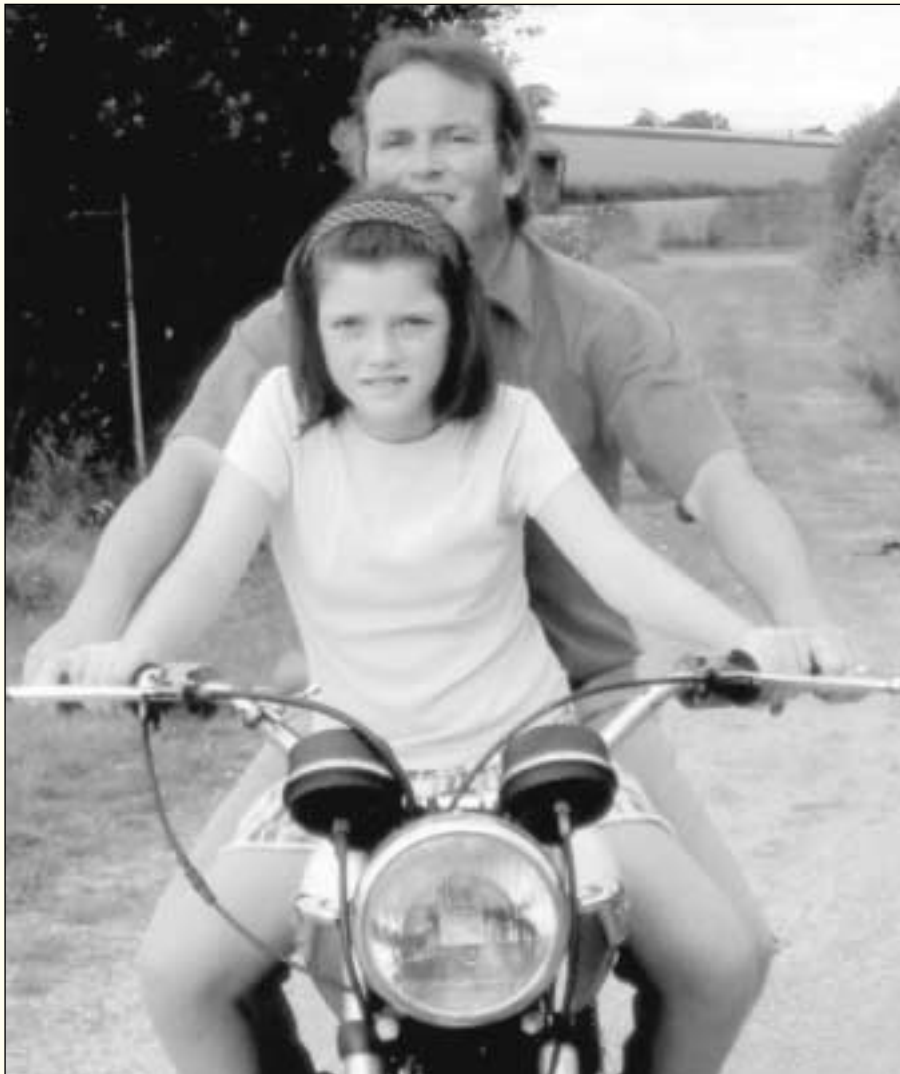


Niemann-Pick News

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

Spring 2007

Vol 14 No 1



Annie and Norman Pyne

CONTENTS

Chairman's Chat	2	Benefits News	17
Research News	3	Family Focus	19
Research Report	8	National Development Manager Report	23
Family Conference 2006	10	Jackie's Journal	24
Fundraising News	13		

Registered Charity
Number 1061881

NP Clinical Support Nurse 0161 922 2414
NPDG(UK) 24 Hour contact number 0191 415 0693

**Niemann-Pick Disease Group (UK)
National Development Manager**

Toni Mathieson
National Development Manager
11 Greenwood Close
The Pastures
Fatfield
Washington
NE38 8LR
Tel/Fax: 0191 415 0693
Email: niemann-pick@zetnet.co.uk
www.niemannpick.org.uk

Chairman:

Jim Green
Tel: 01592 713409
Email jimgee@zetnet.co.uk

Secretary:

Bill Owen
Tel: 01225 865 273
Bill_owen43hotmail.com

Treasurer:

Richard Brooks
Tel: 01249 811311
Email r.m.brooks@btinternet.com

Management Committee

Janice Brooks
Tom Brooks
Richard Rogerson
Dave Roberts
Roger Franklin
Caroline MacDonald
Ian Sixsmith

Patrons

The Rt. Hon. The Earl Cairns CVO
CBE
The Rt. Rev. Dominic Walker OGS
Bishop of Monmouth
Dora Bryan OBE
Sir Robin Calford KCVO CBE
Rt. Hon. The Lord Bassam of
Brighton
Prof. Martin Rossor MA MD FRCP
Guy Johnston
Nicholas Mathias ARAM

Medical Adviser

J.E. Wraith MB ChB FRCP
Willink Biochemical Genetics Unit
Royal Manchester Children's Hospital
Pendlebury
Manchester M27 4HA

Clinical Nurse Specialist

Jackie Imrie RCN
Willink Biochemical Genetics Unit
Royal Manchester Children's Hospital
Pendlebury, Manchester M27 4HA
Tel/Fax 0161 922 2414
Email Jackie.imrie@cmmc.nhs.uk

CHAIRMAN'S CHAT

The aim of our group is to try to "Make a Difference."

Our work this year has focused on refining that aim by consolidating our activities and evaluating what we are doing to see if we can "Make a Difference" faster or, "Make a Greater Difference". We have asked you what you think by way of a questionnaire, we are looking at our performance over a range of different aspects and



above all we are trying hard to maximise our use of resources. We are collectively looking to the future and hence it seems only natural that our conference this year (June 23rd & 24th) has the theme "Focus on the Future". Why is it that we can now begin to do something which, for many of those affected by these diseases, has not so far been possible? Because, relatively speaking there is so much happening now that wasn't happening before which provides us with the light of hope. No false dawns just a gradual increase in the light.

At this year's conference you will hear of the first trial of enzyme replacement therapy for NP type B. This is the first time, for NP type B, that something with the potential to treat the disease has been trialled. News too of the trials and investigations into drugs that will hopefully make a difference to those with NP type C.

As many of you know on the patient support side Jackie Imrie, our clinical nurse specialist and Liz Jacklin our clinical research nurse have been providing huge support to families. What some don't realise is that both Jackie's and Liz's posts, although operated by the NHS are funded by the Group through the fund raising support received from families and grant giving bodies. We are looking to continue to further develop this unique service, thanks to the work of many different people.

Making a difference has never been easy but it can and does happen when people work together.

I hope you enjoy the newsletter.

Jim Green

RESEARCH NEWS

Research Nurse Update



Where has the past 12 months gone! I have been in post for a year now and am still enjoying it!

As some of you may be aware I have now taken over from Helena Prady on the OGT918 (Zavesca) study. Helena has moved on to pastures new and I personally was very sad to see her go - as I am sure many of you were too. However, she is only at the other side of the hospital so we still see her! I know she wants to stay in touch with the group so will no doubt be at our Conference in June.

The study is nearing completion, with some patients having been on the drug for 5 years. We are still waiting for a decision from the drug company about what will happen at the end of the study. We are hoping to have some answers very soon.

The Natural History Paper of Patients with NPC has been published by the Journal of Inherited Metabolic Diseases and has sparked some international interest which we are very pleased about. The aim now is to keep this information updated. I am currently working on a project looking at the affects of Pregnancy and Puberty in patients with NPC. This

entails a questionnaire to all those patients with NPC who have gone through puberty and also those that have had children. We have nearly finished interviewing the patients and their families and are putting the information together. It is hoped that this study will answer some of the questions regarding the effect of puberty and pregnancy on patients with NPC. Our next study, which I have just begun to work on, is looking at the psychiatric manifestations in NPC. We are collaborating with a team in Germany on this work

At the same time I am still learning more and more about the disease and getting out and about with Jackie Imrie meeting patients and their families which I really enjoy. I am also busy at the moment planning the Children's Programme for our Annual Family Conference. We have lots of fun things planned for this year, so I very much hope to see you there.

Liz

Elizabeth Jacklin RGN
Niemann-Pick Disease Research Nurse
Telephone: 0161 922 2728
Email: elizabeth.jacklin@cmmc.nhs.uk

First year combined results of the juvenile/adult and paediatric Clinical Trials of miglustat (registered name Zavesca, previously known as OGT 918).

MIGLUSTAT IN NIEMANN-PICK TYPE C (NPC) DISEASE: A 1-YEAR INTERIM ANALYSIS

M.C. Patterson¹, D. Vecchio¹, H. Prady², L. Abel³, N. Ait Alissa⁴ and J.E. Wraith²

1. Departments of Neurology and Pediatrics, Columbia University, NY, USA; 2. Royal Manchester Children's Hospital, Biochemical Genetic Unit, Manchester, UK; 3. Department of Optometry and Vision Sciences, University of Melbourne, Australia; 4. Actelion Pharmaceuticals Ltd, Allschwil, Switzerland

INTRODUCTION

- Niemann-Pick type C (NPC) disease is a fatal neurodegenerative disorder linked to dysfunction in lipid trafficking^{1,2}
- No treatment exists for this disease aside from palliative care
- Clinical features include pulmonary fibrosis and liver disease in infancy, and progressive neurodegeneration in later onset cases
- The disease is characterized by accumulation of unesterified cholesterol and glycosphingolipids (GSLs) in the lysosomal compartments of many tissues³
- The abnormal storage of GSLs is associated with axonal dendrodegeneration and megacystic formation, characteristic features of NPC⁴
- Miglustat (Zavesca[®]) is a small molecule inhibitor which can reversibly inhibit glucosylceramide synthase, the enzyme that catalyses the first committed step of GSL synthesis⁵
- In physics chemical properties allow miglustat to cross the blood-brain barrier⁶. As such, it may have potential as a therapeutic agent for NPC disease, including the neurological symptoms
- This study is the first to test a potential medication for NPC disease in juveniles and adults, as well as in children

OBJECTIVES

- This study aims to evaluate the safety and efficacy of miglustat as a treatment for NPC disease.

PATIENTS & METHODS

- The main study included juvenile/adult patients randomized to receive either miglustat or standard care. In the paediatric study, all patients received miglustat
- The study cohort comprised male or female patients with NPC (confirmed by abnormal cholesterol esterification and abnormal lipin staining), with normal renal function, who were able to ingest a capsule, and who did not suffer from clinically significant diabetes
- Patients <4 years, and those with medical conditions or who were on concomitant medications that would render them unsuitable for the study were excluded
- Patients were assessed for the primary endpoint, horizontal saccadic eye movement (HSEM) during the screening period and at Month 12. On each occasion, assessments of eye movement velocity were performed twice within 24 hours
- Swallowing ability was assessed at screening, Month 6 and 12
- Neurological examinations and quality of life assessments (for juveniles/adults) were performed at screening, Months 3, 6, 9, and 12, and at follow-up
- Safety assessments were performed at screening, every 3 months, and at follow-up. Adverse events (AEs) were recorded at each visit/monitoring visit
- Measurements presented in the figure were taken at baseline and at Month 12 (last visit)
- The two treatment groups (miglustat vs. standard care) were compared using an analysis of covariance (ANCOVA) model, a non-parametric statistical Wilcoxon test was used to compare the treatment groups with respect to patients' swallowing ability. Data from the paediatric group when compared to those from the juvenile/adult group (descriptive statistics were used for the analysis)

Juvenile/adult Study

- This was a randomized, controlled phase III study in 29 juvenile and adults (12 years and) with NPC
- Patients were randomized 2:1 to 200 mg miglustat three times daily (i.i.d.) or standard care for 12 months
- Patients were given the option to enter or continue active treatment for an additional year after the initial 12 month trial period

Paediatric Study

- This was a non-randomized phase I/II study
- The study involved 12 children (<12 years old) with NPC
- Miglustat dose was adjusted according to body surface area

Criteria for evaluation

- Efficacy:** HSEM measurements, swallowing assessments, neurological tests (including neurological examination and neuropsychological tests), and quality of life assessment. Juveniles/adults also underwent motor and organ volume assessments
- Safety:** AEs, laboratory analysis, vital signs, concomitant medications, and physical examination

RESULTS

PATIENTS

- The demographics and characteristics of the juvenile/adult and paediatric patients at baseline are summarized in Table 1
- At baseline, all patients suffered from various neurological manifestations, including vertical supranuclear gaze palsy, cognitive impairment, ataxia, dyspraxia, dysarthria, and swallowing difficulties. The proportion of patients from each treatment group manifesting these neurological symptoms at baseline is summarized in Table 2

Table 1. Summary of patient demographics and baseline characteristics

Characteristics	Juvenile/Adult		Paediatric
	Miglustat (n=20)	Standard care (n=9)	Miglustat (n=12)
Gender			
Male	10	5	7
Female	10	4	5
Age (years)			
Mean (SD)	24.4 (8.8)	22.0 (7.0)	12.3 (4.4)
Range	14-40	12-32	6-17
6-17 years	0	0	5
18-17 years	0	0	0
18-17 years	20	0	7
Weight (kg)			
Mean (SD)	70.3 (22.0)	64.7 (17.0)	21.7 (14.0)
Height (cm)			
Mean (SD)	173.7 (16.0)	165.7 (13.4)	124.4 (13.0)

Table 2. Neurological manifestations at baseline in juvenile/adult and paediatric patients

Manifestation of NPC	Juvenile/Adult		Paediatric
	Miglustat (n=20)	Standard care (n=9)	Miglustat (n=12)
Vertical supranuclear gaze palsy	10 (50%)	7 (78%)	12 (100%)
Cognitive impairment	19 (95%)	7 (78%)	8 (67%)
Ataxia	20 (100%)	8 (89%)	8 (67%)
Dyspraxia	16 (80%)	6 (67%)	7 (58%)
Dysarthria/dysphagia	12 (60%)	6 (67%)	6 (50%)

EFFICACY

- Horizontal saccadic eye movement (HSEM)**
 - Primary analysis of miglustat treatment vs. standard care shows an improvement in HSEM in both in juvenile/adult and paediatric patients (Fig. 1)
 - Inclusion of patients who were on benzodiazepines (known to affect HSEM velocity) showed statistically significant improvements between miglustat treatment vs. standard care (Fig. 1)

- In paediatric patients, an improvement in HSEM was seen after 12 months of miglustat treatment (Fig. 1). This result was comparable to that of the juvenile/adult patients

Swallowing and auditory acuity

- Miglustat was found to improve measures of swallowing in juvenile/adults in nearly all cases, with no overall deterioration (Fig. 2)
- Miglustat had no effect on swallowing ability in paediatric patients. However, over 80% of paediatric patients treated with standard care had swallowing difficulties at baseline, in contrast to the juvenile/adult population, which showed impaired swallowing at baseline
- An overall trend of improved auditory acuity was seen in juvenile/adults treated with miglustat, while the standard care group showed overall worsening (Fig. 2)

Figure 1. Improvements in HSEM in juvenile/adult (J) and paediatric (P) patients

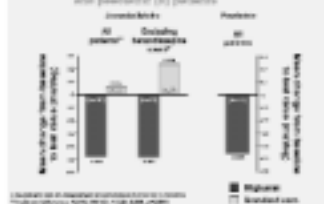
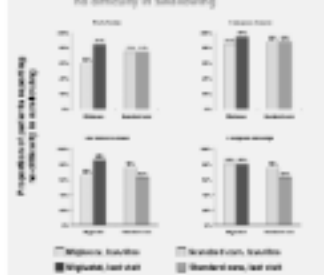


Figure 2. Proportion of juvenile/adult patients reporting no difficulty in swallowing



Effects of miglustat on other secondary endpoints

- Miglustat treatment resulted in improvements in Mini-Mental Status Exam (MMSE) scores in juvenile/adults (Table 3)
- Slower deterioration of Standard Auditory Index scores with miglustat treatment (<0.2) vs. standard care (<0.7) in juvenile/adults was observed. No changes were observed in the paediatric group
- Neurological examination and neuropsychological testing did not reveal any notable changes from baseline or shifts from normal to abnormal
- Improvement of several quality of life domains were observed with miglustat treatment, as compared to standard care, including quality of life, general health, social functioning, mental health and physical component parameters

Figure 3. Proportion of juvenile/adult patients reporting normal auditory acuity

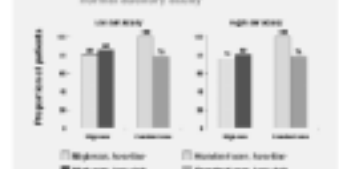


Table 3. Treatment with miglustat improves Mini-Mental State Examination (MMSE) score in juvenile/adults

	Miglustat (n=20)	Standard care (n=9)	Change (SD)
Baseline	20	9	19.5 (5.0)
Month 12	20	9	20.1 (5.1)

Significant differences between treatment groups were observed at 12 months (p=0.0001).

Adverse Events (AEs)

- In adults, the most frequently occurring individual AEs were diarrhoea (95%), flatulence (70%), and weight loss (55%). A summary of adverse events is given in Table 4
- The most frequently occurring AEs were diarrhoea (97%) and flatulence (83%) in paediatric patients
- Discontinuation due to AEs were reported in 3 patients: in 1 paediatric patient due to memory impairment and in 2 adult patients due to constipation (1 patient) and diarrhoea (1 patient)
- No deaths were reported

Table 4. Summary of adverse events

Adverse Event	Juvenile/Adult		Paediatric
	Miglustat (n=20)	Standard care (n=9)	Miglustat (n=12)
Diarrhoea	19 (95%)	7 (78%)	12 (100%)
Flatulence	14 (70%)	6 (67%)	10 (83%)
Weight loss	11 (55%)	5 (56%)	11 (92%)
Constipation	2 (10%)	1 (11%)	1 (8%)
Headache	2 (10%)	1 (11%)	2 (17%)
Stomach pain	2 (10%)	1 (11%)	2 (17%)

DISCUSSION & CONCLUSIONS

- Treatment with miglustat resulted in improvements in the primary endpoint, HSEM, in miglustat-treated patients compared to standard care
- Miglustat treatment showed trends for improved swallowing capacity and auditory acuity, compared to standard care in adult/juvenile patients
- Slower deterioration in Standard Auditory Index was observed in miglustat-treated juvenile/adult patients (untreated patients showed a greater worsening)
- Miglustat had a positive effect on cognitive function in juvenile/adult patients (assessed by MMSE score)
- No unexpected miglustat toxicity was found
- Results indicate that miglustat favourably influences function in some neuronal populations based on improvements in several clinically relevant parameters
- Miglustat is a potential treatment option for patients with NPC, a disease with unmet medical needs

REFERENCES

- Green ED et al. Science 1977;197:683-685
- Levy G, Leclercq G, Mouton RP, et al. JAMA 1982;247:1211-1215
- Wraith JE, et al. JAMA 1982;247:1211-1215
- Wraith JE, et al. JAMA 1982;247:1211-1215
- Wraith JE, et al. JAMA 1982;247:1211-1215
- Wraith JE, et al. JAMA 1982;247:1211-1215

As you will see from the poster the first year results of the juvenile/adult and paediatric Clinical Trials of miglustat are encouraging. Following is a brief summary of the results and what they will mean for Niemann-Pick Type C patients.

What do the results mean?

In summary the results showed that patients who were treated with miglustat showed less deterioration in their condition when compared to a similar group who did not take the drug. Each of the areas measured – eye movements, swallowing, hearing, mobility and cognitive

function – showed either slower deterioration or a trend towards improvement with no unexpected side effects. The main side effects experienced were diarrhoea, flatulence and weight loss.

What benefits will they bring?

The results indicate that miglustat has a number of positive effects on some patients with Niemann-Pick type C and that it could potentially be used as a treatment option in such patients.

What happens next?

At the moment miglustat does not have a license for

general use in the UK, but work is underway to address this.

Will I/my child be able to take miglustat (Zavesca)?

The decision as to who is prescribed miglustat will very much depend on the license that the drug is given and on the clinical decision of the patient's doctor.

We are all eager for the day when there is an effective treatment for this disease; these results take us one more step towards this goal and we should all take encouragement from them. If you have any questions or would like to discuss these results in more detail, please contact Jackie Imrie on 0161 922 2414 or Liz Jacklin on 0161 922 2967, both will be pleased to help you.

Ara Parseghian Medical Research Foundation

NP-C Natural History Study Update

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

Date: March 9, 2007

The NP-C Natural History Study at the National Institutes of Health (NIH) was launched in July 2006. According to Dr. Forbes "Denny" Porter, who heads the Study, the enrolment rate has been much higher than anticipated. This is good news for the NP-C cause.

In total 15 patients have enrolled in the study as of March 15, 2007. They represent 6 females and 9 males, ages 3 to 32. We are very grateful to these families and patients for committing to this very important Study. At this point, the families will be visiting the NIH in Bethesda, Maryland every six months with visits lasting 3-4 days.

Two patients in the study have already returned for their second visit. The comparison of the same tests over this six-month period will give a great deal of clinical information. Although some interesting indications have been observed that could be very meaningful if they continue with the second visit of all the patients, it's too early to make definitive statements about certain outcomes.

The families who have taken part have been very complimentary of Dr. Porter, of the Study nurse Nicole Yanjanin, as well as the many specialists who are conducting the numerous tests. Families are realizing that although it is a task to make time in their schedules, leave other family members at home, and travel to Bethesda...knowing that their child is being seen each six months by clinicians who are seeing 15-20 other NP-C patients is reassuring. These special NP-C families also realize that this is one more way they can contribute in the battle against Niemann-Pick Type C disease.

The following is taken from an email message sent to the Parseghian Foundation from Laurie Friedl, an NP-

C mother who enrolled her daughter in the Study: "We've returned home from our trip to NIH...Dr. Porter and his nurse are wonderful! It was a long week, but we're very glad we went. Ashton actually cried when we were leaving, because she wanted to live at the Children's Inn. It's amazing how quickly kids forget about the bad things and focus on the good!

We wanted to thank you again for informing us about the study. If there are other parents who are considering becoming involved, we'd be happy to talk to anyone or help in any way that we can." Laurie Friedl may be contacted at: lfriedl@excite.com

NP-C families/patients from outside of the United States are welcome to enrol in the study. The NIH would cover the travel expenses from a port-of-entry city in the US to Washington DC, as well as make arrangements for ground transportation to and from the airport to the NIH, and for the family to stay in the Children's Inn on the NIH campus or another close-by facility free of charge. The family (usually one parent and the patient) would be required to pay for their own round-trip flights from their homes to New York or Washington DC. There may be potential funding to cover the costs of these flights, but at this point this funding has not been finalized.

If any family is interested in enrolling their NP-C child in the Natural History Study, please contact Nicole Yanjanin at: nyanjanin@mail.nih.gov

Any UK families considering taking part in this study are urged to consult with Jackie Imrie at the Willink for a full explanation of the tests involved. Jackie can be contacted on 0161 922 2414 or by email jackie.imrie@cmmc.nhs.uk Further funding may be available for flights to the USA, if you would like more information please speak to Jackie.



Update from Genzyme

Paul L. Kaplan, Ph.D., M.B.A.
 Senior Director of Program Management
 Genzyme Corporation (www.genzyme.com)
 e-mail: paul.kaplan@genzyme.com

The Phase 1 clinical study of recombinant human acid sphingomyelinase (rhASM) for treating ASM Deficiency (Niemann-Pick Disease, Type B) began on January 30, 2007 when the first patient was infused at Mt. Sinai School of Medicine (MSSM). Everything went as planned and the patient tolerated the rhASM infusion well. Additional patients are being screened for the study and scheduled for treatment and assessment visits. While there is still much work to do before enzyme replacement therapy for ASM deficiency becomes a reality, this is nevertheless a tremendous milestone for patients. A big thank you goes to Drs. McGovern, Wasserstein, Schuchman, and Desnick at MSSM and an international team consisting of Dr. Wraith at the Royal Manchester Children's Hospital, Dr. Giugliani in Brazil, Dr. Bembi in Italy, Dr. Mengel in Germany and Dr. Vanier in France for all the hard work that they have done to help prepare for this clinical trial. We are also grateful to the patients who have been anxiously waiting for this day to arrive.

The main purpose of this Phase 1 study is to evaluate the safety of rhASM given as a single dose to adults with ASM deficiency. Based on their medical history, potentially eligible individuals are being contacted by the study staff. The trial itself will take place only at MSSM. However, anyone interested in participating in the clinical trial can learn more about the study at www.clinicaltrials.gov/ct/show/



NCT00410566?order=2.

Participation in the trial will require up to four visits to MSSM. Travel expenses and study related medical treatments are being paid for by Genzyme Corporation, which is sponsoring the study.

During the study, successive groups of patients will receive increasingly higher doses of enzyme. This type of study is called a sequential dose escalation study because the data from each group of patients must be analyzed before deciding whether to proceed to the next higher dose. The decision about whether to proceed to the next dose level will be based upon how well the rhASM is tolerated. It is expected that study enrolment will continue at least through the end of this year, after which it will take several additional months to collect and analyze all of the data. The information learned about dosing in this study will then be used to help design future studies that will evaluate the effectiveness of rhASM in different groups of patients. Such efficacy studies will take several years to complete and must be submitted to regulatory authorities around the world as part of the comprehensive registration package needed for drug approval.

National Niemann-Pick Disease Foundation

At their Board meeting in February the NNPDF were pleased to approve the following grants.

Dr Fran Platt, Department of Pharmacology, University of Oxford. \$99,090 (£49,545)

Evaluation of anti-inflammatory intervention (NSAIDS) as an adjunctive therapy in Niemann-Pick Type C1 disease.

Dr Platt aims to test whether anti-inflammatory drugs may be a beneficial add-on therapy in NPC. She will do this by studying how the NPC mouse model responds to anti-inflammatory drugs, either alone or in combination with the substrate lowering drug miglustat (Zavesca).

The Lysosomal Storage Disease Research Consortium

\$10,000 (£5,000)

The LSD Research Consortium (LSDRC) is a collaborative research-funding group, comprised of LSD patient support groups and private family research foundations. The LSDRC has entered into an agreement with the National Institute of Neurological Disorders and Stroke (NINDS) for the purpose of a jointly sponsored program to provide financial, scientific and administrative support towards preclinical or translational research specifically addressing the neurological aspects of lysosomal storage disorders (LSD).

You can read more about the LSDRC on their website <http://www.lsdresearch.org/>

Susan H Green

Update from GOLD, Global Organisation for Lysosomal Diseases

Dr Ann Hale, Executive Director.

Firstly, I'd like to somewhat belatedly wish all friends at the NPDG-UK a very happy New Year – although with the way the time is rushing by, it feels about half way through already!

Membership of GOLD has been growing steadily, and we now have 140 member organisations in 34 different countries. Recently, we have added China to our international network of scientists, clinicians, patient organisations and pharmaceutical companies, working to help support families and find a cure for all lysosomal storage diseases (LSD). The UK Niemann-Pick Disease Group was one of the first members of GOLD back in 2004.

We have been developing new member only areas of the GOLD website, and adding to the video presentations on the site. As the NPDG (UK) is a member organisation, any of its members can access the member areas by registering at the website, and selecting "Niemann Pick Disease Group UK" as your organisation. The latest addition to the video presentations is the Guest Lecture from GOLD's Annual general Meeting, given by Professor Tim Cox, whom many of you will know from NPDG (UK) conferences. Prof Cox's lecture, "Medicine for Lysosomal Diseases: Past Imperfect, Future Tense" reviews the development of therapies for LSD, and discusses the important issues of funding and access to treatment. With treatments in the pipeline for NPB and NPC, this will soon become an issue for NP patients.

GOLD recently collaborated with the Gaucher Association to film their 15th Anniversary Family Conference. This work is being edited and will be on the website soon.

For those interested in reading up to date scientific reviews, Scriver's Online Metabolic and Molecular Basis of Inherited Diseases (OMMBID) is available through the GOLD website. This is the "bible" textbook for metabolic disease. The section on LSD has been available for a few months now. However, recently McGraw Hill Digital Publishing have had a few issues with access and are changing the technology platform. Over the next couple of months, if you try to use this and have any problems, please contact me. By the time the migration is complete, access should be easier. The Niemann Pick A and B chapter is written by Drs Ed Schuchman and Robert Desnick, and the NP-C chapter by Drs Marc Patterson, Marie Vanier and colleagues.

Another area recently added to the GOLD website is the "Member's news" page. There is a picture of



Jackie and Toni receiving the runners up award for NPDG (UK) as Patient Group of the year. Congratulations to the group from GOLD.

I've been busy fundraising for GOLD. Due to an ankle injury, I had to stop going to the gym, so decided on swimming as an alternative exercise. As this is not exactly my preferred form of exercise, I decided to add a challenge and force myself to continue by raising some much needed funds for GOLD at the same time. So, Annie's Channel Challenge was conceived in the local swimming pool! I decided the English Channel was a suitably challenging distance, but as I am not a good swimmer, and completely stupid (or suicidal) the real thing wasn't actually an option! So, I decided on the distance of the English Channel, over a fixed time period. The staff at the local pool logged all my visits and signed a record form for corroboration, and I went swimming at lunchtimes, evenings and weekends until I'd completed the 34Km distance – 1360 lengths of my local pool. During January, I was swimming at least 6 days a week, and wondering if the smell of chlorine would ever wear off! Thankfully, it has now as I finished the challenge on February 2nd.

So, I'd like to take this opportunity to say a big thank you to members of the Niemann Pick Disease Group who supported this effort, and sponsored me.

If you have any questions or would like to make any comments to or about GOLD, please send them via Toni by email niemann-pick@zetnet.co.uk or telephone 0191 415 0693, or direct to Annie at ann.hale@goldinfo.org or via the GOLD website www.goldinfo.org.

RESEARCH REPORT

BY BILL OWEN, RESEARCH CO-ORDINATOR

Disease Prevention – Risk Reduction Opportunities

It has been over seven years since the NPC1 gene was identified and sequenced at Dr Peter Penchev's laboratory in the National Institutes of Health and I remember thinking at that time, that this will give my family and others similarly affected, hope that an effective treatment is just around the corner. A corner, being a sharp transition, is the wrong description. We are on a long steep bend with high walls either side restricting visibility ahead. Every so often we encounter an object in the shape of a new discovery which renews our hope but the end of the road remains hidden from our sight. The discoveries are becoming more frequent and perhaps this is an indicator that the end is approaching, or perhaps, not just yet. We can only wait and see – very frustrating.

In previous Newsletters, mainly Winter 05/06 and Summer 06, I wrote about the possibility of pursuing a disease prevention strategy as a complementary approach to finding a cure. I also discussed some of the problems associated with this approach and how individuals and families could contribute towards defeating the disease, or at least helping to reduce the incidence in future generations. Not necessarily what families with affected children would like to hear as their problem is here and now. But if we can successfully promote disease prevention, then perhaps families in the future, some of whom may be our own descendants, will be spared our ordeal.

An outline of the strategy is given below together with a brief commentary on feasibility, risk and some problem areas. The basis of the strategy is that once disease causing gene mutations show themselves, at great cost to individuals and their families, they should not be allowed to return to anonymity but should be tracked throughout the current generation of relations and through time.

Where does this lead?

The problem arises when a person who is known to be a carrier of a disease causing mutation, wishes to enter into a relationship with a person who has no record of having the disease in their family (a person from the general population) and, they wish to have children. Until recently it would not have been possible to test the status of 'normal' people except

for the common European mutation I1061T. The situation has now moved on in two ways which will allow meaningful testing to be undertaken. Firstly, and because of the misfortune of other families being diagnosed with NPC, the number of disease causing mutation known to date is approximately 250. There are probably many more to come but only time will tell.

Secondly gene chip technology is no longer something for the future, it is commercially available now and allows concurrent testing of a normal persons gene(s) for any of the known mutations. Biotech companies offer genetic testing services via the internet at costs which are within the budget of medium sized charities. An exploratory quotation I received for a 200 mutation chip was approximately £20,000 setting up costs and just over £100 per subsequent test. The chip mutation population is expandable so that newly identified mutations can be added.

From the website I looked at, I see that a number of gene chips for a range of diseases including many eye conditions and cystic fibrosis, are in use. (see www.asperbio.com for further information.)

What are the main problems?

There are many individuals diagnosed with genetic diseases such as Niemann Pick whose mutations have not been identified. There are a number of possible reasons for this and it is an area requiring further research.

Any test cannot be 100% as all the disease causing mutations are not yet known. Testing should therefore, be regarded as a risk reduction measure, remembering that the chances of being a carrier are low to start with.

The next issue is who would set up the organisation and running of the service? In the UK we expect that the NHS should do this but with the growing pressures on resources, it may be necessary to consider other options; eg collaboration with other charity groups in Europe and the USA; applying for grants from organisations who would fund this type of activity.

A further problem is that of obtaining the support of the families. This is not a simple thing to do as there are many issues to be considered, not least having a good understanding of family trees.

Are there any other measures that can be taken?

The gene chip process discussed above may be regarded as stage 1 of a risk reduction programme. There are further options for testing which involve demonstrating that a gene from a normal member of the population is able to produce a functional protein; ie it can be shown to correct a range of cellular defects which arise because the resident protein in a diseased cell, is defective. This can be done at present but the effort in skilled labour and, the need for specialist laboratory facilities means that it is out of reach of standard healthcare services. What is required is a research programme to productionise the process such that it can readily be conducted in standard laboratories.

Is any work being carried out by the health authorities to progress these tests?

Not to my knowledge. The general attitude of the authorities to a disease such as Niemann-Pick is that it is so rare any effort needed would be disproportionate and of no benefit to main line needs. This attitude is wrong in that prevention can not only save considerable distress to affected individuals and their families, but applied to the many thousands of rare diseases, would significantly reduce the burden of social and medical care required, hence saving money in the longer term. Rarity should not be used as an excuse for inactivity. If the prevention approach can save lives of children not yet born, why would we not wish to make it happen?

The natural history of Niemann–Pick disease type C in the UK

J. Imrie · S. Dasgupta · G. T. N. Besley · C. Harris ·
L. Heptinstall · S. Knight · M. T. Vanier ·
A. H. Fensom · C. Ward · E. Jacklin ·
C. Whitehouse · J. E. Wraith

Following is a summary of an article that recently appeared in the Journal of Inherited Metabolic Disease. If you would like a copy of the complete article please contact the office on 0191 415 0693

Niemann–Pick disease type C (NPC) is an autosomal recessive, neurovisceral lipid storage disorder. Mutations in two genes (NPC1 and NPC2) produce indistinguishable clinical phenotypes by biochemical mechanisms that have not yet been entirely clarified. The wide spectrum of clinical presentations of NPC includes hepatic and pulmonary disease as well as a range of neuropsychiatric disorders. Late-onset disease has been increasingly recognized as the biochemical diagnosis of NPC has been more widely applied in adult neurology clinics. The clinical presentation and follow-up of 94 patients with NPC is described, 58 of whom were still alive at the time this report was prepared. The age at diagnosis ranged from the prenatal period (with hydrops fetalis) up to 51 years. This review of NPC patients in the UK confirms the phenotypic variability of this inherited

lipid storage disorder reported elsewhere. Although a non-neuronopathic variant has been described, most patients in this series who survived childhood inevitably suffered neurological and in some cases neuropsychiatric deterioration. While symptomatic treatment, such as anticholinergic and antiepileptic drugs, can alleviate some aspects of the disease, there is a clear need to develop a specific treatment for this progressively debilitating neurodegenerative disorder.

Conclusion

The results of this study demonstrate that NPC can present at all ages and that early observation of symptoms is not always an indicator of possible disease progression. This is important when discussing prognosis and future reproductive history for families as it may affect decision making. With a more accurate understanding of the natural history of NPC it is possible to produce individualized care plans for patients, mobilizing the full range of available community services. Greater knowledge of the clinical course of the disease will be of vital importance as more treatments are developed in an attempt to treat the disorder.

The Peter Carlton Jones Memorial Award 2007

Submissions for the Peter Carlton Jones Memorial Award 2007 have now been received and are being considered by the Award Committee. The winning submission will be revealed at our 2007 Annual Family Conference in June and will be published in the autumn issue of Niemann-Pick News

CONFERENCE 2007

“Focus on the Future”

NPDG (UK) Annual Family Conference 2007

23 / 24 June 2007

Hilton Hotel, Northampton

Janice Brooks, Conference Chair, writes:

This is my first year as Conference Chair and I have thoroughly enjoyed being involved in the organisational process of Conference 2007. There are many exciting things happening in the field of NPD at the moment and our Conference theme “Focus on the Future” reflects this.

The programme is very busy and exciting, with many interesting speakers; this year will be especially important for us as a family (my grandson James has NP-B) as the clinical trials for Enzyme Replacement Therapy for NP-B have just started in the USA. Dr Margaret McGovern will be joining us from the States to update us on the progress of the trial, so this year's Conference is a must for all Type B families. However, when you look at the enclosed programme, you will see that there is something for everyone, including six Breakout sessions, Family Voice and a 'Questions and Answers' Session, giving you the opportunity to ask our Speakers a question of your choice.

We will also be joined by Dr Lizzie Burns; since 2003, Lizzie has used her unique background in science, art and teaching to develop a wide variety of workshops to inspire children and adults about the biomedical sciences. She has collaborated with scientists to find new ways to communicate their subject and convey their passion for science. Lizzie's work will be displayed throughout the Conference and she will be running workshops for both adults and children.

The Children and Young Adult's Programme is looking as exciting as usual with an overall theme of 'Pirates and Princesses'. Liz Jacklin, our own Clinical Research Nurse, is running the programme this year with help from a team of experienced volunteers. Trips to the bowling alley and the cinema are planned



for the Saturday, plus fairground games, crafts and many other fun activities.

Saturday Evening will begin with a drinks reception, followed by dinner and entertainment. The entertainment will be very special this year, as we are pleased to announce that it will be provided by one of our very own 'Niemann-Pick' families, Carl Henry and Emma Jane, whose son Calum has NP -C. They are much in demand and have re-arranged their busy schedule to join us at Conference. We thank them for that and are very much looking forward to their performance. During the evening there will also be entertainment provided for the children and of course, our annual raffle with many great prizes. Tickets for the raffle are available now, if you think you could help our fundraising efforts by selling a few, please contact the office on 0191 415 0693 or email niemann-pick@zetnet.co.uk. We also plan to run the Tombola again, as it was so very popular last year.

To book your place for Conference 2007 simply complete the enclosed form and return it to the office

// Start by doing what's
necessary, then do
what's possible and
suddenly you are doing
the impossible

St Francis of Assisi //

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

We do hope you will be able to join us for this exciting weekend, as well as hearing the latest important information, this weekend is all about sharing information and strengthening our family support network, so there will be plenty of opportunity to

meet other families, make new friends and speak directly to the professionals involved in this field.

Finally, whether you are a regular attendee or joining us at Conference for the first time, we want you to have a positive experience. When planning this year's event, we have taken into account the feedback received in previous years and hope Conference 2007 will meet the needs and expectations of you all.

See you in June!

Janice
Janice Brooks
Conference Chair



Comments from Conference 2006:

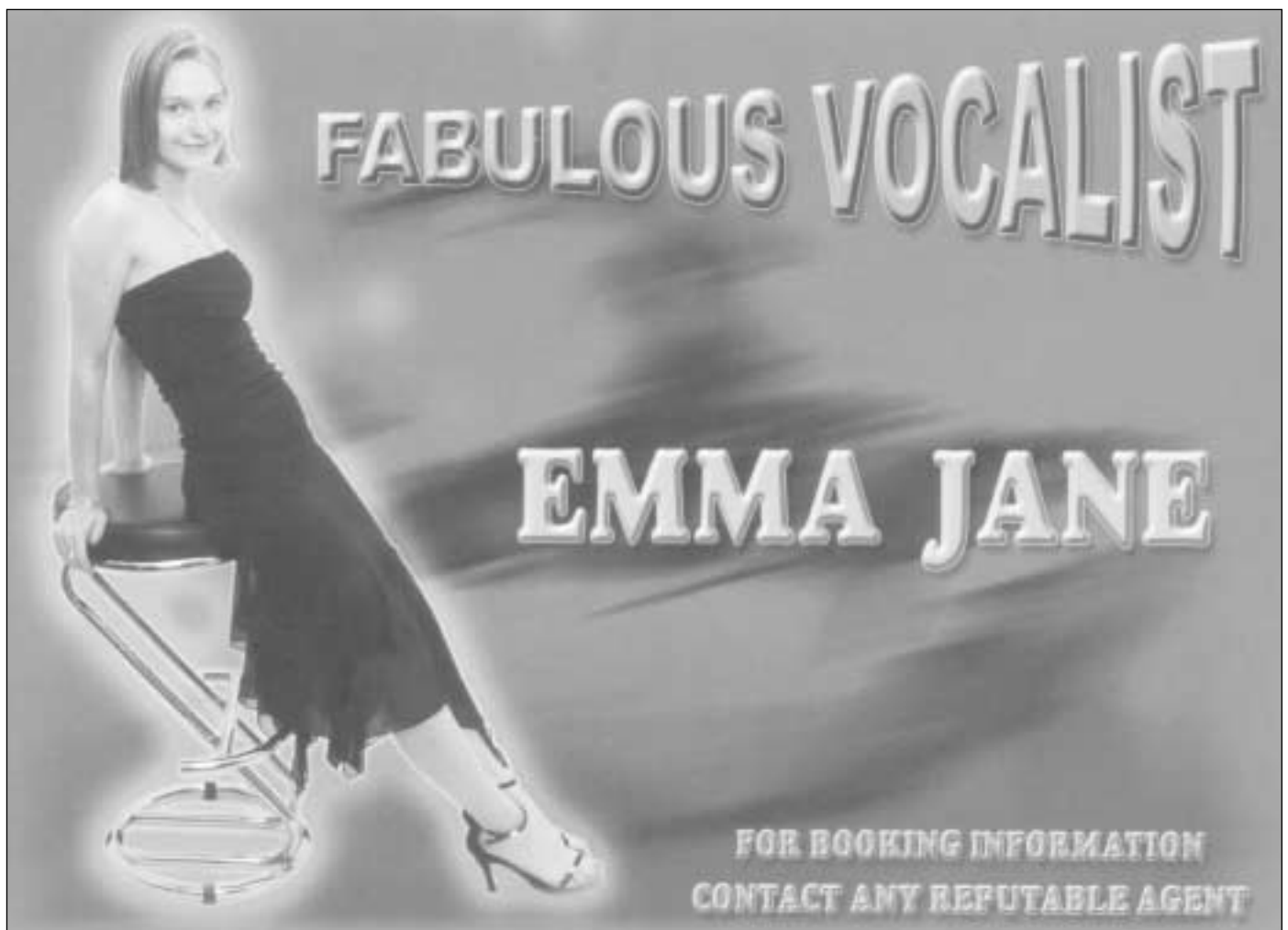
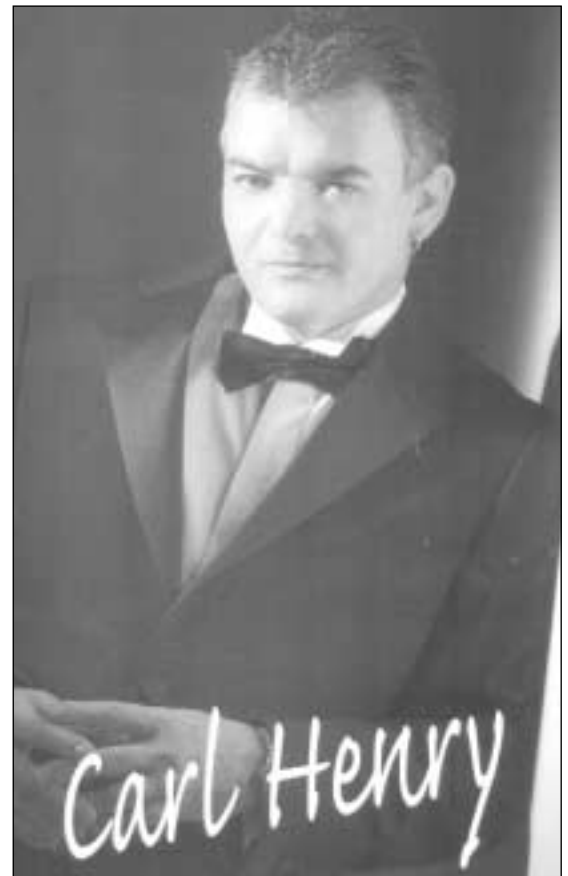
'Excellent. Very informative – great variety'

'As a family we really look forward to our weekend away'

'The venue was very good – easily accessible with spacious accommodation'

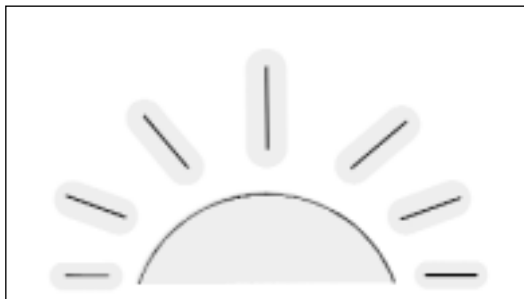
'It was very useful to speak to other families in our situation'

'The Children's Programme was excellent and kept them entertained'



FUNDRAISING NEWS

Awareness Week 2007 – July 16th – 22nd 2007



Our 4th Annual Awareness Week is fast approaching! We hope that those of you who joined in the fun in previous years will once again help to raise awareness of Niemann-Pick diseases and raise funds to support the work of the Group. In 2006 we decided to move Awareness Week from October to July to take advantage of the better weather and allow more outdoor events to take place. This proved to be a popular decision, with many participants organising their own event, such as a 'Niemann-Pick-Nic', balloon race or garden party, throughout the country. We would like to say a big thank you to all those who took part, your hard work is much appreciated!

This year, we once again invite you to organise a 'Niemann-Pick-Nic' of your own. Perhaps you could involve your local school or community group. Our aim is to have many picnics all over the country, attracting the attention of the media. After all it is all about awareness!

Of course you can organise an event of your choosing, not just a 'Niemann-Pick-Nic'. If you would like some ideas, or a fundraising pack please get in touch with us at the office on 0191 415 0693 or by email niemann-pick@zetnet.co.uk

Remember, any event, whether it is large or small is welcome. We can provide you with advice on how to organise an event and supply you with a fundraising pack, containing; a t-shirt, stickers, a poster, an ideas sheet, collection boxes, information leaflets and a sponsor form. We also have other items you can use to help raise funds, such as wristbands and balloons.

I hope you will join us once again as we further our aim of raising awareness of Niemann-Pick diseases – we need your help to make Awareness Week 2007 a success!

Please contact us at the office if you would like more information or a fundraising pack. Telephone 0191 415 0693 or e-mail us at: niemann-pick@zetnet.co.uk

Awareness Opportunity

Every week Real People magazine runs a short piece on a child with a rare condition or disorder. Although it's a populist magazine the information given on the rare disorder is usually quite straightforward. The piece involves a brief interview with the child's parent, and a paragraph from a medical specialist. Parents would also need to supply photographs which are returned after publication. With a circulation of around 350,000 people, this could be a real opportunity to raise awareness of your child's condition, and help other parents.

If you are interested, please contact Clare Swatman, Health Editor at Real People. Claire only works on Thursdays her telephone number is 020 7339 4591. Alternatively, it would be best to email her at <mailto:clare.swatman@acp-natmag.co.uk>.

Prize Draw Winner

Congratulations to Jackie Humphries, the lucky winner of our Questionnaire Prize Draw, who wins £50.00 of Marks and Spencer vouchers. Thank you to everyone to returned their Questionnaire, your responses will help us to improve the services we offer and hopefully make a bigger difference to families living with Niemann-Pick diseases.

Swiss Ironman 2007

On 24th June 2007, Ian Birchnall will be taking on his biggest challenge to date in aid of Niemann-Pick. He will be endeavouring to complete arguably the hardest endurance event possible, by swimming 2.4 miles, cycling 112 miles and running 26.2 miles in the Swiss Ironman event. Since 1999 Ian has taken on challenges each year to raise funds for NP, but this year will see him really put himself to the test. Since discovering that his Niece has NP, it is a charity that is close to his heart and each year the challenge has got harder to ensure sponsorship is maximised.

Training started in November, and is currently on target to achieve the goal of completing the event in 16 hours (1.5 hours for the swim, 8 hours for the cycle, 6 hours for the run and 0.5 hours for transitions). Training is currently taking about 8 hours per week, but when the lighter nights arrive this will double.

Anyone who would like to contribute to the fundraising total can do so by either emailing (ibirchnall@lear.com) or telephoning (01530 273339) a pledge. All donations received will be given to NP, as costs of the event are being paid for by Ian himself.

The results of the event will be featured in our Autumn Newsletter, so keep a look out for it.

Ian Birchnall



Land's End to John O'Groats... In 24 hours on a classic 1970's Triumph Trophy Motorcycle



Why? You might ask... Well, the reason is Annie.

In July 2005, Norman and Kait Pyne's daughter Annie, who was then 9 years old, was diagnosed with Niemann-Pick Type-C. At that time and in the months since, the family have received help and support from the NPDG (UK). To give something back and to help boost NPDG (UK)'s funds, Norman proposed a sponsored ride on a classic fundraising route from the bottom to the top of the UK mainland on a classic British bike

Norman says "I've often thought about doing the end to end ride on my Classic Triumph Trophy and have decided to do this at Easter 2007 to raise money for the Group. I'll try to make the ride in

just 24hrs. This shouldn't be too difficult, plenty of people have done this trip before but it will require good preparation, care and stamina. (Also some fairly good map-reading because I don't think 'satnav' would be in the spirit of the ride!). The distance of the trip will be about 860 miles."

Norman has created a website dedicated to the trip, visit www.np-ride.org.uk for more information.

**THE
ARONOWITZ
ENSEMBLE**

Chamber music at its finest



FEATURING
Guy Johnston, Jennifer Stumm, Magnus Johnston, Tom Foster, Marie Macleod,
Tom Hankey and Nadia Wijzenbeek

Playing Mozart, Mahler and Schoenberg

**in a concert performance at
St George's Church, Kemp Town, Brighton
on Friday 22nd June 2007 at 7p.m.**

**Tickets at £15 and 12.50p (concessions) available from
THE DOME BOX OFFICE, NEW ROAD,
BRIGHTON. Tel: 01273 709709**

PROCEEDS IN AID OF THE NIEMANN-PICK DISEASE GROUP REGISTERED CHARITY NO. 106081

The Aronowitz Ensemble Concert

Once again, Tony Jellings, former Board member and Fundraising Co-ordinator for the NPDG (UK), has endeavoured to organise a prestigious event to benefit the Group. A Charity Concert will be held on Friday 22nd of June at St George's Church, Brighton. As you will see from the flyer, the concert will feature the immensely talented Aronowitz Ensemble, who last year gave a stunning performance to a large and appreciative audience at St John's Church, Wimbledon, raising a substantial amount for the NPDG (UK). Among the musicians will be our Patron, award winning cellist Guy Johnston and his violinist brother Magnus.

If you would like to attend the event, please contact The Dome Box Office, Brighton, on 01273 709709.

We would like to take this opportunity to thank Tony and the Aronowitz Ensemble for their continuing support and send our best wishes for a very successful event.

Bibby Distribution

Elliott Mark Lister – 17th September 1994 – 15th September 2003

Bibby Distribution is celebrating its 200 year Bi Centenary this year with a full year of charity events. They have very kindly chosen the NPDG (UK) as one of the charities they would like to support. Every pound raised at their events will be matched by the company. The NPDG (UK) was chosen as a beneficiary after being put forward by Mark Lister, who lost his son Elliott to Niemann-Pick Type C in 2003. Mark is a member of the Group and is employed at Bibby's

Leeds depot. They have already held several events and are holding a charity ball in April. Other events planned are 'It's a Knockout' and a five-a-side football tournament. The company have a website dedicated to their centenary celebrations, visit www.bibby2007.com for more information. We would like to take this opportunity to thank Mark and Bibby Distribution for choosing the NPDG (UK) and wish them lots of luck in their fundraising efforts.

Generous Gestures

Happy Birthday Lindsey!

A big thank you to Lindsey Foster for generously requesting donations to the NPDG (UK) instead of birthday presents. Lyndsey held a Masquerade Ball for all her friends and family, who thoroughly enjoyed dancing the night away. The guests were extremely generous, raising the grand sum of £400!



2007 Christmas Card Competition

Firstly, thank you to everyone who so generously supported the Group by purchasing our 2006 Christmas cards. The cards proved to be extremely popular this year, thanks to the wonderful designs by our three budding artists, Annie Pyne, Edward Hurst and William Brooks. The total amount raised was a staggering £2,176.88!

We should like to invite entries for the 2007 Christmas

Card Competition, which will once again be judged by attendees at our Annual Family Conference in June this year. Please send your entries to the central office: NPDG (UK) 11 Greenwood Close, Fatfield, Washington Tyne and Wear, NE38 8LR, to arrive no later than June 11th 2007. Don't forget to put your name, address and age on the reverse. Glitter and glue at the ready!



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

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

William Brooks, aged 6 – 'Angel'



Thank you!

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

BENEFITS NEWS

New help from Contact a Family and Citizens Advice if you are in financial difficulties.

In 2005 Contact a Family and the Family Fund carried out some research which found that many families with disabled children were in financial difficulties and struggling with increasing debts. Many families told us that they weren't able to access help from traditional advice centres because of the long queues.

Contact a Family has joined up with Citizens Advice to secure some funding for a specialist money advice project especially for families with disabled children. The new service offers face to face debt advice and help by home visit in the following areas of England:

- Across the whole of Greater London
- Across the whole of Great Manchester
- Liverpool
- Stoke on Trent

- Walsall
- Wolverhampton
- Sheffield
- Rotherham
- Bradford
- Gateshead

If you would like help get your finances back on track call the Contact a Family freephone helpline on 0808 808 3555.

Adapted from an article which appeared in the Contact a Family Rare Disorders E Newsletter 23rd March 2007.

New Right for Carers to request Flexible Working

Current figures suggest that 52% of carers are **juggling work with their caring responsibilities.**

The government have extended the Work and Families Act 2006 to cover carers of adults and this comes into force in April.

Under this Act carers will now have the right to ask for flexible working arrangements in the same way that parents of young children or disabled children up to the age of eighteen have the right to do at present. Flexible working might be changing your hours or working from home and applies to employees who have worked for their employer for at least 26 weeks.

There is also the right to take a "reasonable amount of time off work to deal with an emergency involving a dependent". It is at the employer's discretion whether or not the leave is paid or unpaid.

Employers will still have the right to say no to your request for flexible working arrangements but must demonstrate clearly why they are refusing. It is important to bear in mind that if an employer accepts a request, then this results in a permanent change to the employee's contract of employment.

If you are finding it difficult juggling your paid work with your responsibilities as a carer it might be helpful to think about what would make life easier at work. It is sometimes the simple things like access to a telephone or getting hold of information that can make a difference.

There are a couple of web sites where you can get further information, these are

www.direct.gov.uk/Employment/Employees/fs/en

or

www.businesslink.gov.uk

or you could telephone the free confidential helpline run by Acas for employers and employees on all aspects of employment law: 08457 474747 (Mon-Fri 8am-6pm).

Carers UK have a booklet for carers on this subject, you can contact Carers UK on 0808 808 7777 or by email info@carersuk.org

Adapted from an article that appeared in the Spring 2007 Fife Carers Centre Newsletter.

Useful Publications

Different Dads

Fathers' Stories of Parenting Disabled Children

Edited by Jill Harrison, Matthew Henderson and Rob Leonard

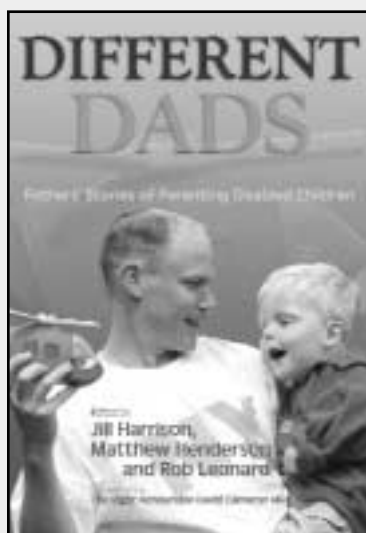
Foreword by

The Right Honourable David Cameron MP

2007 176 pages Paperback ISBN 978 1 84310 454 4 £12.99/US\$19.95

'It is a great idea to draw together stories of fathers' experiences in bringing up disabled children. One of the ways parents manage to cope is to know that others have been through the same experiences. Then you learn that it isn't just about coping – there are positive stories to tell.'

– *David Cameron MP*



Fathers of disabled children can feel overlooked when the focus of much parenting support is aimed at mothers. *Different Dads* is a collection of inspiring personal testimonies written by fathers of children with a disability who reflect on their own experiences and offer advice to other fathers and families on the challenges of raising a child with a disability.

The fathers featured represent a broad spectrum of experience. Their contributions reflect a wide range of cultures; some are single fathers, others are married adoptive fathers. What they all have in common are the challenges that face them and their families in raising a child with a disability. Issues explored include the reactions of family, friends and colleagues, how to deal with the organisations and professionals that support families with a disabled child, and the difficulty of being open about feelings in a culture that doesn't always expect men to have a sensitive or nurturing role.

Offering direct and thoughtful perspectives on being a father of a child with a disability, this book will be a valuable source of support and information for families with disabled children, and also for health and social care professionals who work with these families.

If you would like a copy of this book please contact
Jessica Kingsley Publishers on 0207 833 2307 or email www.jkp.com

FAMILY FOCUS

One Step Closer

We are the Brooks family from Hemel Hempstead in Hertfordshire. James was diagnosed with NPB in May 2000. We had welcomed William into the world the previous January, and things were on the up but the year ended being full of positive and negative emotion for us.

Initially we dealt with just finding out the implications of how this might affect our little boy and in doing so learned that, although nobody had been cured of this, there was an enzyme replacement treatment already developed.

Our expectations were that, within a year or so there, would be an ERT treatment available to him but a few legal issues had to be dealt with over the next few months.

Testing and licensing are things we know little or nothing about and it has been frustrating to wait as these months turned into years. James is now 9 years old and in this time has had no treatment, but he also hasn't let NPB affect him in any way. He visits Great Ormond St and Manchester regularly but this is purely so that his development and progress can be measured.

We have always been fortunate to know that, for James with NPB, an ERT treatment is just a question of time.

Two years ago we learned that there seemed to be a problem with the drug. This had been developed by



Genzyme in the USA working in cooperation with Mount Sinai Hospital in New York. The problem stemmed from the Federal Drug Administration's strict stipulations on what was expected of the drug. This led to a further delay of 18 months or so.

However, the news of the FDA's approval and consequent start of a first stage clinical trial is everything we could hope for. It gives us further hope that James will be able to lead as full and unhindered a life as possible.

The wait is not over, there are further hurdles to come, especially with regard to funding in the UK, but we are one step closer.

Andy Brooks

Wedding Congratulations!

We would like to offer our heartfelt congratulations to Hayley Johnson and Philip Edwards who married in a very quiet ceremony on Monday 5th February 2007, at Southover Grange, Lewes Register Officer. Hayley has Niemann-Pick Type B. Very best wishes to you both!



It's a Small World....

In October 2006 I was on holiday with my sister, Alex and my Dad. We were staying at the Esmerelda Park in Cala D'or on the Island of Majorca.

It was a really hot sunny day and my sister and I decided to go swimming. I was just walking into the pool when I noticed a woman in the pool wearing a wristband. It was an unusual lilac colour and I instantly recognised it as a Niemann-Pick one.

I walked over to her and asked if it was a Niemann-Pick wristband – she was very surprised that I had recognised it. She asked me how I knew what type of band it was and I explained that my mum, Sue, worked as the Administrative Assistant for the NPDG (UK) with Toni, the National Development Manager.

The woman's name was Kait Pyne. She explained that her daughter, Annie, who was the same age as me, had NP Type C and introduced us. Annie, Alex and I got on very well and spent much of the remainder of our holiday together.

Emily Lowe age 11



Emily Lowe (left in both pictures) with Alex (top) and Annie (above)

In Loving Memory

We remember

Richard McAdam Age 37

Bryanna De Souza Age 13



In loving memory: Bryanna DeSouza



From Judy DeSouza (Mom) December 30th, 2006

Bryanna was the greatest gift in my life from God. She was a beautiful child and I was so proud to be her mom. I told her every day I loved her. She brought me so much joy.

When I sadly found out that Bryanna had this terminal illness called Niemann-Pick Disease, I realized that this meant she would be mine to care and hold only for a short time. I wanted to make the most of our life together, so we did many things and travelled everywhere. She made me laugh, cry, taught me patience and smiling through adversity, strength and courage. She touched so many people's lives with her smile and positive spirit.

Watching her body decline with this disease over these past years, brought much deep pain to me to watch her suffer and to know that it was only a matter of time that she would be gone from me. The most difficult and biggest gift of love that I gave her was letting her go into a restful sleep and back to God where she will no longer suffer. I wanted to have one more week or month of that smile, but that would not have been fair to her. There is a huge emptiness in my heart and soul

that can never be replaced with anything on this earth. I can still hear her words to me that, although she found it physically difficult to talk anymore, she would often struggle to get out "Are you okay Mom?" and "I love you mom." She will forever remain in my memory and heart forever. I love you too Bryanna.

Love, mom.

Jackie Imrie writes:

"My daughter Beth and I have known Bryanna and her mum Judy for several years. Beth always had the honour of being Bryanna's helper in the kids programme at the NNPDF Conference in the USA. We'll never forget heading off into the Hollywood hills and not getting back until way after bed time. We'll never forget the expression on Bryanna's face when her mum went on the bucking bronco in Dallas –and we have the photos to prove it. We'll miss your smiling face at conference 2007 but we'll look after your mum."

Love Jackie and Beth

In loving memory: Richard McAdam

19.03.1969- 28.11.2006

It was early in the year 2000 when I first turned my car left at the Forth road bridge instead of going straight on into Fife to visit relatives. This was the first time I met Richard, his mum Patsy and dad Matt, and I've had many visits since.

As well as being able to see Richard's smiling face, one of the reasons I visited was to attend the team meetings; I'm sure Patsy must have said to all her team, "If Jackie can come all that way to attend meetings you can come from Falkirk". In spring 2005 there were 17 of us sat around the table with only one apology! This really did mean that everyone working with Richard worked as a team to offer Richard and his family optimum support throughout the many years when he was so dependant.

The team leader was definitely Patsy who knew what Richard needed and made sure he got it. Patsy, Matt and Richard's sister Anne (who made a wonderful communications book when Richard could no longer talk) always made me very welcome. It was very sad over Christmas to visit and not have Richard sat in his chair in the corner smiling or just dozing. We will all miss him greatly.



NATIONAL DEVELOPMENT MANAGER REPORT

You will probably have noticed that this issue of the Newsletter contains rather a lot of 'Research News'. That is, of course, very good news and shows just how much work is being done around the world to find what we all ultimately hope for – a treatment or cure for Niemann-Pick diseases. I realise that some of our readers find the research articles quite hard to read, I have to admit that I sometimes have to ask Jackie Imrie (our Clinical Nurse Specialist) to summarise them in layman's terms for me! Therefore I thought it would be helpful to include a Glossary of Terms with this issue, which will help to explain the medical terms used in the articles. If you still have questions regarding any of the articles we have included, please do contact Jackie on 0161 922 2414 or Liz Jacklin (our Clinical Research Nurse) on 0161 922 2967.



Although Christmas is now just a distant memory, I have to mention our amazing Christmas card sales, as the cards simply flew out of the office! I would like to thank the winning children (Annie, Edward and William) for the hard work they put in designing the cards for the competition and all who so generously supported the Group by purchasing them. I am looking forward to seeing the entries for this year's Christmas Card Competition, which will be on display at our Family Conference in June, where the 2007 winner will be chosen.

The Conference Sub-Committee have been busily planning our 2007 Conference since September 2006. Thanks to their hard work, the programme has now taken shape and we are looking forward to a very exciting weekend. This year's theme is "Focus on the Future" – the amount of work taking place in the field of NPD (as highlighted in this Newsletter) gives us renewed hope and encouragement that we will one day beat this group of diseases. I hope you will join us at Conference this year to make new friends, catch up with old ones and hear the latest information. If you have not attended before and would like to know more, contact me at the office on 0191 415 0693 or email niemann-pick@zetnet.co.uk I will do my best to help.

Along with Jackie, I have been juggling many different projects, all of which we hope to have completed in time for Conference. The booklet we are working on with Colin Cosgrove of the Alzheimer's Society, 'Dementia in Childhood', is almost ready to go to print and, thanks to the help and involvement of our family members, the NP-C Care Manual is making good progress. The NPDG (UK) website is in the process of being completely re-designed. We hope the new look website will be on-line later this year.

Thank you to all of you who took the time to complete and return the Members Questionnaire we sent out earlier this year. The results of this will be published in our autumn newsletter. Your comments and suggestions will help us to shape the service we offer to better meet your needs in the future. There is no need, however, to wait for the next questionnaire if you would like to discuss any aspect of the service we provide. Please just pick up the phone, we are always eager for feedback about our work.

I hope to see you at Conference in June, 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

With spring around the corner, here in Manchester we are wondering if winter is over as we have had so little frost and snow. That suits me fine as I head off to Huddersfield and Newcastle. Little stops me getting to family homes when needed but snow on the highest motorway in England (M62) certainly does.

Here at the Willink we have seen a few changes. Helena Prady has now gone on to pastures new and she faces a lot of challenges in her new role. She will be really missed here, but whilst we are waiting for Actelion to finish collating all of the data on the Zavesca trial Liz Jacklin will be caring for those patients in the extended use phase of the trial.

We have been lucky to get funding from NSCAG to allow another 6 patients nationally to be prescribed Zavesca whilst awaiting the trial outcomes. As with those patients on the trial, we will be monitoring these young people very closely for safety and efficacy.

Talking of Actelion, I was recently asked if I would talk at ZOE, the Zavesca Own Event in Lisbon. When I was asked several months ago it was not made clear that I'd be up on stage in front of over 260 delegates (Thanks Charlotte!) As it happened it went fine as I explained what the end points in the trial meant to patients. I met many eminent physicians and scientists including Hans Klunemann from Germany and Marie Vanier from France (pictured at the faculty dinner) Hans will be working with Liz and I looking at the psychiatric manifestations of NPC and Marie is very much involved with the diagnosis of NPC, so we are frequently in contact. It was great to meet up again with the metabolic nursing teams from throughout the UK and all of the Actelion team.

As most of you are aware all adult patients with metabolic disease are now seen at Hope Hospital by Dr Waldek, our adult physician, with Dr Wraith in attendance. I am also there when our NPD patients



are at clinic. So far this seems to be going well and the nurses at Hope are appreciating meeting families with other metabolic diseases.

As you can see from several references in the newsletter, plans are well underway for this year's conference. If any of you have not attended before and are unsure about it please do give me a ring. It is always hard to come to conference for that first time, but we will support you all the way. It will be very exciting, especially on Saturday night when some of Liz's scouts will be manning the fairground games. Obviously the talks on Saturday and Sunday are important and interesting but this weekend is for you, the families, to meet with other parents and professionals for mutual support and, hopefully, comfort.

Hope to see you there.

Jackie

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

From the editor...

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

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

Please send your articles for the Autumn Newsletter by 31 August 2007

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