

## Partners For A Cure

As we wait desperately for a cure for our children, often the isolation and despair can be overwhelming. However, we are not without hope. A cure will be found. We must all join together to make the difference. The Lee King Benefit Buddy Tournament is partnering with Stratos Boat Company to promote the need for research into NPD. The 4th Annual ~ Lee King Benefit Buddy Tournament ~ will be held on June 12, 2004 at Lakepoint Marina at Lake Eufaula in Eufaula, Alabama. For your \$100 donation to the LKBTT, your name will be entered into a drawing for this 2004 Stratos 285 Boat & Mercury 15 HP Motor. The drawing is to be held on June 12, 2004 at the weigh-in for the Lee King Benefit Buddy Tournament. You do not have to be present to win. We will deliver the prize package to any location in the continental United States. All proceeds benefit the National Niemann-Pick Disease Foundation, Inc.

Please contact Melissa King at 334-687-7753 or go to the [www.leekingbenefit.org](http://www.leekingbenefit.org) for more information. If 200 of our NPD families promote 5 \$100 donations ~ we have the opportunity to raise \$100,000 for NPD research. Can you help?



Our son, Lee, test driving the boat which was generously donated from Stratos Boats.

## Notice Of 2004 Family Conference

Sherry and Chris Richardson along with Amy Lee of Orlando, Florida will act as co-hosts for our "Twelfth Annual NNPFD Family Conference". The **2004 Family Conference**, will be held from July 22nd to 24th at the award-winning Rosen Centre Hotel (1-800-204-7234; [www.rosencentre.com](http://www.rosencentre.com)). Special family conference rates of \$75 per night will be available at the beautiful 17-acre Rosen Center Resort for three days prior to the start of the conference and for three days following. These special room rates will allow our families to take advantage of this exciting family orientated location and plan an extended stay for more family fun. You never know who may surprise us with their attendance at the conference.

~ ~ **Special Theme Park Ticket Offer Available to our NNPFD Families** ~ ~ An offer has been made by a local support group in Orlando to assist our families with NPD children under the age 17, in obtaining complimentary tickets to some popular theme parks. If you wish to receive additional information and an application form ~ please contact the foundation office ([nnpdf@idcnet.com](mailto:nnpdf@idcnet.com)) for additional information.



# NIEMANN-PICK NEWSLETTER

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## Families Unite in Support of Continued NPD Research Efforts



Back Row: Paul and Elisa Beraldo, Chris and John Taft, Lucy and Enio Liegghio, Dr. Marc Patterson  
Front Row ( Seated) Allan, Michael, and Tammy Vaughan, Dr. Joe Clarke

The 3rd Annual "Life For Luke" Dinner Gala was held in Vaughan, Ontario Canada on Friday, March 5th, 2004. Lucy and Enio Liegghio have a strong network of friends and family who help them to pull together a wonderful fundraising event with proceeds to benefit NPD Type C research. This year other NNPFD families traveled to Vaughan to take part in this event as well. John and Chris Taft of Monroe, Michigan, parents of "Little John" and "Baby Kate" who both sadly lost their battle with NPD Type A felt very fortunate to attend the event and noted that it is always so uplifting to see the love and joy that an event like this can generate.

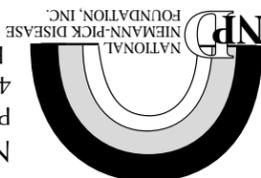
John noted, "The only way to describe the event is WOW!! It was a professional event from the beginning to the end. It is a testament to what kind of people that Luke, Lucy and Enio are to see the support of their family, friends, and community."

They were joined by Tammy and Allan Vaughan of Durham, Ontario along with their son Michael. Last year the Vaughan's hosted their 1st Annual Memorial Golf Tournament in memory of their 2 children, Alex and Laura, who died from NPD Type C. Paul and Elisa Beraldo, of Canada, also made the journey to the Dinner Gala in honor of Luke. The Beraldo's learned recently that their daughter, Taylor age 7, has NPD Type C.

See a recap of this event from the Liegghio's on page 4 of this newsletter.

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From Left: Hunt, Hunter, McNair and Annette Ozmer

## From the Chairman

My name is Hunt Ozmer and I am the new chairman of the National Niemann-Pick Disease Foundation. I am honored for the opportunity to serve you and look forward to working together to find a cure for this dreadful disease.

My life began in North Florida in a now popular area known as Amelia Island. Job opportunities, however, moved our family to a small south central Virginia city known as South Boston. As a Halifax County High School "Fighting Comet" senior I knew it was time to spread my wings. At the Virginia Military Institute in Lexington, VA, I quickly learned how to survive as a "rat" at the "I". Four years and many demerits later I graduated with a BA in history. Thank you God!

December 27, 1981 I made the best decision of my life, I said "I do". My bride Annette has shown me what love and dedication can do to strengthen a relationship. God blessed us with two beautiful children McNair (21) and Hunter (18 NPC). I thank God everyday for giving me these precious gifts of love.

My career has transitioned from restaurant owner to life insurance sales to my current position as packaging product manager, of now 20 years, with NSW,LLC., where sales and marketing have been my strengths.

Over five years ago Hunter and I met, by chance, a man who has come to be a true friend to our family. You all know him as "JB", James Brown, of FOX NFL but to his loving wife, Dorothy, he is known simply as James. "JB" has given generously of his time and talents to help the foundation move forward with its fundraising events and will continue to assist the foundation furthering awareness and research. We are all very grateful for his love and commitment to Hunter and all the Niemann-Pick children.

My goals for the foundation are simple ~ find a cure or treatment for all forms of Niemann-Pick Disease. As a foundation, I am confident we can exceed \$1,000,000 dollars in donations for research within two years. That's a fifty percent increase! To make this happen **you** will have to be involved with the work of the foundation. New board members have stepped up with fresh ideas for advocacy, fund-raising, marketing & public relations and awareness. We must **all** be involved to make a difference.

Thank you for your continued prayers and support as we work together for the future.

Sincerely,  
Hunt Ozmer  
Chairman, National Niemann-Pick Disease Foundation

## A Special Thank You

As I pass the torch of leadership, I would like to thank everyone for making my term as NNPfD Chairman the past four years a very positive and rewarding experience.

One of the main goals during my four-year term has been to establish a base to support continuity and stability for the foundation. With the establishment of a Central Headquarters, the development and implementation of the NNPfD Director's position, and a very strong Board of Directors, I feel, we have systems in place for a smooth transition in leadership. In order for the foundation to continue to grow and prosper, I will now step aside so the new chairman can bring in fresh ideas and new direction to take the NNPfD to the next level.

I know I am leaving the foundation in good hands with Hunt Ozmer as Chairman. Hunt has served on the board since 1999 as Vice Chairman. He has the tenacity and dedication to continue the fight. I am confident that Hunt will help lead the Foundation with a renewed sense of passion.

I will continue to serve on the board as a Director-at-large, volunteering as needed for program services, and overseeing the Central Office in Fort Atkinson.

It has been an honor and a privilege being the chairman of an organization full of such inspiring people. Thank you not only for your personal support but also for your continued support of the NNPfD.

With heartfelt gratitude,  
Barbara Vorpahl



From left: Barb, Gary, Stacey and Kimberly Vorpahl

## Your Child's Summer Reading List...

The NNPfD is an Amazon.com associate. So any purchases you make from Amazon through the NNPfD website earns a commission for the Foundation.



Go to [www.nnpdf.org](http://www.nnpdf.org) and click on the Amazon logo to shop and make a difference for the children...

## Cate Walsh Vockley joins the NNPfD as National Coordinator of Education, Referral and Advocacy

My name is Cate Walsh Vockley, and many of you know that I have been involved with the Niemann-Pick community for a number of years through my role in the Mayo Clinic project on DNA analysis of the NPC1 and NPC2 genes. I recently moved to Pennsylvania as my husband continues his work in research and clinical genetics at the Children's Hospital in Pittsburgh. I am very pleased to announce that I will be the new National Coordinator of Education, Referral and Advocacy for the National Niemann-Pick Disease Foundation. You may have heard that Lori Seidman decided to step down from the position due to new family priorities, and I am happy to continue her important work. I see the focus of the position in 3 major areas: education, advocacy and research. The educational component in part will involve development of packets of materials for families of newly diagnosed patients and for their primary care providers for each of the 3 conditions. I plan to collaborate with the testing laboratories to get these promptly delivered as needed. I will also catalog information about ongoing studies related to NPD, including information about eligibility requirements, and will facilitate participation for interested families. Advocacy comes in many forms, ranging from one-on-one work with families accessing services to interaction with other organizations and agencies to increase awareness of the need for further research in NPD and other rare disorders.

I bring many years of experience in working with families to this position. I obtained my Master of Science degree in genetic counseling from the University of Colorado Health Sciences Center. This education provided a unique combination of knowledge about the scientific and medical aspects of hereditary disease while also emphasizing the psychological effects of such diagnoses. Over the last 17 years, I have worked in Denver, at Yale University and at the Mayo Clinic. I

have always seen myself as an advocate for patients and families, whether working to educate about new diagnoses, obtain appropriate health care, respite care, or insurance coverage, or in working for support for continuing research. In each of my positions, I have worked in a metabolic disease clinic, providing ongoing education, counseling and support for families as they have dealt with challenges of a chronic medical condition in a loved one. Through this work I have developed a relationship with a national network of health care providers with experience in metabolic disease. This will serve as an excellent resource as we strive to obtain experienced providers and prompt diagnoses. I have also been involved in developing programs at an institutional, state and national level for providing and improving services to families, including recent work on the Minnesota planning grant for follow up of newborn screening across the state.

On a more personal note, my husband Jerry and I have been married for almost 24 years (I was sooo young when I got married!), and we have 2 sons, Aaron age 13 and Ben age 10 (or almost 11 according to him). And we can't forget the dog, Zeke, a 90+ lb. Lab mix who loves our new country home. We are busy settling into a new routine and meeting new friends here in Pittsburgh. As a family we share our interests in hiking, camping, canoeing and kayaking, scouting, all kinds of music and, for now at least, yard work! I look forward to continuing my work with all of you, and hope that you will call me when I can help. For now, I can be reached through the Foundation Office. Once the details of my position at the Children's Hospital are finalized, we will publish new contact information so you can reach me there.

## Scientific Advisory Board "Up Close"

### Dr. Robert Maue

Bob Maue's career in science started with an interest in environmental extremes like the bottom of the ocean or the top of Mount Everest. He studied seals at the South Pole but finally found the mysteries of the brain were the ultimate challenge.

Bob's lab began with studies of how neurons develop and, specifically, the development of electrical activity in neurons. Their focus has become "ion channels" and how they are regulated. Bob defined "ion channels" in this way: "Ion channels" are proteins in the cell membrane that, when appropriately triggered, undergo extremely small and rapid changes in shape and conformation, and in doing so form small "pores", "holes", or "channels" in the membrane through which specific ions are able to flow....All electrical signals are the result of ions flowing one way or another across the cell membrane. Without correct expression of these proteins, our nervous system would not function..." Bob's expertise with ion channels prepared him for his current studies on the electrical activity of neurons from the NPC mouse.

Bob's wife is Leslie Henderson, also a neurobiologist. Both teach at Dartmouth Medical School in Hanover, New Hampshire. Leslie has done some work on Niemann-Pick disease in the past and formerly served on the NNPfD's Scientific Advisory Board. Her current focus is understanding the basis for differences in the male and female brain. She studies how sex hormones like estrogen and testosterone affect the properties of nerve cells in a region of the brain known as the hypothalamus.

When asked for the "best" and "worst" of having a career in science, Bob says: "The best part of science is the constant intellectual challenge, the excitement of discovering something you know is new, and the interactions with all types of interesting scientists from around the world - its never a boring job. The worst part is the constant "pull" between spending time with your family and spending more time at work, and the pressure to keep getting research grants."

Bob and Leslie have two boys, Tyler, 16 and Casey, 14. When not working, they enjoy hiking, canoeing, skiing, cycling, and other outdoor activities. However, just now, Bob says he and Leslie spend their "down time" driving all over New Hampshire and New England watching soccer, basketball, hockey, and lacrosse games. I'm an avid reader and like doing landscaping projects around the house."

When asked if there is any message he'd like to give the NP families, Bob said: "Keep supporting each other - I have always been impressed with the strength and support of the NP community, and it has served as a source of inspiration for us during our research efforts."

Dr. Maue joined the Dartmouth Medical School faculty in 1989 and is currently Professor of Physiology and Biochemistry. He received his doctorate in Physiology and Pharmacology at the University of California in San Diego in 1986. He has received fellowships from Brandeis University Department of Biochemistry (1986); from New England Medical Center Division of Molecular Medicine (1987-89); and from the Alfred P. Sloan Foundation (1990-92).



The Maue-Henderson Family

Disclaimer: Information which appears in this newsletter is for the purpose of providing information and awareness and does not necessarily reflect the views of the foundation.

# Research Highlights

by Director of Research, Janet Ward Pease

## ***“Cellular and Molecular Mechanisms Underlying Neurodegeneration in Niemann-Pick Type C Disease” - Makoto Michikawa, M.D., Ph.D.***

Dr. Michikawa works in Japan's National Institute of Longevity Research and became interested in NPC after doing work in Alzheimer's and other dementia-causing conditions. He recently reported that his results to date “suggest that synapse formation and subsequent neuronal network formation are affected in NPC mouse brains. In addition, [his lab] found that the neurons prepared from NPC... brains showed more vulnerability to neurotoxic reagents [than neurons from NPC carrier brains or from normal mice].” Going forward, Dr. Michikawa says: “We are currently performing experiments to elucidate the mechanisms underlying [NPC]-specific impairment of neurite extension and the high vulnerability of [NPC] neurons to neurotoxic reagents. If these mechanisms are clarified, they would provide us insights or clues for the development of therapeutic approaches to stimulating neurite outgrowth, promoting synapse formation, and preventing neuronal death caused by the neurotoxic reagents, leading to inhibition or prevention of neurodegeneration found in NPC disease.”

## ***“Reduction of Lipid Accumulation in Niemann-Pick Fibroblasts by Modulation of Membrane Trafficking” - Amit Kumar Choudhury, Ph.D.***

Last September, Dr. Choudhury, recipient of an NNPDF 2-year fellowship who is working under the sponsorship of Richard E. Pagano, Ph.D at the Mayo Clinic and Foundation, reported:

*“During the first six months of this fellowship we have identified a new trafficking defect associated with the [NPC] diseased cells. We also identified a protein whose function is perturbed in these diseased cells and have been able to alleviate disease phenotype in cell culture by overexpression of this protein. During the next 6 months I plan to continue this work by investigating the mechanism by which this protein alleviates the disease phenotype as well as examine other proteins whose function is perturbed in this disease.”*

Dr. Choudhury's next status report is due March 15<sup>th</sup> (too late, unfortunately, to go into this issue of our newsletter) however a lay summary of his report, along with lay summaries of all NNPDF-funded projects and their associated status reports, are posted on the NNPDF website when they are received. See the “Additional information” column on [www.nnpdf.org/grants.asp](http://www.nnpdf.org/grants.asp) and [www.nnpdf.org/fells.asp](http://www.nnpdf.org/fells.asp).

## **NEW FUNDING**

### ***“SSRIs as a potential therapy for NP-C” - Synthia H. Mellon, Ph.D.***

One of the most impressive “therapeutics” presentations at last year's 2nd International Conference on NPC was that of Dr. Synthia Mellon of the University of California, San Francisco. Her presentation showed the results of her work using the compound allopregnanolone to delay neurological decline and extend the lifespan of NPC mice. After the conference, the NNPDF solicited a grant application from Dr. Mellon to test the possibility of using selective serotonin re-uptake inhibitors (SSRI's) as a treatment for NPC. (The idea for the project is based on the fact that SSRI's have been shown to stimulate the generation of allopregnanolone in the brains of rats.) At its recent Board meeting, the NNPDF joined with Dana's Angels Research Trust ([www.danasangels.org](http://www.danasangels.org)) to award a 1-year grant to Dr. Mellon for this study. Details of the project will be posted on the NNPDF grants page ([www.nnpdf.org/grants.asp](http://www.nnpdf.org/grants.asp)) as soon as a start date for the project is set.

### ***“NMDA receptor hypofunction in Niemann-Pick Disease Type A” - Stuart A. Lipton, M.D., Ph.D.***

A one-year grant was recently awarded to Dr. Stuart Lipton of the Burnham Institute in La Jolla, California. Dr. Lipton describes his project as follows: Our idea all started when we made the observation that the pattern of neurodegenerative events in the brains of Niemann-Pick Type A (“NPA”) disease patients had some striking similarities to two other known disease syndromes, namely the fetal alcohol syndrome (FAS) and the intoxication with anesthetics (IWA). These syndromes are caused by the chemical suppression of the activity of a very important brain protein (the NMDA receptor), which is in part responsible for the communication between nerve cells, and thus for many learning, memory, perception and movement processes. We wondered whether some endogenous chemical compound might accumulate in the brains of NPA disease patients that might have a similar suppressive effect on NMDA receptors, and we have identified in preliminary experiments one candidate chemical compound having this activity. Our project will] ... characterize how this compound suppresses NMDA activity, and ...test in animal models of NPA disease whether the activity of NMDA receptors really is inappropriately suppressed during the course of the disease...If our preliminary data are confirmed, our results would have a direct impact on the development of investigational drugs for NPA disease, since other drugs have had a beneficial effect on the aforementioned “sister diseases” (FAS and IWA). These drugs might then be immediately tested in animal models of NPA disease and eventually in humans.

## **RESEARCH COLLABORATION PROPOSED**

All three types of Niemann-Pick disease are included in the category “lysosomal storage diseases” (LSD's). This means that, in each type of NP, something gets stuck (i.e. “stored”) in a processing compartment inside the cell called a “lysosome”. In types A and B, the stored substance is sphingomyelin; in type C it is cholesterol and other lipids.

Other rare diseases which are LSD's include: Batten, Fabry's, Gaucher's, Krabbe, Pompe, Tay-Sachs, and the mucopolysaccharidoses (MPS) and mucopolipidoses (ML) disorders.

There has recently been discussion among the LSD support groups, including the NNPDF, about jointly funding research projects that are of common interest to all. This would allow us to leverage our research dollars to tackle large projects which no single support group could afford on its own. One example of such a project is research on the blood/brain barrier which is of particular interest to LSD's with central nervous system (i.e. brain) involvement (which includes NPA and NPC). Figuring out how to move therapeutic compounds from the bloodstream to the brain through this barrier would simplify delivery of treatments for all diseases in this group.

A followup on this proposal for collaboration will appear in the next NNPDF newsletter.

# NNPDF 2004 Board Meeting Highlights

The Annual Board of Directors Meeting was held on February 6th–8th, 2004 in St. Louis Missouri. Highlights were:

- A changing of the guard took place with **Barb Vorpahl** stepping down from the Chairman's position at the end of her term. **Hunt Ozmer** was elected as the new Chairman of the Foundation. Also, new to the board are **John Taft**, **Stephanie Cortes** and **Jonathan Jacoby** as Directors-at-Large
- The **2004 budget** was approved with a fund raising goal of \$500,000 and a research budget of \$322,000. Last year, Foundation members and supporters raised over \$480,000.
- **Cate Walsh-Vockley** was selected as the National Niemann-Pick Coordinator of Education, Referral and Advocacy. The position is being expanded to encompass all forms of Niemann-Pick. Cate will be working with the University of Pittsburgh Medical Center to assist families and to develop education programs for health care providers and medical professionals.

- **Research – Synthia Mellon** was awarded a grant to study the effect of SSRIs on Niemann-Pick Type C. **Dr. Stuart Lipton** was awarded a grant to study similarities between Niemann-Pick Type A and Fetal Alcohol Syndrome.
- **Rod Carter Communications** was selected to assist the Foundation with new public relations and fund raising initiatives. Details of these new efforts will be announced in the coming weeks.
- **Strategies for advocating for NPD Disease** at the legislative level and joining forces with other rare disease in the “Global Movement” now taking place for lysosomal storage diseases were outlined. A committee was formed to move forward with these strategies.

We wish to thank Nancy and Art Sullivan for planning this year's meeting and donating meals and refreshments for the weekend. We also offer a special thanks to Cynthia MacLean and Red Robin Gourmet Burgers, Inc for sponsoring Saturday evening's dinner and transportation in St. Louis.

## “Changing of the Guard”

### Introducing new members to the NNPDF Board of Directors



*Stephanie, Tristen and Brian Cortes*

#### **STEPHANIE CORTES ~ NNPDF Secretary**

My name is Stephanie Lyn Cortes. I live in Los Angeles, CA with my husband Brian and my eight-year old son, Tristen.

I work for a cosmetics company doing accounting and I teach spinning on the weekends at a gym called, Bodies In Motion. My son, Tristen, was diagnosed with Niemann-Pick Type B on January 31<sup>st</sup>, 2003.

I am sure as most of you experienced, my life was turned upside down by first learning there was something wrong with Tristen, then the year of performing every test under the sun on him, and followed by the diagnosis. After learning about the diagnosis and how rare the disease is, I was overjoyed when I came across the NNPDF and the listserv. I immediately realized the need for fundraising for all types of Niemann-Pick and went to work on a fundraiser. The First Annual Spin for Tristen was quite a success, bringing in close to \$20,000. The Second Annual Spin for Tristen will take place on April 4<sup>th</sup> of this year.

I am excited to serve as a member of the NNPDF Board for several reasons and feel I have much to offer as a member. My goals as a board member include: spreading awareness about Niemann-Pick Disease to as many communities as I am able to touch, raising as much money as I am physically able to, having a vote when it comes to how that money is spent, helping to unite the Type B families to get them involved in fundraising and understanding the importance of it, giving support and encouragement to newly diagnosed families, offering my financial skills to help in a way they can be useful and giving all of my energy, enthusiasm and complete determination towards finding a cure for all people affected with this disease.

Thank you for the opportunity to serve as a member of the National Niemann-Pick Disease Foundation Board.

#### **JOHN TAFT ~ NIEMANN-PICK DISEASE TYPE A FAMILY REPRESENTATIVE**

I am honored to have been elected to the board of the NNPDF as the NPD Type A Family Representative. My wife, Chris, and I look forward to helping the NNPDF and other NPD families in anyway possible. Chris and I reside in Monroe, Michigan and are fortunate to have a large, strong and supportive family network in our hometown. Chris and I began dating in high school in 1985. Yes, we are high school sweethearts. We were married on June 25, 1994 and after several years of trying, John Taft IV, “Little John”, was born on September 30, 1998. We received the diagnosis of NPD Type A for “Little John” when Chris was pregnant with our second child, Kathryn Grace. “Baby Kate” joined our family on March 27, 2000 and we were stunned to learn that she as well had NPD Type A.

The diagnosis of NPD Type A propelled our families to take action to make a difference in the lives of “Little John”, “Baby Kate” and all NPD children. What a difference they made! Along with our family and friends, we hosted an “Ice-Cream Social” that raised awareness into NPD and funds for research. The response from our families and friends was amazing and the event was a huge success.

Sadly, “Little John” passed away on January 20, 2001 and “Baby Kate” passed away on February 17, 2002.

We have since been blessed with two more children that are Niemann-Pick free and they certainly keep us busy as a family. Jackson Lee joined us on June 27, 2002 and Sarah Grace was born on November 22, 2003. In addition to family activities, I work as a Real Estate Developer and Manager. I also run a Residential Real Estate Appraisal Business ~ in my spare time!! We are fortunate that Chris can be a “stay-at-home” Mom with Jackson and Sarah and we both are thrilled with their daily activities and milestones.

We look forward to helping the National Niemann-Pick Disease Foundation in anyway possible. It's our hope that we'll be able to do whatever is necessary to raise awareness, support families, and ultimately find a treatment & cure for all types of Niemann Pick Disease.



*Chris, Sara, Jackson and John Taft*

## Meeting The Challenge .....

In 2003, NNPDF supporters did a tremendous job!! Totals for the year were in excess of \$483,000. This amount was made possible through the overwhelming success of Niemann-Pick Awareness Month, the First-Time Fundraiser Contest, and fundraising events both large and small throughout the year.

The 2004 annual goal for the NNPDF has been set at \$500,000. To help reach this goal, we need everyone's time, talent and financial support. We hope you will join us in the challenge to make 2004 a true milestone in the foundation's history.

Luke Liegghio



## Teamwork and a "Spirit of Family" propel the 3<sup>rd</sup> Annual Life for Luke Dinner Gala towards another successful year.

It gives us great pleasure to recap our 3<sup>rd</sup> Annual Life for Luke Silent Auction and Dinner Gala.... First, a little bit of history on the event, it almost did not take place. It was not due to the lack of support but due to the fact that our energy level had been depleted over the last year. The only inspiration we needed was the courage and the smile of our hero and guest of honor, Luke Liegghio. With our team of 25 committee members, "Luke's Angels" the wind beneath our wings, we had only six months to move forward.

From the onset guests had to make strategic decisions whether to try and "bring home the bacon" behind our "crack the code" game, purchase a "Luke Bag" or be the lucky one to score the big prize with the NFL Pigskin Team Logo lottery. This was even before they had to decide which of the 200 plus silent auction items to bid on. In addition, everyone was asking themselves the big question of the evening: "How much will the Big Cheese wheel in?"

### The Life for Luke Dinner Gala motto is everyone goes home a winner!

Auctioneer, Claudio "Hopper" Peluso was welcomed with a thunderous round of applause. The big question was finally going to be answered. The Big Cheese was on the auction block again. Claudio had the crowd in a frenzy when we surpassed last year's bid with a bidding war resulting in a \$2,000 final price tag. The big cheese did not leave the Liegghio family as Luke's Great Uncle Onorio, last year's winner's brother, took home the prize. Not only did our auctioneer work his magic on the auction he did a tribute to our committee and guests by performing a lip sync to U2's "ONE". The song was fitting as we are "ONE" together in this fighting battle to find a cure for NPC.

Aside from all the great games that were taking place and the wonderful prizes to be won or purchased we had a packed banquet hall of 340 people who were able to enjoy the sumptuous dinner from Il Cavaliere Banquet Hall and listen to the music arranged by Soundtek DJ.

The event was attended by some especially important people in the fight against NPC: Dr. Marc Patterson from Columbia University in New York and Dr. Joe Clarke from the Hospital of Sick Children in Toronto. We were also strongly supported by three other families afflicted with NPC and NPA; the Beraldo, Taft and Vaughan families.

- Enio & Lucy Liegghio, Luke and Vanessa's Parents

## Our First Time Fundraiser Contest Continues

If you missed last year's contest, don't feel left out. The NNPDF is sponsoring a "First Time Fundraiser" Contest right now.

**The rewards of participating are great (even if you aren't the prize winner):**

- A sense of accomplishment at doing something you thought impossible
- Moving the fight against NPC forward
- Recognition at the Family Conference and in the Newsletter
- Winner receives a complimentary suite at the Family Conference and dinner for four at a local Restaurant

**The rules are simple:**

- This must be your first fundraising event for NNPDF
- Only events held after June 30, 2003 will be counted
- Results & receipts must be to the foundation by July 1, 2004
- NNPDF Board members, officers, and relatives are not eligible
- You can hold more than one fundraiser during the year if you wish. Everyone participating will be First Timers - so everyone has an equal chance at being the top fundraiser. Last year's contest generated an astounding \$72,000 from new events. But whether you raise \$50 or \$5,000, it all helps in the fight against Niemann-Pick. Be sure to notify Nadine Hill at the NNPDF Office that you are participating so she can track your results. **Good luck to everyone ~ we are all winners with this project!!**

## I Am Not There

Do not stand by my grave and weep.

I am not there, I do not sleep.

I am a thousand winds that blow,

I am the diamond glints upon the snow.

I am the sunlight on ripened grain and

I am the gentle autumn rain.

When you awaken in the morning's hush,

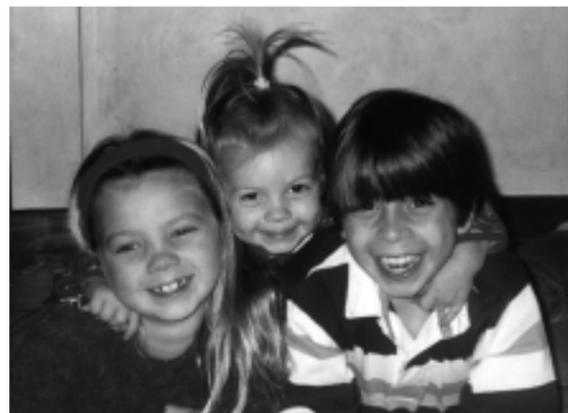
I am that swift uplifting rush,

Of quiet birds in circled flight.

I am the soft star that shines at night.

Do not stand by my grave and cry,

I am not there, I did not die.



Riley, Logan and Blake Donegan

Unbeknownst to me, all of these elated feelings would soon be changed into a life of fear and anxiety. It was October 28, 1996 when Blake looked "funny" to me. I took him to the doctor and told him that he didn't look right; he had lost his beautiful skin tone and started to get dark circles under his eyes. That Dr. looked at me as if I was a pathetic, neurotic, first time mom. He sent me home saying he is just losing his tan and everything was fine. Within two days, October 30<sup>th</sup>, Blake was running a fever, and throwing up. I remember these dates so well because Blake missed his first Halloween that he could walk and say, "Trick or Treat." For the next three months, I was bringing Blake back and forth to the doctor. Finally, after many doctors and fellows examined him, our dooms day had arrived: "Niemann-Pick Type C, we think." The next thing I knew, we were cutting a piece of skin off his arm and sending it off to the National Institute of Health.

While we were waiting the 3 months for the results, we started looking into Niemann-Pick Disease. We ran across the foundation phone number and started to gather all the information about NPC that we could. I was told there was a family conference coming up at the Mayo Clinic in Rochester, MN. My husband and I both attended along with our now two year old son. I couldn't believe my eyes. What a wake up call. Michael and I decided from that point on that we would do what we could to help children with NPC, regardless if Blake was afflicted or not.

Life since Blake's diagnosis has been an emotional roller coaster. I am anxious, scared, happy and sad all at once, and just don't know whether I am coming or going on a daily basis. I worry for Blake's future. I worry that his two younger sisters may have to bury their brother. I hate that the things he loves to do the most are being taken from him by this monster called NPC. Blake is starting to lose his balance. He can no longer remember his addition facts. He has trouble walking up and down stairs. He is not allowed to be in gym class at his school anymore because they are so afraid that he may fall and rupture his spleen. What kind of life is this for a nine year old?

Blake knows something is wrong with his spleen and liver. What he doesn't know is that this is only a symptom of a much bigger problem. He is starting to notice that he is not allowed to do some of the same things that other children his age are doing. He is not permitted to jump on a trampoline, roller blade, play ice hockey, football, soccer, gymnastics, climb trees, or just play a regular boys game of tag. I hate that he has to sit on the sidelines of life. I want so much for my son. I want the sky to be the limit. But, we all know, this is not the case. I fear for the day Blake can no longer eat food due to swallowing difficulties or walk without the assistance of a wheel chair or walker. However, I think the worst thing I can possibly imagine short of my son dying is when he is no longer able to say, "I love you Mom."

For now, I treasure every moment; the almost perfect scores on a test, or even the terrible scores on a test. For while he may have a horrid score on an exam, he is in the classroom, alive and socializing with other children his age. I think all parents want the most out of their child's life. For Blake, or a child with NPC, those things are different from other children, but they are just as important.

Blake touches most every life he comes in contact with. He is so fun and full of life. For every person he interacts with, I know he has touched his or her life forever. I believe one of Blake's purposes on this earth is to help others realize how precious life is and to be thankful for every minute that they are gifted. His time here on earth may be shortened due to this disease, but what an impact he will make. Some people don't realize what it takes to live, or even get up in the morning knowing what we know about our child's future. The struggles that lay ahead for him and how overwhelming the thoughts can be inside our minds. The milestones that some might think are nothing or little, are HUGE in a child with NPC. I thank God for every minute we get to spend together with Blake and his sisters as a family. And, I pray for the day to come when there is a cure for Niemann-Pick. Blake is so special to Michael and me. I can't imagine life without him. We will continue to fight this battle against NPC and look forward to the day that we are all victorious.

### Please take a moment to remember those we have lost recently to Niemann-Pick Disease.

Kayla Barney "Lil Roy" Daniel Teller ~ 13 Corbin Cain ~ 25 Kyle Fitzherbert ~ 14  
Benita Arlene Mitchell Harper ~ 31 Karolee Alice Jane Koonce ~ 25  
Mareena Pamela McNab ~ 14 David Wayne Slater ~ 44 Robert Whitehead ~ 23

Our hearts go out to their family and friends