



# Niemann-Pick

FALL NEWS 2007

Toll Free 1-877-287-3672



**BARBARA VORPAHL**  
NNPDF Board Chair

I love this photo from the family conference. It embodies so well the spirit of this year's event... "today well lived." Through our children, we have come to realize the importance of celebrating each day. Celebration has always been the key to our conferences. ...celebrating our children... celebrating our friendships.... celebrating the hope offered for tomorrow.

At the conference, we commemorated the 15th Anniversary of the NNPDF, taking time to recognize the Foundation's achievements and those individuals who have played a key role in those achievements.

One of those key individuals, Susan Green, stated it best in her remarks from a discussion paper on our accomplishments, "For those of us who were around at the start of the Foundation, it seems that 'yesterday is but a dream'. It was a bad dream - one in which our children were diagnosed with a devastating disease, a

disease that had no hope of a treatment and no hope of a cure. It was a world where we were told, 'Niemann-Pick disease is so rare that you are one of only a few families in the world with this disease'. We all felt so alone but, when we eventually came together with other families to start the Foundation, we had hope. We had a vision of a tomorrow where there would be 'something' that would help our children. In order to realize this new dream we knew that 'somebody' had to do something - so we did! We have been so busy doing 'something', constantly striving for that elusive cure, that we often don't realize what we have already accomplished."

As you read, the Research Highlights Article inside this edition, you will see our "vision of hope for tomorrow" is becoming a reality. Fifteen years ago we were told that a treatment for this disease was not possible. Now, scientists are increasingly certain that they will find effective treatments for all types of Niemann-Pick disease. It is no longer a question of 'if' but a question of 'when'.

The NNPDF is the only Niemann-Pick

family support organization in the United States and Canada. The foundation has always been "families helping families" passing on what they have learned to make things a little easier for others dealing with the same struggles. Fifteen years ago all that was available to families was a few paragraphs of information. Today families and individuals facing a diagnosis of NPD receive a multitude of support services.

You only have to take a look at "NPD Awareness Month" events in this edition to realize we are still doing "something". Families continue to come forward past their own pain and suffering, to raise funds for research. They share their stories putting their families in the "public eye" to create awareness. They reach out asking for YOUR help because we know we can't do this alone.

As we take a step back to acknowledge our accomplishments over the past fifteen years we realize... Together we are a very powerful force that can not and will not be stopped.

*"Family Services ~ Activity Updates"*

**NNPDF CELEBRATES 15TH ANNIVERSARY**

***Family Support, Services, and Research!***

Anticipation ran high as families gathered in the beautiful lakefront city of Milwaukee, Wisconsin this past August for the 15th Annual NNPDF Family Support and Medical Conference. Research, medical and clinical updates for all types of NPD were intermixed with several anniversary celebration events, which helped to elevate this year's conference to a new high!

Our family conference hosts, Barb and Gary Vorpahl, their family and a drove of volunteers, were excited to have the opportunity to welcome conference attendees to Milwaukee for a weekend filled with new family experiences. Conference committee members worked to develop activities and agenda topics that supported this year's theme, "Yesterday, Today, Tomorrow".



*Families attending the summer 2007 family conference took time out of their busy schedules to gather for this group photo at the Saturday evening banquet celebration.*

Agenda highlights included sessions featuring the multi-disciplinary approach to treating the entire child/young adult, educational assistance in approaching and developing an I.E.P., as well as, symptom progression of the disease and a historical perspective on scientific research into a treatment and cure for NPD.

Organizing a conference of this magnitude is not an easy task and we are deeply appreciative of the time and talent that our host family, speakers and volunteers contributed to the overwhelming success of this event. Special thanks go to the Actelion Pharmaceuticals Limited and Genzyme Pharmaceutical Company, which assisted with conference support via educational grants.

Plans for the 16th Annual NNPDF Family Support and Medical Conference are currently underway. Our 2008 conference, hosted by Melissa and Jimmy King, is slated to be held in the Atlanta, Georgia. Watch our web site ([www.nnpdf.org](http://www.nnpdf.org)) and future newsletters for updates.

**"THE NNPDF MEDICAL AND FAMILY CONFERENCE EXCEEDED OUR EXPECTATIONS. WE WERE TOUCHED BY THE CARING AND EMPATHY FROM THE DOCTORS, SCIENTISTS AND RESEARCHERS INVOLVED IN THE CONFERENCE TO THE PROFESSIONALISM AND SUPPORT OF THE NNPDF COMMITTEE MEMBERS AND VOLUNTEERS. HAVING THE OPPORTUNITY TO BOND WITH THE OTHER FAMILIES THROUGHOUT THE DAY AND EVENING EVENTS WAS MEANINGFUL AND MEMORABLE. THE CONFERENCE PROVIDED US ENCOURAGEMENT AND STRENGTH KNOWING THAT WE ARE NOT ALONE IN OUR STRUGGLE FOR A CURE."**

*Family Conference Attendee ~ Summer 2007*

**NEW EQUIPMENT EXCHANGE PROGRAM**

As NPD patient and family needs change during a chronic illness there may be equipment stored in homes that is no longer needed and could be put to use by another family. The NNPDF will assist families who wish to donate this equipment.

The foundation will retain and post on its web site a log that describes the type, size, and

location of available equipment and information about equipment needs. Please contact the central office for more information on this program.

This program is being funded in large part by the Jami and Lisa Chavez family in memory of their daughter, Breann, who passed away from NPD Type C in June 2006, at the age of 4. Their annual "Ducks for Bucks" event



generated the monies to assist in offsetting the cost to ship equipment from home to home.

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**VISIT OUR NEW WEBSITE**



[www.nnpdf.org](http://www.nnpdf.org)

**MEMORIALS**

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

- Spencer A. Bisio (NPC) ~ 31 years
- Anna Marie Evans (Philips) (NPA) ~ 3 years
- Mariah Hoss (NPA) ~ 16 months
- Caroline Jeanne Kirk (NPB) ~ 9 years
- Edward James McCormick (NPC) ~ 6 years

- Vincent K. Steven VanDetta (NPA) ~ 2 years
  - Malayna Renee' Varela (NPA/B) ~ 6 years
  - Nichole Christine Vivian (NPC) ~ 20 years
- Our hearts go out to their families and friends.*

*So much of me is made of what I learned from you. You'll be with me like a handprint on my heart. Because I knew you I have been changed for good.*

# Making Our Voices Heard

"The NNPDF ~ Where Success Begins with You!"

## Papier Family & Friends Fund Raisers



**1st Annual Dillon Papier Charity Golf Outing**  
In honor of: Dillon Papier  
Frederick, Maryland

**"Grandma" Maris Walker's Birthday**  
In honor of: Dillon Papier  
Frederick, Maryland



**Papier Family Bowie Baysox Benefit**  
In honor of: Dillon Papier  
Frederick, Maryland



**Greensboro Grasshopper Fund Raiser**  
In honor of: Dillon Papier - Frederick, Maryland



**Recke Family "5K Run/Walk For a Cure"**  
In honor of: Adam Recke  
Allentown, Pennsylvania



**Corvettes For Adam**  
In honor of: Adam Recke  
Allentown, Pennsylvania



**PSEA Golf For A Cure Tournament**  
In honor of: Adam Recke  
Allentown, Pennsylvania

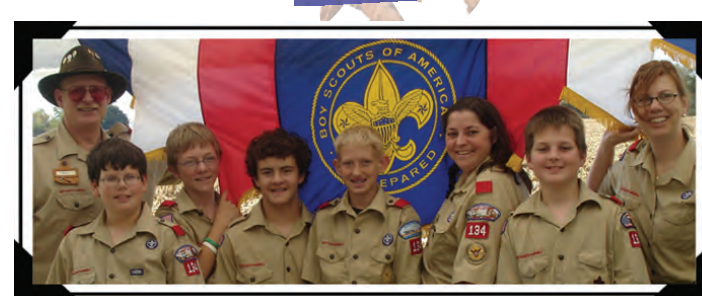
Look to this day  
For yesterday is but a dream,  
And tomorrow merely a vision.  
But today, well lived,



**Tomorrow's Hope Walk Fest**  
In honor of:  
All Members of the NNPDF  
Jefferson, Wisconsin



**"Drive For The Cure" 11th Annual Hunter's Hope Golf**  
In honor of: Hunter Ozmer  
Hanging Rock Golf Course - Salem, Virginia

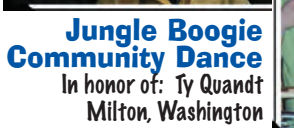


**Tour de Hope Bike Fest - Tomorrow's Hope**  
Members of Boy Scout Troop 134  
In honor of:  
October as NPD Awareness Month and in support of Tomorrow's Hope  
Jefferson County, Wisconsin

Makes every yesterday  
a dream of happiness  
And every tomorrow a  
vision of hope.  
Look well, therefore,  
to this day.



**Ty's Fire Engine Birthday Surprise**  
In honor of: Ty Quandt  
Milton, Washington



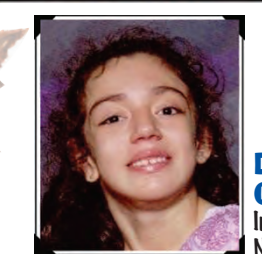
**Jungle Boogie Community Dance**  
In honor of: Ty Quandt  
Milton, Washington



**Essence Food Studio Evening of Entertainment**  
In honor of: Mitchell Harrower  
Melbourne, Australia

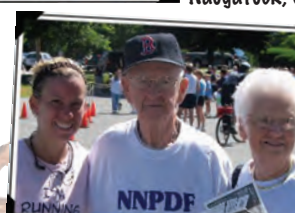


**Chavez Family Memorial Conference Candles**  
In memory of: Breann Chavez -  
Farmington, New Mexico

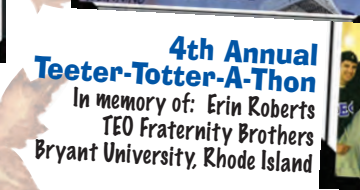


**DeSouza Family Yankee Candle Sale**  
In memory of: Bryanna DeSouza  
Naugatuck, Connecticut

**"Ducks for Bucks"**  
In memory of: Breann Chavez  
Farmington, New Mexico



**Hanover Community Walk Fest**  
In memory of: Erin Roberts - Hanover, Massachusetts



**4th Annual Teeter-Totter-A-Thon**  
In memory of: Erin Roberts  
TEO Fraternity Brothers  
Bryant University, Rhode Island



**Sumbad Family Golf Outing & Silent Auction**  
In honor of:  
Katlyn Rose Sumbad  
Modesto, California



**Vinnie VanDetta ~ Memorial Donations**  
In memory of: Vincent K. Steven VanDetta  
Hamlin, New York

## Tyrrell Family & Friends Fund Raisers

**"Little Feet Making Big Strides" Walk-a-thon**  
In honor of: Naomi Tyrrell  
Shelton, Connecticut



**Jessica Harstmayer Kick-a-thon**  
In honor of:  
Naomi Tyrrell  
Shelton, Connecticut

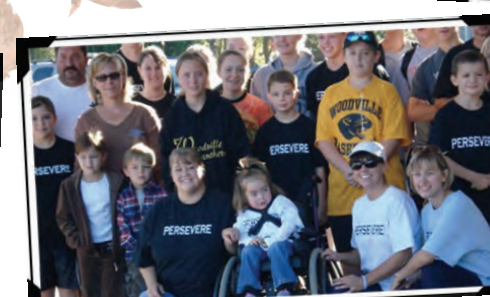
**Tyrrell Family "PERSEVERE" Wristband & "PERSEVERE" Wear Campaign**  
In honor of:  
Naomi Tyrrell  
Shelton, Connecticut



**Heinz Family ~ Faith Awards Dinner**  
In memory of: Tyler Heinz - Elsie, Michigan



**Charlie Green Ironman Triathlon**  
In memory of: Tyler Heinz  
In honor of: Faith and Katie Heinz - Elsie, Michigan



**Riley Corbit Walk-Fest**  
In honor of: Riley Corbit - Woodville, Alabama



**Bailey and Brianna's "Ride For Life" ~ Poker Run**  
In memory of: Bailey Patterson  
In honor of: Brianna Patterson  
Kennewick, Washington

**Klondike Middle School Awareness Event**  
In memory of: Braden Smith  
In honor of: Keaton and Riley Smith  
West Lafayette, Indiana



Brianna & Bailey Patterson



# Research Highlights

*It is no longer a question of 'if' but a question of 'when'.*

## NNPDF RESEARCH OVERVIEW

As we enter this fifteenth year we should be excited by the promise we see in the scientific advances being made towards treatments and a cure for all of the Niemann-Pick diseases. Fifteen years ago very few scientists were working on NPD there were only a handful of research papers available. Today, a Pubmed search will find more than 1800 research papers. Through the efforts of many families and friends, the NNPDF raised funds to support identification of the NPC1 and NPC2 genes. Families also contributed to this work by providing blood and tissue samples. Carrier testing is now available for those families where the genetic mutation is known as are prenatal diagnosis and, in some cases, Pre-implantation Genetic Diagnosis (PGD).

NONE OF THIS WOULD HAVE BEEN ACHIEVED SO QUICKLY WITHOUT THE INVOLVEMENT OF FAMILIES WHO CAME TO THE FOUNDATION FOR INFORMATION AND SUPPORT.

Our Foundation has promoted the trials to a growing number of families and supported those families involved in the studies. In all types of Niemann-Pick disease, we are now entering the treatment era! The current clinical trials have the potential to slow or stop the progression of these diseases, providing scientists and families with more time to make further advances.

IN ONLY FIFTEEN YEARS, NPD RESEARCH HAS MOVED FROM GENE DISCOVERY TO PROMISING CLINICAL TRIALS.

New treatments are being developed based on the scientists' current understanding of how the proteins made by these genes work in the body. None of the progress being made would have taken place without advances in basic understanding. Much of what scientists know about the basic, underlying mechanisms involved in Niemann-Pick disease, they have learned from investigating similar genetic structures in animal and yeast models. The Foundation

has provided funding to study the disease process in these models and to help develop and maintain valuable colonies of mice and cats.

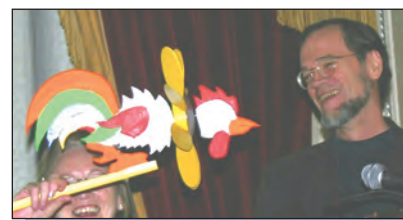
The NNPDF was crucial in identifying patients, to enable statistically viable data to be gathered in preparation for the clinical trials, in recruiting patients for the trials and in supporting families through the process. Without the families there would be no clinical research.

Fifteen years ago when this Foundation started, many were told that a treatment for these diseases was not possible. Now, scientists are increasingly certain that they will find effective treatments for all types of Niemann-Pick disease.

IN ADDITION TO THE FUNDS OUR FAMILIES AND FRIENDS RAISE FOR RESEARCH, OUR BIGGEST RESEARCH RESOURCE IS THE FAMILIES THEMSELVES.

## SCIENTIFIC ADVISORY BOARD CHAIR CHANGE

Our Scientific Advisory Board serves an essential role in providing expert guidance to the NNPDF Board on the Foundation's research strategy. The SAB also help the Foundation to assess the quality of research proposals and decide how best to support research that will move us toward an effective treatment or cure. The dedicated scientists and physicians who serve on the board share their expertise willingly, without expectation of compensation. Little did Dr. Steve Walkley know, when he informed the Board that he would be stepping down as SAB Chair after 4 years, that he would be well-rewarded for his efforts. Thanks to Nadine's husband Mike, Dr. Walkley is now the proud owner of a chicken weather vane (see photo)! Be sure to ask Dr. Walkley about his chickens and personally thank him for his generous service to the Foundation. We are very grateful for his gift of time and talent, especially as he has agreed to continue to serve as a member of the SAB (5 years and counting...).



At the same time, we welcome a long-standing member of the SAB as our new SAB Chair. Dr. Dan Ory has been a member of the SAB for 5 years and has agreed to step into the Chair position.



Dr. Ory is an Assistant Professor of Medicine, and Cell Biology & Physiology, Cardiovascular Division, in the Department of Medicine at Washington University School of Medicine in St. Louis, Missouri. Dr. Ory earned his M.D. from Harvard

Medical School, and did his extensive post-graduate clinical and research training at Brigham & Women's, Harvard, Massachusetts General Hospital and the Whitehead Institute for Biomedical Research. He has received numerous academic awards including Physician Scientist and Career Development awards from the National Institutes of health, as well as RO1 funding. His current research focuses on identification and characterization of genes that function in the uptake, intracellular transport, and export of lipoprotein-derived cholesterol.

We thank Dr. Ory for taking on this new role within the Foundation!

VISIT OUR NEW WEBSITE  
  
[www.nnpdf.org](http://www.nnpdf.org)

*The NNPDF is seeking to fund excellent projects that directly investigate Niemann-Pick Disease (NPD). Pilot Studies of up to \$50,000 per year are funded for one year for researchers who are new to the NPD field or established NPD investigators who wish to*

*test an innovative idea. Research Grants of up to \$100,000 per year are funded for two years to investigators who are studying either basic mechanisms or treatment approaches of direct relevance to NPD.*

*Applications are accepted from U.S. and non-U.S. scientists and consideration for larger studies are accepted with prior approval from the Chair, NNPDF Research Committee, Susan Green (E-mail: [susangee@zetnet.co.uk](mailto:susangee@zetnet.co.uk)).*



# 2007 Year-End Gift Ideas

Each year, hundreds of families around the world depend on the NNPDF for medical information, research updates and emotional support as they struggle to care for a terminally ill child or young adult. We, in turn, depend on those with compassion and the financial resources to make a difference in the lives of those who are suffering.

As you consider your year-end tax planning, we at the NNPDF hope you will consider making good use of the income tax charitable deduction. Your 2007 year-end gift can significantly reduce your income taxes, while providing meaningful support for the work of the NNPDF.

Here are some of the best 2007 year-end gift ideas. We would be pleased to provide you further information upon request.

**GIFTS OF CASH** ~ Mail Checks To: NNPDF, PO Box 310; Fort Atkinson, WI, 53538-0310. Via Credit Card At: [www.nnpdf.org](http://www.nnpdf.org)

**TRIBUTE GIFTS** ~ Financial gifts can be made in honor or in memory of a friend, family member or colleague. They can also be made to mark any special occasion, such as an anniversary, birthday or graduation.

**MATCHING GIFTS** ~ Many employers will match charitable gifts, meaning your donation dollars are worth even more. If your company or firm has a matching gift program, simply enclose the form along with your check. We will take care of the rest.

**UNDERWRITING GIFTS** ~ You can help to defray the ever-increasing cost associated with the development and support of the programs provided by the NNPDF. A major gift can help to underwrite and sustain

the organization's overall financial health. You may choose to support a specific program, such as the Annual Family Support and Medical Conference, the position of the Coordinator of Education, Referral and Advocacy, the publishing the Niemann-Pick Newsletter, or research ventures.

**UNITED WAY CONTRIBUTIONS** ~ Employee directed contributions can be made via United Way by "writing in" the National Niemann-Pick Disease Foundation, Inc. Tax Exempt ID #: 35-1844264.

**COMBINED FEDERAL CAMPAIGN (CFC)** ~ Contributions can also be made by Federal and State employees who wish to support the foundation by selecting the NNPDF #10121 located under National/International Independent Organization on your CFC pledge card.

**GIFTS-IN-KIND** ~ Donations of printing, advertising, promotional items, office supplies, etc. assist the foundation in keeping our overhead costs low and move more funding directly towards family support and research.

**GIFTS OF STOCK** ~ A transfer of stock and other securities is almost always more tax-wise a contribution than cash.

**PLANNED GIVING** ~ Bequests made through a will, trust, life insurance, or other estate planning can have a significant impact on NNPDF programs and services will into the future. Please contact the Foundation office for more information.

Your charitable gifts make an important difference in what we are able to accomplish. Thank you for your generosity.

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

*Celebrating 15 Years of Support, Service and Research!*

NATIONAL NIEMANN-PICK DISEASE FOUNDATION

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