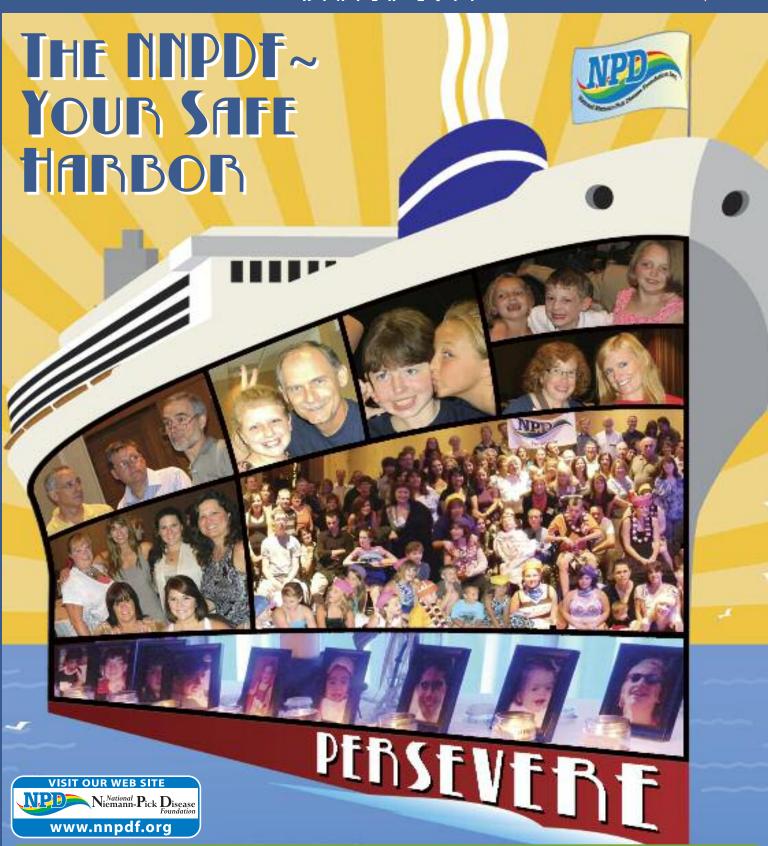
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WINTER 2011

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A publication of The National Niemann-Pick Disease Foundation (NNPDF)

NATIONAL NIEMANN-PICK DISEASE FOUNDATION

"Foundation Updates"

"Nothing in this

world can take the place

of persistence. Talent will not.

Genius will not. Education will not.

Persistence and determination

alone are omnipotent.

The slogan 'press on' has solved and

always will solve the problems



Karen Quandt NNPDF Board Chair

2011 is coming to a close and the National Niemann-Pick Disease Foundation (NNPDF) has had a busy and productive year, persevering in our Quest for a Cure. Our heartfelt thanks goes out to you and all our

supporters for the many roles played in advancing our mission to find effective treatments and a cure for all types of Niemann-Pick Disease. This newsletter highlights exciting progress made by the foundation, its families, researchers and supporters this year.

The NNPDF year officially began on a snowy weekend in February, when the Board of Directors met in Milwaukee, not far from the foundation's home office in Fort Atkinson, Wisconsin, to take stock of the previous year's accomplishments and set goals for the year ahead.

Among those yearly goals were ambitious plans to continue driving NPD research forward. It is research that will find effective treatments and a cure for NPD, and we are very excited about the projects the foundation has been able to support. Today's dedicated scientists are building on the knowledge gained by those who have gone before – their perseverance brings us ever closer to our goal.

With your generous support, the NNPDF is currently funding four postdoctoral research fellows and an NPC calvin Coolidge cat colony. Further, researchers have identified medicines to bring to clinical trials for both ASMD (NPD Types A and B) and Niemann-Pick Disease Type C (NPC) families a patients. Our anticipation grows as plans for a clinical trial for cyclodextrin takes shape. You can read more about all these exciting research projects and developments later in the now and newsletter.

NNPDF families were updated on the latest research at the 19th Annual NNPDF Family Support and Medical Conference held in

Norfolk, Virginia, in July. Our families are the heart and soul of the foundation and the conference is a family reunion where we can connect with others who understand the day-to-day struggles of Niemann-Pick Disease, and find answers to questions about the disease. Visit our Web site at www.nnpdf.org for photo slideshows from the conference.

Many successful awareness and fundraising activities were held by our families and supporters during October Awareness Month and throughout the year, providing essential support for the NNPDF's programs of Research and Family Services. The NNPDF's 11-11-11 Challenge spread awareness about Niemann-Pick Disease across the country and the world.

The International Niemann-Pick Disease Alliance (INPDA) met in November in Talavera de la Reina, Spain, to share updates and to reinforce and develop worldwide networks. Representatives from patient support groups (including those from

the NNPDF and CCNNPDF) and pharmaceutical companies, along with clinicians and scientists from around the world, were in attendance.

Please take a few moments to check out the rest of this newsletter for more details about the foundation's activities. In addition to this newsletter, the NNPDF works hard to keep families and supporters informed about the latest news and research via the Web site, listserv groups, email, e-newsletters, Facebook, the blog, and of course the telephone and mail. Stay tuned to stay in touch!

Throughout the year and around the globe, the NNPDF has persevered in the Quest for a Cure for Niemann-Pick Disease. It's the generosity of people like you that allows the NNPDF to PERSEVERE year after year in support of all families affected by NPD, and it is purely through perseverance that we will find effective treatments and a cure for all types of Niemann-Pick Disease. We thank you for your dedicated support

now and through 2012!

Karen R. Quandt, R.N., M.S.N. NNPDF Board Chair

VISIT OUR WEB SITE

Negational Pick Disease Foundation

www.nnpdf.org

The 2011 NNPDF Annual Meeting will be held in Milwaukee from Friday, February 10th through Sunday, February 12th. The Foundation's annual board meetings are open to all members of the NNPDF. Please refer to the Web site (www.nnpdf.org) for more details or contact the NNPDF Central Office if you are interested in attending.

MEMORIALS

Melana Marie Elfe (NPC) ~ 35 years Daniel Jonathon Flinton (NPC) ~ 4 years Guy Michael Borsetti Heikila (NPC) ~ 31 years Jimmy "Lee" King (NPC) ~ 20 years Stephanie Francis Lawrence (NPC) ~ 24 years Gavin Luiz Lopez (NPC) ~ 7 years Tehyez Brandi Minde (NPA) ~ 2 years Sawsan Moubarak (NPC) ~ 10 years Adam James Edward Reimer (NPC) ~ 24 years Michelle Rose Trombley (NPC) ~ 24 years Mia Jeanette Walts (NPA) ~ 2 years Right now I'm in a different place, and though we seem apart, I'm closer than I ever was, I'm there inside your heart. ~ Abbey Press

Please take a moment to remember those we have lost recently to Niemann-Pick Disease. Our hearts go out to their families and friends.

NATIONAL NIEMANN-PICK DISEASE FOUNDATION

"Our Quest for a Cure"

2011 NNPDF Family Support and Medical Conference

The National Niemann-Pick Disease Foundation's 2011 Family Support and Medical Conference, held in Norfolk, Virginia, was a resounding success, with the largest attendance at any Family

Conference to date! Families from the U.S., Canada, and several other countries came together in late July to learn the latest in research and clinical care, and to share in supporting one another along the difficult journey presented by Niemann-Pick Disease.

The NNPDF Annual Family Conference strives to maximize opportunities for families to interact not only with each other, but also with the country's eminent physicians and scientists working on Niemann-Pick Disease. Families in attendance heard medical/clinical and research updates, with separate sessions for ASMD (NPD Types A and B) and NPC (NPD Type C), including the opportunity to submit questions to the panel of experts.

Conference attendees enjoyed the waterfront setting and a plethora of sightseeing opportunities in the historic Norfolk area during their free time. On Friday evening many families attended the nearby Peninsula Pilots' baseball game, dedicated to the memory of Kevin Eadie (NPC), who had served as a batboy for the team. The conference closed with a celebratory banquet, complete with the traditional "Singing in the Rain" and "Chicken

Members of the NNPDF board and staff extend their greatest appreciation to the many volunteers and speakers, the staff others who went above and beyond the call of duty

to make the conference such a wonderfully enriching experience. Special thanks to our Family Conference sponsors – Genzyme and Ducks for Bucks!

For more information and photos from the 2011 Family Conference, visit the NNPDF Web site (www.nnpdf.org).



First-Time Fundraiser Winner



Simon and Heather (Patenaude) Taillefer of Quebec, Canada, were the winners of the 2011 First-Time Fundraiser contest. Heather and Simon are the doting parents of Monica Taillefer, the spirited two-year-old who has captured our hearts, beating the odds and exceeding all expectations in her battle against Niemann-Pick Disease Type C (NPC).

The Taillefers dedicated themselves to raising awareness and funding, holding a bowling event with raffles, door prizes, food sales, etc. They also wrote letters to their families and friends updating them on "Princess Monica's" progress and challenges, asking for donations to the foundation in honor of their beloved baby girl.

As the First-Time Fundraiser contest winners, the Taillefers enjoyed a complimentary hotel room for the three nights of the 2011 NNPDF Family Support and Medical Conference held in Norfolk, VA.

The foundation is so very grateful for the awareness and fundraising efforts of the Taillefer family and all other families, friends and supporters who make the NNPDF's programs of research and family services possible. Together, we can make a difference!

CANADIAN CHAPTER OF THE NATIONAL NIEMANN-PICK

DISEASE FOUNDATION INC

www.nnpdf.ca



Canadian Chapter Update

The Canadian Chapter of the NNPDF (CCNNPDF) has put in another busy and successful year raising awareness and funding and providing support to families affected by Niemann-Pick Disease.

A sister chapter to the National Niemann-Pick Disease Foundation, the CCNNPDF receives administrative support from the NNPDF. Money raised through the CCNNPDF is invested in Niemann-Pick Disease research

through the NNPDF's research program with guidance from the NNPDF's Scientific Advisory Board (SAB), Board of Directors, and Research Committee. This partnership helps eliminate redundancy and maximizes the impact of our research dollars. In 2011, the CCNNPDF contributed \$75,000 CAN toward Niemann-Pick Disease research initiatives.

Tammy Vaughan and Sandra Cowie serve as representatives of the CCNNPDF on the NNPDF Board of Directors and are both active members of the foundation's Research Committee.

During 2011 many Canadian families, including the Taillefer family, winners of the First-Time Fundraiser contest, hosted awareness and fundraising activities in support of the CCNNPDF's essential programs of Research and Family Services. Two other major fundraisers were hosted by

the Tammy and Allan Vaughan family, in memory of their children Alex and Laura (both NPC), and the Lucy and Enio Liegghio family, in memory of their son, Luke (NPC).

Many CCNNPDF families participated in October Awareness Month activities and 20 percent of the families who took on the NNPDF's 11-11-11 Fundraising Challenge were Canadian. Additionally, several CCNNPDF

> member families have signed on to make regular contributions through United Way payroll deductions. This generous support, combined with the resources of the NNPDF, continues to drive progress against NPD forward.

> The CCNNPDF's collaborative efforts reach beyond North America, with two representatives of

the CCNNPDF having attended the recent International Niemann-Pick Disease Alliance (INPDA) meeting in Spain, along with Dr. Julian Raiman of the Division of Clinical and Metabolic Genetics at the Hospital for Sick Children in Toronto.

We want to express our deep appreciation to the members of the CCNNPDF for their generous support of our Quest for a Cure during 2011, and we look forward to continued success together in 2012!





Dillon Papier Charity **Golf Tournament Bowie Baysox, Frederick** Keys, and Baltimore **Orioles Baseball Benefits Boy Scout Fundraiser** Collin's Lemonade Stand **Dunkin' Donuts Grandma Maris**¹ Vendor Party

Irwin Family Golf Outing, Jewelry Party and Birthday Party **Rebel Race**

Nanny Charlotte's 90th Birthday Nanny Rose's 95th Birthday Foster's Grille **Monthly Family Night Subway Donation Jar** In honor of Dillon Papier (NPC) Frederick, Maryland

In honor of Andrew Coppola (NPC)

Bristol, Pennsylvania

Yankee Candle Sales

Otis Spunkmever

In memory of

10th Annual Life for Luke

In memory of Luke Liegghio (NPC)

Gala and Auction

Vaughan, Ontario, Canada

Cookie Dough Sale

Bryanna DeSouza (NPC)

Naugatuck, Connecticut



"What Matters Most"

Heritage Day Booth

Chuck Beckman (NPC)

Fundraiser Boutique,

Bake Sale and Raffle

Scentsy Candles and

Simi Valley, California

Baseball Game

Kevin Eadie (NPC)

Hampton, Virginia

In memory of

Premier Design Jewelry

In honor of Mitchell Brubaker (NPC)

Wylder Nation Kickoff Event

Facebook Page and Blog

Scottsdale, Arizona

In honor of Wylder Laffoon (NPA)

Rook Sales

In memory of

Jasper, Alabama

Family Birthday Donations Yukon Quest and Iditarod Musher Races In honor of Lily Beaupre (NPA) Enterprise, **NW Territories, Canada**

Pillows for Pat

Fundraiser

Patrick Gaffney

Naperville, Illinois

In honor of



Research Is Our Only Hope

"The NNPDF ~ Where Success Begins with You!"

Wine Tasting **PTO Penny Drive** Letters and **Brochures Campaign**

Feather Hair **Extensions Events** Governor's and Mayor'

Proclamations In honor of Collin and Cohen Parody (NPB) Dexter, New York



Facebook Cause

In memory of Gavin Lopez

4th Annual Ryan's Ride American Legion Poker Run Motorcycle Raffle

In memory of Ryan Richardson (NPC) Crestview, Florida

Casino Night

Grain Valley, Missouri

Brisan and Parker Stults (NPC)

In honor of



2nd Annual Ride4Tylor

RAGBRAI Team

Walk for Wishes

Hair Cut-a-Thon

Tilt4Tylor

Coins for Comments Blog

Dubuque Bruisers Football

Moms and Me Presentation

n honor of Tylor White-Richardson

Dubuque, Iowa, and Galena, Illinois

5th Annual Katlyn Sumbad Golf Tournament Katlyn's Birthday Donations In memory of

PERSEVERE Wear and Products Sales Calendar Sales ladarola Family Collection Palomba Family Can Collection











In memory of "Little Roy" Teller (NPC) Chula Vista, California







15th Annual **Hunter's Hope Memorial Golf** Tournament and Auction In memory of Hunter Ozmer (NPC)

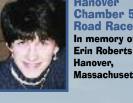




Erin Roberts (NPC)







Johnathan's Dreams





School of the Arts

Blue Hawaii Party

Edgewood, Washington

Notre Dame Presentation

Governor's Proclamation

n honor of Ty Quandt (NPC)

Walk-a-Thon

Loganfest IV

Katlyn Sumbad (NPA/B) Modesto, California











National Pick Disease

www.nnpdf.org

Our Ouest for a Cure ~ Families Unite to Host **Marcello and Matthew Perugini Memorial Walk New Britain Rock Cats Baseball Benefit** Niemann-Pick Disease Awareness Events and Activities In memory of Matthew and Marcello Perugini (NPC)
Waterbury, Connecticut Across the United States and Canada Save Gavin

2nd Annual Zoe's Fun Run In memory of Zoe Bachman (NPA/B) King's Mountain, North Carolina



Kaitlyn Bourgeault (NPA) Charlotte, North Carolina







Poker Run Cycle Ride 5th Annual Woodville Student Government **Association Walk** In memory of Riley Corbitt (NPC)

Ducks for Bucks

Willowwicks Candle Sales

Governor's Proclamation

In memory of Breann Chavez (NPC)









Bowling Event

In honor of

Grand-Mere,

Quebec, Canada

and Fundraiser

Monica Taillefer (NPC)



lex and Laura Vaughan emorial Golf Tournament

Alex and Laura Vaughan (NPC) Durham, Ontario, Canada

Annual Golf FORE Staces **Memorial Golf Classic** and Silent Auction **Barrie Elementary** Mini-Grant Fundraise **Governor's Proclamation** In memory of Stacey Vorpahl (NPC) Fort Atkinson, Wisconsin



Scentsy Wickless Candles Fundraiser

In memory of Damon Cardinal (NPA)

Edmonton, Alberta, Canada

Awareness

Michael Cordero

Yard Sale

In honor of





Vinyl Banner Sales Donations







ADVANCING KNOWLEDGE ~ TRANSFORMING LIVES

"Research Updates"

NNPDF's 11-11-11 Challenge Raised Funds for Research

In light of the unique, once-in-a-lifetime date of 11-11-11, the NNPDF issued a special challenge to its members and supporters to raise awareness and funding for essential research. Those who accepted the challenge asked their families, friends, neighbors and co-workers to each donate \$11 to the NNPDF by 11-11-11. One hundred percent of the proceeds raised by the Challenge will go to support research, designated by disease type (ASMD/Types A & B or NPC).

Many thanks to all who took the Challenge! Their combined efforts raised \$20,000 for Niemann-Pick Disease research!

- Bexton Family (MI) in Honor of Kaitlyn Bexton and in Memory of Alexandria Bexton (both NPC)
- Bourgeault Family (NC) in Honor of Kaitlyn Bourgeault (NPA)
- (NPA) Cardinal Family (Alberta, Canada) in Memory of Damon Cardinal
- Chavez Family (NM) in Memory of Breann Chavez (NPC)
- Owie Family (Canada) in Honor of Sandra Cowie (NPB)
- Onegan Family (IL) in Honor of Blake Donegan (NPC)
- Fagan Family (MI) in Honor of Mindy Fagan (NPC)
- Flinton Family (NY) in Honor/Memory of Daniel Flinton (NPC)
- Follett Family (WA) in Memory of Emma Shaffer (NPC)
- Glassman Family (VA) in Memory of Sarah Glassman (NPA)
- Higgins Family (NC) in Honor of Melissa Higgins (NPB)
- Hughes Family (TX) in Memory of Riley Corbitt (NPC)
- Mehoe Family (RI) in Memory of Graham Kehoe (NPC) and Kevin Kehoe
- Melly Family (RI) in Memory of Erin Roberts (NPC)
- (NPA) Laffoon Family (AZ)- in Honor of Wylder Laffoon (NPA)
- Lane Family (NY) in Honor of Collin and Cohen Parody (both NPB)
- Marocco Family (MD) in Memory of Scott Marocco (NPC)
- Martino Family (Canada) in Honor of Mariarosa Martino (NPB)
- McGuire Family (CT) in Honor of Kelly Thompson (NPC)
- Minde-Redcrow Family (Canada) in Memory of Tehyez Minde (NPA)
- OConnor Smith Family (FL) in Honor of Annie OConnor Smith (NPB)
- Papier Family (MD) in Honor of Dillon Papier (NPC)
- Patterson Family (CA) in Memory of Bailey and Brianna Patterson (both NPC)
- Purpura Family (NY) in Honor of Ashlyn Purpura (NPC)
- Quandt Family (WA) in Honor of Ty Quandt (NPC)
- Quesada Family (NY) in Memory of Valeria and Fernanda Vargas Arias (both NPC)
- Reedy DiGiovanni Family (VT) in Honor of Chase DiGiovanni (NPC)
- Roberts Family (MA) in Memory of Erin Roberts (NPC)
- St. Germain Family (Canada) in Honor of Kaiden St. Germain (NPC2)
- Stults Family (MO) in Honor of Brisan and Parker Stults (both NPC)
- Taillefer Family (Quebec, Canada) in Honor of Monica Taillefer (NPC)
- Teller Family (CA) in Memory of "Little Roy" Teller (NPC)
- Vaughan Family (Canada) in Memory of Alex and Laura Vaughan (both NPC)
- Vorpahl Family (WI) in Memory of Stacey Vorpahl (NPC)

NNPDF-Funded Research

Research into Niemann-Pick Disease is a key focus of the NNPDF's Quest for a Cure, and the foundation is pleased to be able to support the projects of researchers dedicated to unlocking the mysteries of the disease.

During 2011, the foundation, with the recommendation of the experts on the NNPDF's Scientific Advisory Board (SAB), selected two additional research projects to fund:

 Andrew Munkacsi, Ph.D. (Columbia University Medical Center) – "Defining the molecular basis of HDAC inhibitors to treat murine, feline, and human models of NPC"





 Charles H. Vite, D.V.M., Ph.D. (School of Veterinary Medicine, University of Pennsylvania – Philadelphia) – NPC Cat Colony

These two projects bring the total number of studies being supported by the NNPDF/CCNNPDF to six, including the recently completed project of Ian Williams, Ph.D. (University of Oxford, UK), and the ongoing projects of Fabrizio Vacca, Ph.D. (University of Geneva, Switzerland), Dorothea Maetzel, Ph.D. (Whitehead Institute for Biomedical Research, Cambridge, MA), and Nicholas Cianciola, Ph.D. (School of Medicine, Case Western Reserve University, Cleveland, OH).

Support for this essential research is possible due to the hard work of our NNPDF/CCNNPDF families to raise awareness and funding, and the continued generosity of our donors and friends.

NNPDF-funded researchers provide regular updates regarding the progress of their work. Please visit http://www.nnpdf.org/npdisease_14.html for more details and links to their reports.

The application deadline for the next funding cycle is May 1, 2012. Please visit http://www.nnpdf.org/PeterPentchev ResearchFellowship.html or email nnpdf@nnpdf.org for more details.

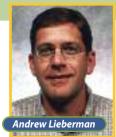


NNPDF Welcomes Two New Members to Scientific Advisory Board

The NNPDF is pleased to welcome two new members to the Scientific Advisory Board (SAB). Fran Platt, Ph.D., Professor of Biochemistry and Pharmacology at the University of Oxford, and Andrew Lieberman, M.D., Ph.D., Associate Professor and Director, Neuropathology, University of Michigan Health System, both recently joined the SAB.

The SAB gives guidance to the NNPDF's Board of Directors in determining policies and funding decisions related to research. The commitment and guidance of the SAB has been critical to the NNPDF's success and we greatly appreciate all their hard work on behalf of our families.





ADVANCING KNOWLEDGE ~ TRANSFORMING LIVES

"Research Updates"

Genzyme Raises Awareness During October Awareness Month



Genzyme conducted an in-house awareness campaign for October **Niemann-Pick Disease** Awareness Month. Posters featured a photo of Collin and Cohen Parody (both NPB), grandsons of NNPDF **Board Member Kathy Lane.** Genzyme remains committed to their efforts to advance the enzyme replacement therapy trial to Phase 2 for our Type B patients. Pictured are members of Genzyme's Clinical Development and Patient Advocacy teams.

NIH to Develop Clinical Trial for Cyclodextrin

The National Institutes of Health (NIH), in collaboration with the Therapeutics for Rare and Neglected Diseases Program (TRND), is developing a clinical trial utilizing cyclodextrin for Niemann-Pick Disease Type C (NPC) patients.

The clinical trial is in the planning phase and many criteria must be met and numerous approvals granted before the trial can begin. Dr. Forbes "Denny" Porter, Senior Investigator at the NIH, and Dr. Daniel Ory, NNPDF Scientific Advisory Board (SAB) Chair from the Washington University School of Medicine, are working collaboratively to bring this trial to our NPC patient community.

To keep families affected by NPC up to date on plans for the trial, the NNPDF hosted an open conference call in May, and Drs. Porter and Ory gave a presentation at the 2011 NNPDF Family Conference in late July. Most recently, a meeting was held between TRND and the U.S. Food and Drug Administration (FDA) to talk about plans for the trial. Dr. Porter reported after the meeting that there is still

plenty of work to be done in preparation for the trial, but that the meeting was very positive.

With regard to the TRND team (see photo below), Dr. Porter said, "It is an impressive collection of expertise and this group has greatly facilitated moving the effort to develop a safe and effective therapy for NPC forward. We know the photo is missing one critical facet of the team - the families."

While everyone is eager to learn more about the upcoming trial, the criteria for participation have not been set as of our publication date. Further information will be posted to the NNPDF's Web site (www.nnpdf.org) as it becomes available, and the NNPDF will advise member families when applications for participation are open. (NPC families -- Please be sure the NNPDF has your current email and mailing addresses. Email changes to nnpdf@nnpdf.org.)

To read the complete report from the TRND/FDA meeting, please visit http://www.nnpdf.org/Cyclodextrin.html.



FRONT ROW (LEFT TO RIGHT): Ilona Scott (J&J), Pat Frenchick (RRD), Sandie Morseth (RRD), Kimberly Lilly (RRD), Mark Kao (J&J), Nicole Yanjanin (NIH/NICHD), Liz Ottinger (NIH/TRND), Nuria Carrillo-Carrasco (NIH/TRND), Xin Xu (NIH/TRND)

SECOND ROW (LEFT TO RIGHT): Steve Walkley (Albert Einstein College of Medicine), Charles Vite (UPenn), Chuck Finn (RRD), Joy Vander Wal (RRD), Steven Silber (J&J), John McKew (NIH/TRND), Denny Porter (NIH/NICHD), Dan Ory (Washington University School of Medicine), Chris Austin (NIH/TRND), John Heiss (NIH/NINDS), Wei Zheng (NIH/TRND) NOT PICTURED: Juan Marugan (NIH/TRND), Bill Pavan (NIH/NHGRI), and Pramod Terse (NIH/TRND)

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Gifts that Keep giving...

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 2011 year-end gift can significantly reduce your income taxes, while providing meaningful support for the work of the NNPDF.

Hold Fast

Stick To 1

Pursue

ersever

Keep At

Hang

NNPDF, PO Box 310, Fort Atkinson, WI 53538-0310

Via Credit Card at: www.nnpdf.org

Gifts of Cash ~

Mail Checks To:

Tribute Gifts ~ Memorials, Anniversaries, Birthday Celebrations

Matching Gifts ~ Employee Business and Corporate Matching

Underwriting Gifts ~ Support a program or service that has special meaning to you

United Way Contributions ~ Via employee payroll deductions ~ Tax Id #: 35-1844264

Combined Federal Campaign (CFC) ~ Via employee payroll deductions ~ #10121

Gifts-In-Kind ~ Donations of product and services in support of our programs

Gifts of Stock ~ A transfer of stock is a strong tax-wise contribution over cash

Planned Giving ~ Bequests through wills, trusts and estate planning

VISIT OUR WEB SITE Niemann-Pick Disease www.nnpdf.org

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

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.

THE MADE-YOUR SAFE HARBOR

Get It Done

Fort Atkinson, WI 53538-0049 National Niemann-Pick Disease Foundation



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