



NIEMANN-PICK NEWSLETTER

Volume 3, Issue 35

Toll Free 1-877-287-3672

Winter 2004

NNPDF Central Office Contact Information

Phone: 920-563-0930
Toll Free: 1-877-287-3672
Fax: 920-563-0931
E-mail: nnpdf@idcnet.com
Website - www.nnpdf.org

Office Address

415 Madison Ave, P.O. Box 49
Fort Atkinson, WI 53538-0049

NNPDF Coordinator of Education, Referral and Advocacy

Cate Walsh Vockley
Phone: 412-692-7349
Children's Hospital of Pittsburgh
Medical Genetics
E-mail:
catherine.walshvockley@chp.edu

Contributions and Membership To NNPDF

Post Office Box 310
Fort Atkinson, WI 53538-0310

Scientific Advisory Board

Steven U. Walkley, DVM, Ph.D.
Chair
Rose F. Kennedy Center

Robert J. Desnick, Ph.D., M.D.
Mount Sinai School of Medicine

Yiannis A. Loannou, Ph.D.
Mount Sinai School of Medicine

Laura Liscum, Ph.D.
Tufts University School of Medicine

Robert A. Maue, Ph.D.
Dartmouth Medical School

Dan Ory, M.D.
Washington University School of
Medicine

Marc C. Patterson, M.D.
Columbia University, NY

Peter Penchev, Ph.D.

Edward H. Schuchman, Ph.D.
Mount Sinai School of Medicine

Steven Sturley, Ph.D.
Columbia University College of
Physicians and Surgeons

"A Collector of Hearts"

Families unite in their efforts to promote October 2004 as National-Niemann Pick Disease Awareness Month.

Once again, we have learned that the success of the NNPDF begins with our families! Excitement, involvement and interest continue to build each year as NPD families from around the Nation and Canada join together to spread awareness and raise funds in support of our battle against Niemann-Pick Disease.

This year, the children of the foundation were invited to take part in a "Junior Collectors Day" at the Queens Museum of Art in New York City. In addition to displaying their collections, information about the work of the Foundation and Niemann-Pick Disease was distributed to the attendees. Melissa King, our Director of Fund Raising, wrote the following article for the museum event in honor of her son, Lee.



Our son, Lee, is now 13 years old. He was diagnosed with Niemann-Pick Disease type C at age seven. He is now in a wheelchair, he is fed through a feeding tube and he no longer speaks.

I was asked recently about taking a picture with Lee and his "collection" because we had the opportunity to raise awareness about Niemann-Pick. It is an exciting idea but I did not know what to do. Healthy boys at this age have many different possible collections ~ baseball cards, girls' phone numbers, sports trophies....the list could be endless.

My child can't make a collection for himself he really can't tell us what his interests are. So, I tried to think of what to take a picture of. He has an extensive video collection given in place of real gifts because no one has an idea of what to buy for him. He has many, many stuffed animals from several hospital visits. He has albums of pictures taken at different events over the years, where money was raised to help fund a cure for his disease. He

has a few autographed items that celebrities have given him when they wanted to do something nice for him.

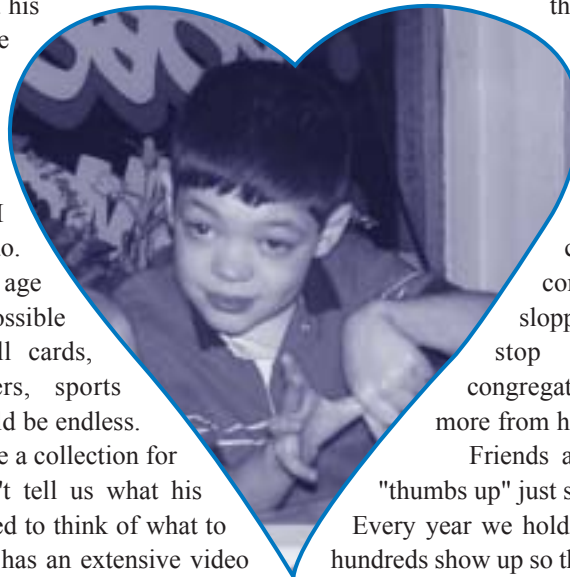
But, there is nothing tangible that Lee collects.

As I thought about this right down to the deadline for turning our entry in, I realized it would be really cool, if we could collect on film all the lives our little boy has touched. You see, our son has collected hearts. He gives complete strangers hugs and sloppy kisses. His laughter will stop an entire church congregation just so they can hear more from him.

Friends and family beg him for a "thumbs up" just so they can smile with him.

Every year we hold events to raise funds and hundreds show up so that hopefully a cure will be found and he can live. Our son is a gift and people fall in love with him wherever he goes ~ he is a collector of hearts.

Submitted by Melissa King
NNPDF Director of Fundraising



From the Chairman

I'm sure you have heard the expression "no man is an island." This expression takes on special meaning for all of the families affected by Niemann-Pick Disease. We cherish the love, support and fellowship of other families who understand what it means to fight for the life of a loved one. For it is when we stand together that our voice rings the loudest. This concept is gathering momentum for the Niemann-Pick Disease Foundation and for other non-profit patient organizations.

We have begun to partner with other Lysosomal Storage Disease (LSD) groups to increase our numbers and improve our chances for government funding of research. This partnering has taken the number of affected patients from less than 300 in the USA for Niemann-Pick to over 1 in 7000 births that are affected by some form of rare LSD.

The National Institutes of Health (NIH) has begun a vigorous program to support and encourage this collaboration by funding broad application research which can benefit many rare LSD's including Niemann-Pick.

I am leading my first full Board of Directors meeting in February 2005 and I would appreciate the support of your prayers. We can beat this disease but we can't do it alone. My vision for our foundation includes increased support of public relations. James Brown of FOX NFL recently recommitted himself as our National Spokesperson to help us increase awareness. JB and my daughter Hunter recently taped public service announcements at a Comcast studio which, thanks to JB, donated all the studio time. We will distribute these PSA's along with press kits to all families who are willing to take their message to the media for increased awareness in their communities.



Our foundation is blessed to have the services of Cate Walsh Vockley serving as the National Coordinator of Education, Referral and Advocacy. Cate comes to us with a broad educational background and years of experience in genetic counseling most recently at the Mayo Clinic in Rochester, MN. Cate is helping newly diagnosed families with information and support and is providing a clear direction for undiagnosed families. Cate has developed a detailed library of information related to Niemann-Pick and I encourage all families to seek her assistance.

We recently enjoyed the many exciting activities and events taking place throughout the country in recognition of October as Niemann-Pick Awareness Month. In Roanoke, VA. we listened to the Christian music of "This Day" and then Annette and I gave a power point presentation describing our work locally as well as the wonderful work being done by the foundation and all the families throughout the US. We had a nice crowd in attendance and were able to also get our local CBS affiliate to provide coverage on

their evening news.

I am looking forward to 2005 as we continue to spread the word of the plight of our precious children and adult family members who are stricken with Niemann-Pick Disease. I appreciate your support and look forward to working with you as we stand together, voices united, to make a difference.

Hunt Ozmer
NNPDF Chairman

headquarters in Los Angeles as well. They readily agreed to support my Fox NFL Sunday colleagues in wearing the NPD lapel pins on our broadcast over the Christmas Holidays. I can't tell you how thrilled I am. Finally, do know, as I've mentioned previously, I am committed to doing all I can to increase awareness and support for research regarding Niemann-Pick Disease.

Be assured, Dorothy and I will continue to pray fervently, knowing that a cure is at hand.

GOD bless you,
James Brown
Fox NFL Sunday

James Brown and the Fox NFL Sunday team will be wearing the NNPDF lapel pins for their December 26th pregame show. The pregame show begins at Noon (EST) and the explanation for the pins is expected to be the first item discussed. Please let your friends, family, and supporters know about this event so they can watch also. NNPDF is working to get additional local coverage from Fox affiliates. If you can assist with media contacts or would like more information, please contact the foundation office at: nnpdf@idcnet.com or 920-563-0930.



Hello from "JB",

Recently, my wife Dorothy and I were blessed to open our home to Hunter (NPC) and Hunt (her dad) Ozmer. While we have bonded as families over the years, this overnight stay allowed us to witness firsthand the determination, perseverance, commitment and love that parents and NPC kids alike, display daily in meeting challenges. It strengthened our resolve to be capable and effective partners

in any way we can. I'm so thankful that the Bethesda, MD based Comcast Sports operation, where I do a weekly sports report, enthusiastically agreed to let us use their studio to tape several Niemann-Pick Public Service Announcements. As usual, Hunter was the star of the show, and made a wonderful and lasting impression on EVERY crew member in the studio. Her impact was a lasting one on my co-workers and executives at Fox Sports

Meet the New Vice Chairman of the NNPDF



Doug Pease was recently elected as the new Vice-Chairman of NNPDF but he has been involved with the Foundation since attending his first family conference in 1997. Doug and his wife, Janet, are the uncle and aunt of Adam, Chelsea, and Amanda Ward. Adam and Amanda have Niemann-Pick Type C. After Adam was diagnosed, Doug and Janet went to their first family conference when Adam's parents couldn't attend.

"We went to the conference almost totally uninformed. There was little information available about Niemann-Pick and all we really knew was that it was a terminal disease. I had no idea what to expect and was pretty nervous. Then I met this wonderful group of people and immediately became part of the Niemann-Pick family."



"Season of Hope"

I have been sitting at my computer trying to think of something inspiring to write to our membership. Something wonderful that might help to refresh some of you,

something intriguing to those of you who are new to the NNPDF, but most of all, something to motivate ALL of you. As I sit here typing, thinking, deleting, retyping, I realize, I really have nothing new.

We've lost so many loved ones this year. Some who were here at the beginning of this foundation. Because of such losses, it can become incredibly hard to continue to hope for a "timely" cure of this disease. So many of our children and young adults are growing worse and we have to painfully watch them lose pieces of themselves ~bit by bit. And still, newly diagnosed families are joining the foundation and list serve all the time ~hoping. Hoping that we'll have answers, hoping there is a treatment or a possibility of a treatment, hoping their child won't begin to lose their abilities ~ unable to imagine facing wheelchairs, feeding tubes and hospital beds.

The Peases volunteered to develop a website. "Our first email from the website was from a family in New Zealand who had never talked with another NPC family. Since then I have heard from people in Japan, the Middle East, Europe, Canada, and South America. I'm always amazed at the impact of the website and grateful that I could do something to help."

Joining the Board in 1998 as Director of Computer Services, Doug later volunteered to be chairman of the Bylaws Committee. He continues to maintain the Foundation's website.

The One Child Left Behind page on the website features Adam from ages 2 through 9. Nowhere is it said that Adam passed away a year later. Amanda, who can no longer walk or talk, is in the Photo Gallery as a toddler, along with pictures from families around the world. "It's my way of remembering who they were and who they could have been. It's a way of not giving up to this disease."

Originally from Massachusetts, Doug migrated to New Orleans in 1979 where he met Janet. They were married in 1992. Doug has two children from a previous marriage and two grandchildren. He works for Affiliated Computer Services managing the computer center of the local water utility.

Doug and Janet, along with Jim and Missy Ward, graciously hosted the Foundation's 10th annual family conference in New Orleans.

So here we are, smack dab in the Season of Hope. So I sit here wondering, what hope can we offer to these families? I'll be honest, as I look at our son, Lee, it becomes harder to believe that there will be a cure for him on this side of Heaven. And as that thought settles into my being, a sweet peace comes ~ I have an assurance that we have done everything possible to help him. He has the best possible care we can give him, he is loved beyond measure and we have continually raised funds and awareness to help find a cure that may or may not come in his lifetime.

So.....here is my "cheerleader" part. We families (old and new) raise funds because we have no other choice. There will be a cure some day. Already, research has brought some very real possibilities for therapies. We still have a long way to go. A hard lesson to learn is that anything worthwhile takes time and diligence. We persist because we must. As research builds on research, a cure will be found. I challenge each and every family to be able to stand up on that day and say, "I did my part."

Merry Christmas and God Bless
Melissa King ~ Mom of Lee age 13, NPC
Director of Fundraising



NNPDF Central Office Year-end Wrap Up

2005 Family Conference

"The Magic of Our Children"

It is with great pleasure AND excitement that we pass along the details for the 13th Annual NNPDF Family Support and Medical Conference to be held in Manhattan Beach, Los Angeles in August of 2005!

Our gracious family hosts' for this upcoming event are Stephanie Cortes of Granada Hills along with Brian and Deborah Demeules of Torrance, California. We would like to offer them a GRAND note of thanks for stepping forward and taking on this task ~ it is appreciated more than you know!!

Stephanie, Brian and Deborah toured a number of hotel locations in and around the Los Angeles area and found a wonderful hotel for the conference event that will offer many sightseeing and outside activity options for families to enjoy. The conference will be held at the Manhattan Beach Marriott at 1400 Parkview Avenue in Manhattan Beach, CA 90266. Check out their web site for more details about the hotel at: www.marriott.com/laxmn

The location and hotel site is just beautiful and is located close to the beach boardwalk, shopping and movie theatre options ~ not to mention the many restaurants which are within walking distance as well. We have also been assured that there isn't ANY humidity in California!!

The conference dates are:

Thursday ~ August 4th thru Sunday ~ August 7th, 2005.
The group event room rates are: \$109.00 per night with a 10% tax and 12 cent resort fee ~ apx \$120.00 per room per night. (These rates will be available to families 3 days prior to the conference dates and three days after.) You may call directly to the Marriott Reservations line at: 1-800-228-9290 or (310)546-7511 to make your reservations, please be certain to note that you are with the National Niemann-Pick Disease Foundation or NNPDF.

We will be sending out additional information and registration packets to families in early 2005 and the Cortes and Demeules families have plenty of ideas in store for everyone. We look forward to working with all of the families for, yet another, successful NNPDF family support and medical conference.

Happy Vacation Planning!!

First Time Fundraiser Contest

We have had such positive feedback and involvement from new families who wish to take part in raising awareness and funds to support the work of the NNPDF that we will continue to sponsor the "First Time Fundraiser" contest again this year. If you missed last year's contest, don't feel left out ~ the NNPDF is sponsoring another "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 may have thought impossible.
- Moving the fight against NPD forward
- The top fundraiser will be recognized at the Family Conference and in the Newsletter
- Winner receives a complimentary suite at the Family Conference for 3 nights and dinner for four at a local restaurant. This year's conference will be held at the beautiful Manhattan Beach Marriott in Manhattan Beach California ~ August 4th 7th, 2005.

The rules are simple:

- This must be your first fundraising event for the NNPDF
- Only events held after June 30, 2004 will be counted
- Results and receipts must be turned into the foundation by July 1, 2005
- NNPDF Board members, officers, and their 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. Each year we have had fun and creative events added to the foundation's fund raising events. But whether you raise \$50 or \$5,000, it all helps in the fight against Niemann-Pick. Please 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!!

2005 NNPDF Board Meeting Announcement

Mark your calendars!! The 2005 Annual NNPDF Board Meeting will be held Friday, February 11th ~ Sunday, February 13th, 2005 at the Wingate Inn in Maryland Heights, Missouri (outside St. Louis, MO). Art and Nancy Sullivan have again offered to host this annual meeting. Art has been the foundation's acting Treasurer for the last year and we all appreciate that he and Nancy have offered to take on this project again for 2005. This annual Board Meeting is open to all members. Please contact the NNPDF Central Office if you are not a board member but would like to receive information pertaining to this meeting.

Activity Update

From Cate Walsh Vockley, MS, CGC
Coordinator of Education, Referral and Advocacy

It has been a busy few months since I settled into my new office at Children's Hospital in Pittsburgh on August 29. Actually, my file boxes aren't even unpacked yet because the file cabinets just arrived this week, but that hasn't kept me from being productive. My biggest and most rewarding activity has been the many e-mail and telephone interactions I have had with families. Calls come from parents, grandparents, siblings and other relatives who were looking for information for themselves and for the affected family member. Most of the contacts have been about NPC, but I have had a number of contacts regarding NPB as well. I have made referrals to experienced healthcare providers, tracked test results for families, and have sent information for families and for health care providers.

Several of my tasks were part of my original proposal, while some new things have been added. In keeping with my proposal, I have put together packets of materials on NPA/B and NPC for health care providers, and packets for patients and families will be completed soon. The health care provider packets each contain several medical articles on the condition including an extensive review article, additional testing information, medical specialist information and patient resource information including key worldwide web addresses. I encourage you to let me know if you would like this information sent to your physicians or other health care providers. These packets will be routinely reviewed and kept up to date. I have also collected a comprehensive list of published articles about Niemann-Pick Disease and current research into causes and potential treatments. This will be computerized so that it will be searchable in the event that I am asked a question about a specific feature of one of the conditions. This will also be updated frequently to be sure that it contains the most recent articles about the quickly changing body of knowledge about NPA, B and C. It

may also be helpful to keep a less formal list of articles that families have found useful. If you read something in any magazine, or have a favorite book or other resource that was helpful or addressed issues important to you, please let me know. I will keep a list and Nadine will send it out to everyone with each newsletter.

One new project that I have been working on is development of a presentation or display about the Foundation that can be used at various meetings. For example, Nadine Hill and Rhonda Brown-Kehoe attended the National Organization for Rare Disorders meeting in Minneapolis this October. We had hoped to have the display completed for that meeting, but had too little time to meet this goal. The text for the display is complete, and we will be deciding how to combine it with pictures to most effectively tell the story of the Foundation's goals and activities. When completed, this display can be used at fund-raising activities, professional meetings, or at any gathering where our message can be shared. We are also looking at what professional meetings we should target for use of this display to most successfully raise awareness about NPD.

In discussion with various members of the board, I have also been pulling together information about 2 additional activities. As many of you know, the Coriell Cell Repository in Camden, New Jersey now has a collection of NPC cell lines. Clinical and DNA data about many of these cell lines are also available. These cells are being made available to researchers who are studying NPC. People at Coriell are interested in increasing the number of cell lines available, and we are discussing how this can be accomplished. We are also learning more about development of a Niemann-Pick disease patient registry, another tool that will help as research moves new ideas and possible treatments into the clinical world.

I look forward to hearing from more of you in the future. Please let me know your thoughts and ideas about my work for you and for the Foundation. I may be reached at 412-692-7349 or via e-mail at: catherine.walshvockley@chp.edu.

Family Medical History Day

U.S. Surgeon General's Family History Initiative

Health care professionals have known for a long time that common diseases - heart disease, cancer, and diabetes - and even rare diseases - like hemophilia, cystic fibrosis, and sickle cell anemia - can run in families. Tracing the illnesses suffered by your parents, grandparents, and other blood relatives can help your doctor predict the disorders to which you may be at risk and take action to keep you and your family healthy. To help focus attention on the importance of family health history, U.S. Surgeon General Richard H. Carmona, M.D has launched a national public health campaign, called the U.S. Surgeon General's Family History Initiative, to encourage all American families to learn more about their family health history.

National Family History Day

Surgeon General Carmona has declared Thanksgiving 2004 to be the first annual National Family History Day. Thanksgiving is the



traditional start of the holiday season for most Americans. Whenever families gather, the Surgeon General encourages them to talk about, and to write down, the health problems that seem to run in their family. Learning about their family's health history may help ensure a longer future together.

My Family Health Portrait

Because family health history is such a powerful screening tool, the Surgeon General has created a new computerized tool to help make it fun and easy for anyone to create a sophisticated portrait of their family's health. This new tool, called "My Family Health Portrait" can be downloaded for free and installed on your own computer. This tool will help you organize your family tree and help you identify common diseases that may run in your family. This is a powerful tool for predicting any illnesses for which you should be checked and will help families forward significant family health information to their primary physicians. For information on National Family History Day and to download your free "Family Health Portrait" Form please access: www.hhs.gov/familyhistory.



Annual Membership Renewals Due

We want to send along a reminder that the annual NPD family membership dues expire on December 31st annually. The nominal \$20 membership fee was established to help offset

the printing and mailing costs for our newsletters, direct mailing announcements and family directories. If you wish to renew your membership with the NNPDF please forward your dues payment in the remittance envelope provided within the pages of this newsletter.

Making Our Voices Heard

“The NNPDF ~ Where Success Begins with You!”

Arlington, Texas

Family, friends and classmates of Andrew Limer pooled their resources in Arlington, Texas to host an awareness campaign and fund raising event at Andrew's school. Children and families at Mary Moore elementary school hosted a coin drive run by the Student Council called "Nickels for Niemann-Pick". The drive spanned the last two weeks of October and the kids collected \$1,302.69. On November 11, 2004, the student council, along with principal Debbie Allison and Andrew's teacher Becky Cagle, presented the Limer family with a check in front of the whole school. We are honored and humbled that the community gathered together in such a positive show of support for Andrew, our family and all children of the NNPDF.



Melbourne, Florida

The Sherry and Chris Richardson family hosted their second annual October NPD Awareness Month in Melbourne, Florida in spite of the “triple threat” of hurricanes-Charley, Frances and Jeanne. Their entire community suffered from these storms, but everyone still pulled together and they had a wonderful event, which was hosted by Mr. and Mrs. Borrelli of Sonic Drive Inn. Once again, the Richardson's friends from Sonic ensured that the event was a fantastic hit. They planned a classic car show, jumping gym, face painting and the Richardson's were able to distribute and promote the Stratos boat tickets and NPD educational information for the foundation. Another wonderful couple, Ted and Maxine of Accent Marine, Inc. of Tampa drove over a 285 Pro XL Stratos Boat to be displayed. The main cheerleader and guest of honor at this event was Little Ryan Richardson. Thanks to everyone who helped make this event a success.

Sherry, Christopher, Rachel, Jordan and Ryan



October 2004 ~ 3rd Annual Niemann-Pick Disease Awareness Month

Once again, we were all overwhelmed and truly inspired as families across the country and Canada planned awareness campaigns and fund raising events. We thank everyone for joining together to spread the word and for your financial support to assist the NNPDF in the battle against Niemann-Pick Disease.

New York, New York

This year, the children of the NPD foundation received a special invitation to take part in a "Junior Collectors Day" at the Queens Museum of Art in New York City. In addition to displaying their collections, information about the work of the Foundation and Niemann-Pick Disease was distributed to the attendees. The organizer of this event, Harley Spiller, ~ aka ~ Inspector Collector, has advised the foundation that Junior Collector's Day at the Queens Museum of Art was a giant success. The Museum was overflowing with more than 40 kids and their cool stuff. Nearly 200 adults marveled at the 36 tables of collections, including a comprehensive set of Tweety Bird toys, a 143 pound rubber band ball, a collection of miniature mugs, and a mermaid collector who dressed up as a mermaid and hid among over 100 different mermaids. The NNPDF was represented by Ryan Richardson, who displayed his quilt made of police emblems donated from precincts across the United States, Stacey Vorpahl sent a photo of herself with her "snazzy sock collection" and Melissa King, our Director of Fund Raising wrote the "Collector of Heart's" article from the front page in honor of her son, Lee.



NPD literature was distributed to hundreds of visitors, many of whom were learning of NPD for the first time. Thank you to David and Sandra Hale, Uncle and Aunt of Ryan Richardson. They represented the NNPDF and the Richardson family and answered many questions about Niemann Pick Disease. They enjoyed their day by seeing different types of hobbies children have done and were honored to represent their nephew Ryan and all the children of the foundation. They also received patches to add to Ryan's blanket.



A special thanks to Harley Spiller ~ Inspector Collector, along with the members of the education department at The Queens Museum of Arts for their dedication and support in encouraging our NPD children to take part in this event, helping to raise awareness into NPD and for ensuring that every single participant's involvement was a positive experience.

New York, New York

Jonathan Jacoby and his wife, Donna Bojarsky, worked to raise awareness into Niemann-Pick Disease through a “Family and Friends” letter. When their two-year son, Joshua, was diagnosed with NPD Type C they became committed to learn all that they could about the disease, possible treatment options and on-going research. They learned that the accumulated research has shown that progress towards treatment of NPD-C may help in the search for a cure to other disorders that involve cholesterol metabolism, including Alzheimer's Disease. There is also recent evidence of a link between NP-C and Cystic Fibrosis. Pharmaceutical companies have already begun to show interest in NPD-C research, which could geometrically increase funding. Jonathan and Donna shared this information with their family and friends in a hope to educate them into the plight of their precious son, Joshua, and all children diagnosed with NPD and in a continued effort to raise additional funds for the essential research that is needed to find a treatment and cure.



Geneva, New York

The 3rd Annual “Angels for Ashley” Awareness event took on a new twist this year. Fourteen-year-old Ashley Sims, NPD Ty C, worked along side her Mom, Juanita Aikens, and many family and friends to develop a “NPD Awareness Youth Night” for Ashley's friends. The local YMCA offered the use of their facility gymnasium and pool for a night filled with swimming, games, basketball, dancing and fun. The Aikens' family approached many of the local business in Geneva, NY to request product donations that could be used for door prizes and to help offset overhead costs. We all know how much high schoolers can devour so these donations were especially appreciated. Ashley was able to shine with all of her friends and family at the “Youth Night” as they offered their love and support while working to educate her friends and neighbors about Niemann-Pick Disease. The event went off without a hitch and the outpouring of support that they received from their community once again, overwhelmed the Aikens family.



Plainville, Connecticut

Twelve-year-old Bryanna DeSouza (NPC) continues to inspire family and friends with her engaging spirit and never-ending smile. Bryanna's mom, Judy, worked with the Governor of Connecticut to garner a proclamation declaring October 2004 as National Niemann-Pick Disease Awareness month in the state.



In addition, Judy also received support from her friends and co-workers at Bic Pen, once again, as she distributed the NPD awareness campaign material along with a Yankee Candle fund raising catalog throughout the office.

Roanoke, Virginia

On Saturday, November 6th, the Hunt and Annette Ozmer family held their October Niemann-Pick Disease Awareness event at Cave Spring Baptist Church in Roanoke, Va. A local Christian band named "This Day" played music and lifted praises to God for Hunter's life and the work the Ozmer's do to help make a



difference for Hunter and for all NPD children. Hunt and Annette gave a very powerful presentation at intermission to describe the way that God is working in Hunter's life and to explain the disease and how it affects her. Hunter called out the winning ticket numbers for the door prizes. The local CBS television affiliate covered the event on their 11 pm news.

Lexington, Texas

The 2nd annual "Walk For A Cure" sponsored by the Lanny and Linda Jo Ruthven Family and held in memory of their daughter, Lanna Jo, took place on October 23rd, 2004. Although dangerous storms rolled through the area the night before the event the weather had cleared in time for the "Walk For A Cure" kick off and didn't dampen any spirits.

Lanna Jo's family continue to receive support from the many members of their community who still work diligently to raise funds so that a cure can be found for this horrible disease. The Ruthven's noted that this event wouldn't have been a success without the ongoing commitment of family, friends and businesses within the town of Lexington. Their hope is that the "Walk For A Cure" will inspire others to join along in supporting the work of the NNPDF and the tireless efforts of our researchers and scientists.



Fort Atkinson, Wisconsin

The community of Fort Atkinson rallied around the family of Stacey Vorpahl upon learning of her death from NPD Type C on October 9th. The Vorpahl's received hundreds of condolence letters and were overwhelmed by the many acts of love, support and kindness offered them from family and friends around the world. In another tribute to Stacey and the NNPDP, the Vorpahl's requested that all memorials be made to the foundation in memory of their beautiful and courageous daughter.

In early September, the Vorpahl family had begun to take steps to work with Governor James Doyle to proclaim October as NPD Awareness Month for the state of Wisconsin. In late October, Barb and Gary journeyed to the State Capital to meet privately with Governor Doyle to receive this proclamation and to discuss the work of the foundation, which Stacey had inspired. Local Representative James Ward joined Governor Doyle and the Vorpahl's to commemorate the awareness proclamation for the State of Wisconsin. Pictured from left to right are Local Representative James Ward, Governor James Doyle, and Gary and Barb Vorpahl.



Jefferson, Wisconsin

Cub Scouts of Pack 149 in Jefferson, WI along with Boy Scouts of Troop 134 in Fort Atkinson, WI performed double duty in early October when they hosted a rest stop during a bike race for a Tomorrow's Hope fund raising event. The NNPDP is a recipient of funding from the Tomorrow's Hope group based in Jefferson and the Scouts were working in honor of Stacey Vorpahl and the National Niemann-Pick Disease Awareness Campaign during the month of October.

The Scouts were kept busy preparing and offering refreshments to the 951 bikers who enjoyed a beautiful fall Wisconsin day as they took part in this 2nd annual event to raise funds for Tomorrow's Hope. An informational booth and display with NPD items was available for the bike riders to learn more about Niemann-Pick Disease while enjoying their tasty treats. A copy of the NNPDP promotional brochure was also distributed to each rider. Way to go Scouts!!



Making Our Voices Heard (CONT.) ~ Our Canadian Neighbor's Unite

Ladysmith, British Columbia

Fundraisers can take on many different shapes and forms ~ or shall we say ~ "Corns"!! Once again this year the Murray McNab family worked all summer to develop a corn maze on their farmland that has become a unique tool for both raising awareness into NPD and funds for research. During the month of October the McNab family hosted, "Corn 4 A Cure" Community Care Fund where they hosted exciting trips through the corn maze with proceeds to further research into a cure for NPD. This year's maze included a tribute to the memory of their sweet daughter, Mareena, who died from NPD Type C in January of this year at the age of 14. The family was able to incorporate her name into the maze design.



Mareena's Dad, Murray, sent along this poem which he distributed to the individuals who toured the maze during October ~ he wrote this in memory of Mareena.

*Our little hummingbird's wings, Beat too fast
But with the Niemann-Pick Disease, She just couldn't last.
Like a shooting star in the night, So bright she has shone
Then just short of 15 years, Our little angel was gone.*

Durham, Ontario

"Up and Coming!"

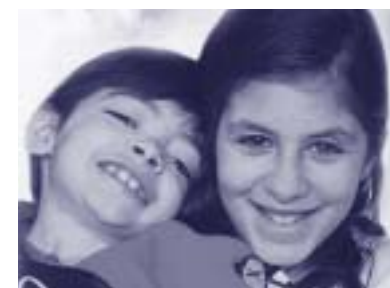
Alex and Laura Vaughan Cross-Country Memorial Bike Ride. Tammy and Alan Vaughan have sponsored two annual memorial golf outings in honor of their children, Alex and Laura, who died from NPD Type C. Not wanting to be left out of the excitement, the children's Aunt, Lynda Reger is beginning to plan a monumental event to help raise awareness and funds for NPD scientific research. In 2005, Lynda and a friend plan to ride their bikes across the United States in memory of Alex and Laura and all those afflicted with NPD, while working to educate towns and communities along their route about NPD and the urgent need for research funding. The NNPDP is working closely with the Vaughan family to choose a route through the U.S. that will match their stops up with other NPD affected families and host communities throughout the course. Watch the NNPDP web site for details! Don't forget your helmets!! Remember ~ Safety First...

Hanover, Massachusetts

Ryan does it again!! On Sunday October 31st, Ryan Kelly, cousin to Erin Roberts NPC, arranged for his fraternity from Bryant College in Smithfield, RI, to sponsor a Halloween Party for Erin and her housemates. Ryan organized the event and all went off without a hitch. Twenty something young men arrived dressed in costumes. These amazing young men purchased gifts for the girls, brought candy, participated in a sing along with Karaoke, but most important was NOT what they purchased, but what they brought. They brought smiles and laughter to four women who are physically and medically compromised. They gave up their Sunday afternoon when they could have been watching football, or sleeping in late, to bring a lot of happiness to four amazing women. Erin sang "Somewhere Over

Vaughan, Ontario

Despite numerous reoccurring illnesses and the frequent hospitalization of their precious son, Luke ~ age 7 NPD Type C, Lucy and Enio Liegghio continue to push ahead with their awareness events and community educational campaigns. The only inspiration the Liegghio's needed was the courage and the smile of their hero and sunshine, Luke. They worked with their state political leader's to obtain a NPD Awareness Month Proclamation for Ontario this past October. In addition, they invited a local newspaper reporter and photographer from The Saturday Sun publication in Toronto to their home to spend time with their super hero "Luke".



Reporter Susan Green and photographer Veronica Henri titled their article, "Little Luke's Fight for Life" and managed to summarize all of the daily chores and challenges that encompass a family living with NPD ~ a child fighting for his life. This article accomplished so much in the area of education and awareness, not to mention the urgent need for additional funding opportunities for continued research.

These efforts capped off another successful "Life for Luke" Dinner Gala, Dance and Silent Auction, which the Liegghio's spearhead annually to benefit NPD Type C Research. The Liegghio family's ongoing efforts are truly inspirational.

Stoney Creek, Ontario

This family event began as a "flicker" of an idea and grew into a "flame" of success for Elisa and Paul Beraldo in honor of their little girl, Taylor ~ NPD Type C. When family members approached the Beraldos with the suggestion of an awareness luncheon in honor of Taylor, no one had an inclining of the events that they were putting in motion. "Touched By Taylor" began as a small luncheon gathering of family and friends and grew into an event attended by over 650 family and friends from the community of Stoney Creek, Ontario. Dr. Marc Patterson was able to travel from New York to speak with the luncheon attendees about the specifics Niemann-Pick Disease and ongoing research into a cure. Overwhelming doesn't quite capture the spirit or level of the support and love that the Beraldo family garnered from this event.

the Rainbow", as well as many of her favorites with her family, friends, and twenty something new faces that she made smile. Earlier this year, Ryan and his gang also hosted a "24-hour Teeter-Totter Marathon" fund raiser for the NNPDP. Thank you Ryan, for once again making a difference in Erin's life and other's affected by NPD. (Editors Note: Family and friends gathered in support of the Roberts family as Erin's health declined in late November. Erin's heartbroken parents notified the foundation that she had died on December 1st from NPD Type C. Our hearts and thoughts go out to her loved ones.)



Cullman, Alabama

Chuck Beckman's mom, Rozetta, has found a unique way to help raise awareness into NPD nationwide. Rozetta hosts an e-bay shopping web site at: www.rozetta treasures.com and has supported a link to the NNPDP web site in honor of her son, Chuck, age 35 NPC. In support of NPD Awareness month, Rozetta has begun to include the foundation's promotional brochure along with an update on her family in all of her packages. We continue to say that creativity is the key and Rozetta has certainly found a new way to raise awareness for the work of the foundation.

Los Angeles, California

Once again, Anthony and Lisa Leoni, along with their extended family and support system hosted a "Spiritual Convergence ~ Pray for Jessica" event via their family web site.

Friends, family and supporters of anyone affected by NPD were encouraged to join in their Community Offering for those affected by Niemann-Pick Type C Disease.

Their web site displayed an ever-expanding journal of the spiritual pledges made for their Spiritual Convergence. The event was specifically held during the month of October to coincide with the Niemann Pick Disease Awareness Month.

Jessica's family worked hard to capture all the various "Acts of Love" on their site. They hope to show how the power of prayer and worship can help those of us affected by this dreaded disorder.



Chatsworth, California

Friends Step In to SCARE CUZ WE CARE! On October 30th, 2004, friends of Tristen, Brian and Stephanie Cortes rallied together to give the ultimate gift. They threw a Halloween Party/Fundraiser on behalf of Tristen and the NNPDP!

In August, Steve Lewis of Chatsworth, CA pumped up a group of his friends to form a committee to put on this party and they all enthusiastically joined in to make it happen. It was a great evening, with incredible haunted decorations, Vegas style roulette, black jack and craps tables, raffles and a whole lotta' shakin' goin' on! Ghosts, goblins and ghouls alike boogied down 'til the wee hours of the night for the best cause Halloween has seen yet!

The Scare Cuz We Care committee has already started thinking about next year and how to continue to make this event bigger and better. A heartfelt thanks goes out from the Cortes family to all of those who donated their time and phenomenal energy to make this happen.



~ ~ ~ ~ Research Highlights ~ ~ ~ ~

"Advancing Knowledge.....Transforming Lives"

Ongoing NNPFD-Funded Research Janet Ward-Pease NNPDF Director of Research

NMDA receptor hypofunction in Niemann-Pick disease Type A

- Stuart A. Lipton, M.D., Ph.D.

Dr. Lipton's lab has found that a compound called sphingosylphosphorylcholine accumulates in Niemann-Pick Type A and that this compound interferes with 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. In cell culture, chemically, he can reverse some, but not all, of this effect. In addition, sphingosylphosphorylcholine seems to influence two additional receptors in the brain as well. Dr. Lipton says "We are currently trying to assess in animal experiments to what quantitative extent the compound is really responsible for the brain dysfunction in NPA, in order to obtain a sufficiently realistic perspective whether there might be a therapeutic potential for targeting the NMDA receptor in NPA."

"The Role of NPC2 (Niemann-Pick Disease Type C2) Protein in Lysosomal Cholesterol Trafficking"

- Heng-Ling Liou, Ph.D.

NNPDF fellow, Dr. Heng-Ling Liou, has now identified several interesting lipids that bind (create a molecular link) to the NPC2 protein. These lipids are lathosterol (a cholesterol precursor), stigmasterol (a plant sterol), and cholesterol sulfate. Also, there appears to be another lipid which may bind to NPC2 but it is not yet identified. The analysis and identification of this lipid is in progress. The significance of these findings is that they add to our understanding of how cholesterol processing is supposed to work in the healthy body. With that knowledge, scientists can understand what goes wrong in NPC disease cells and how that problem might be corrected.

"Cellular and Molecular Mechanisms Underlying Neurodegeneration in Niemann-Pick Type C disease"

- Makoto Michikawa, M.D., Ph.D.

Dr. Michikawa, whose one year grant ended in September, reported his findings that NPC1 mouse brains contain a reduced amount of "synaptic vesicle-related proteins" than normal mouse brains. Per Dr. Michikawa, "...these results suggest that synapse formation and subsequent neuronal network formation are affected in NPC1-/- mouse brains. In addition, we found that the neurons prepared from NPC1-/- brains showed enhanced vulnerability to reagents generating oxidative stress..".

"SSRIs as a potential therapy for NP-C"

- Synthia Mellon, Ph.D.

Dr. Mellon's 6-month status report, submitted in October, explains her activities and findings to date: "Research in our laboratory has

shown that treatment of NP-C mice with the neurosteroid allopregnanolone (ALLO) increases lifespan and delays the onset of the neurologic symptoms typically seen in NP-C mice. ALLO is currently available for research purposes only, and is not available for human use, so immediate clinical trials in patients with NP-C is not possible. To find an additional treatment option for NP-C using an approved medicine, we have been studying the effect of selective serotonin reuptake inhibitors (SSRIs) on increasing the abundance of ALLO in brains. Previous work from several laboratories has suggested that SSRIs such as fluoxetine (Prozac) and paroxetine (Paxil) can increase the abundance of neurosteroids in laboratory rats as well as in human beings treated for depression or premenstrual dysphoric disorder. The main questions we want to answer are: Can SSRIs increase ALLO abundance in brains of wild type mice? Can SSRIs increase ALLO abundance in NP-C mice? At what age of the animal can this occur? and Which SSRIs work best?

We have spent quite a bit of time trying to replicate in mice, the effect of fluoxetine seen previously in rats. We have completed a number of assays in "test tubes" (in vitro) that monitor the conversion of the precursor of ALLO (dihydroprogesterone, DHP), to ALLO, using brain tissues from mice of different ages. So far, we have not seen an effect of fluoxetine or of another SSRI, escitalopram (Lexapro) on the conversion of DHP to ALLO. We have also started an analysis in the animals ("in vivo") to see if we can detect an effect of either of these SSRIs on increasing the content of ALLO in the brain. These studies are ongoing now, and we don't have the answer yet. We have been working exclusively with wild type, control mice, before we start any studies using the NP-C mice, so that we can work out all the conditions on animals that are easily obtained. We have an active NP-C mouse colony at UCSF, so we will be able to do studies using NP-C mice once we are confident in the results in wild type mice."

Clinical Trial Updates

Type B - Enzyme Replacement Therapy

Paul Kaplan, Ph.D., M.B.A., Senior Director of Program Management at Genzyme Corporation says that intensive discussions with regulatory agencies regarding the design of the clinical trial and the clinical development program for ERT are continuing. Based on progress to date, Dr. Kaplan hopes that Genzyme will be able to file an application for clinical trial in early 2005.

Type C ~ Zavesca Clinical Trial Update.

Dr. Marc Patterson, Director of Pediatric Neurology at Columbia University where the U.S. portion of the trial is being conducted, has advised, "The clinical trial of OGT 918 in NPC is now fully enrolled. For the adult patients we anticipate Database lock for the main study period (0 to 12 months) in Q3 2005 and the preliminary results at the end of Q3 2005. For the pediatric part of the study, Database lock for the main study period (0 to 12 months) is due in Q4 2005 with the preliminary results due at the end of Q4 2005. OGT 918 has been generally well tolerated, although some patients in both the UK and the US have withdrawn from the study"

Stacey a True Hero

Members of the extended NPD Foundation were all brokenhearted to learn that Stacey Vorpahl, the daughter of our past Chairperson, Barb Vorpahl, had died on October 9th from NPD Type C. Barb, Gary and Kimberly found themselves wrapped in the arms of love and support, once again, by family, friends and the extended community of Fort Atkinson, the surrounding Jefferson County area and members of the NNPFD. Initially, it was the kind, encouraging and supportive character of this community that brought the NNPFD headquarters to Fort Atkinson, Wisconsin. Following is an editorial, which appeared in the Daily Jefferson County Union shortly after Stacey's death.

We doubt she ever wanted to be a hero, but that is how everyone will remember Stacey Vorpahl.

The 19-year-old Fort Atkinson woman who died Oct. 9 probably would have preferred being known as "that freckled-faced kid" or "the girl with curly red hair" or simply "Stacey," but fate changed her life early on and she became the heroic poster child for a rare and fatal disease.

Stacey was only 18 months old when she was diagnosed with Niemann-Pick Disease-Type C, an illness that causes progressive deterioration of the nervous system by blocking the movement of cholesterol within the body's cells. Cholesterol builds up in the vital organs, causing an enlarged spleen and liver, clumsiness, problems with balance, poor muscle control, impaired eye movements, slurred speech and dementia.

She began showing some symptoms in first grade and started experiencing seizures about age seven. Stacey needed a feeding tube at nine, slowly lost her ability to walk and, by age 10, was confined to a wheelchair. Most recently, her eyesight had dimmer.

Yet, despite the many medications, despite the regimen of physical therapy, Stacey never gave up. Even when she could no longer talk, she retained an infectious smile. Even when her vision began to fail, she kept a twinkle in her eye.



It was that spirit, we think, that took Stacey far past the life expectancy for a Niemann-Pick Type C patient ... that, along with personal attention and care, and the morale and financial support of an entire community.

And it was that spirit, we think, that pushed Barb and Gary Vorpahl to join the families of six other Niemann-Pick children to create a foundation that has raised \$1.9 million to help fund 36 research projects, including more than \$305,000 from Fort Atkinson residents alone. Many strides have been made, including the identification of the gene that causes Niemann-Pick Disease Type C, and most recently, its help in learning about the basic causes and early progress of Alzheimer's and cardiovascular disease.

With only 1,500 people in the entire world having been diagnosed with Niemann-Pick Disease, it would not be a stretch to say that Stacey's infliction and her family and friends' commitment vaulted an otherwise unknown - and unfunded - illness into the public eye.

Yes, Stacey Vorpahl was, indeed, a hero. For nearly two decades, she faced illness and certain death with a joyful smile, courageous heart and hopeful outlook. In turn, Stacey spread that joy, courage and hope to others near and far ... and she continues to do so yet today.

Address and Contact Corrections Requested

One of the strongest tools that we are able to supply to our NPD families is the opportunity to network with other families. Reaching out and sharing their information, knowledge and daily trials with other NPD families in similar circumstances is an extremely helpful support system for all.

Although we work diligently to keep the foundation database current with correct addresses and contact information, we have been known to miss an address change here and there. We would appreciate any address change notifications be made with the remittance envelope included in this newsletter as well. Thank you for your assistance with this matter.

New Funding

No new projects have been funded since our last newsletter, however the NNPFD Board and its Scientific Advisory Board are in the process of reviewing eight grant applications received this month. Funding decisions will be made at the February Board Meeting.



Memorials

Brandon Dominique ~ 22 years
Erin Arline Roberts ~ 24 years
Stacey Lynn Vorpahl ~ 19 years

*Be strong in your resolve...live as fully as you can
Appreciate the people in your life...every woman, child, and man
Fight all obstacles you face, with a maximum degree of fervor
Life is valuable and precious, don't take it for granted.....ever*

*Gone are the needles and tubes. Her medical equipment is still
But.....if we listen quietly and if we have the will
We may bear the distant sounds..... of one new Angel
Tapping, hopping, running and singing above...*

For we know Stacey is free now, and she is sending us her love.

*~ ~ An excerpt from a poem written in memory of
StaceyVorpahl."Stacey's Song" by
Vorpahl family friend Jean Anhalt*

Giving Opportunities

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. We, in turn, depend on those with compassion and the financial resources to make a difference in the lives of those who are suffering. Won't you help in whatever way you can?

For more information or to make a gift, please contact NNPDF's Central Office at (920) 563-0930, or e-mail Nadine Hill, Director at: nnpdf@idcnet.com. Contributions may be mailed to: NNPDF, P.O. Box 310, Fort Atkinson, WI 53538-0310.

*Thank you for
your generosity.*

Partners for a Cure Campaign

Sponsored by "Lee King Buddy Benefit Foundation and Stratos Boats"

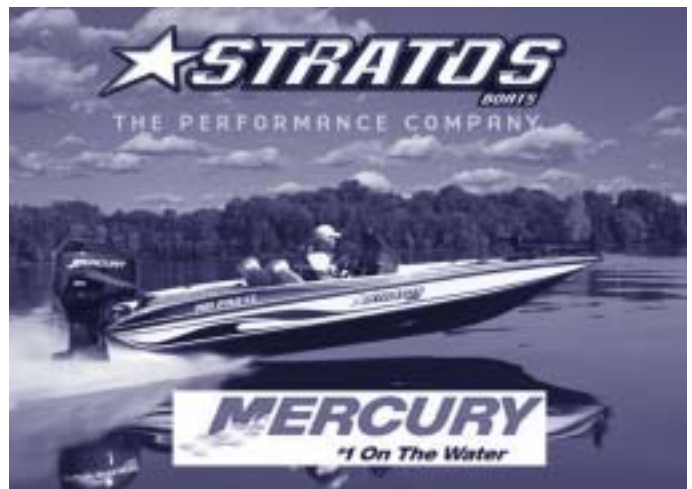
2005 STRATOS 285 PRO XL - 2005 MERCURY XR6 150

Humminbird Electronics - Minn Kota Trolling Motor

Prize Package Is Valued Over \$20,000 -
Rigged and Ready To FISH!!!

Here is your chance to win a top of the line bass boat package AND help fund the finding of a cure for Niemann-Pick Disease that will help children everywhere! Stratos Boats and Mercury Marine have graciously donated a Stratos 285 PRO XL that is powered with Mercury 150 HP XR6 and equipped with Humminbird electronics and a Minn Kota trolling motor to be drawn for and given away at the 2005 Lee King Benefit Bass Tournament. Tickets for the drawing are only \$25 each or buy 5 tickets \$100 and get one FREE!

YOU DO NOT HAVE TO BE PRESENT TO WIN!!! You may purchase tickets by Phone, Mail or ONLINE. By Phone - Call Sam Williams to purchase tickets or more information: (334) 687-6266 or access www.leekingbenefit.org for additional raffle contact information. The duo of Jerry Tice and Al Johnson from Chipley, Florida were the lucky names on the ticket drawn at the Lee King Buddy Benefit Foundation Tournament in 2004 for this beautiful bass rig.



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.



National Niemann-Pick Disease Foundation
P.O. Box 49
415 Madison Avenue
Fort Atkinson, WI 53538

NON-PROFIT
U.S. POSTAGE
PAID
BETHESDA LUTHERAN
HOMES & SERVICES