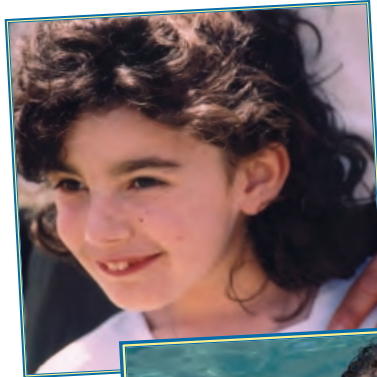




# Niemann-Pick

## SUMMER NEWS 2008

Toll Free 1-877-287-3672



Jessica Leoni ~  
April 2005



Swimming  
with Dad



Jessica  
"The Birthday Girl" ~  
July 6, 2008 ~  
11 years old.

### Thus the journey started...

This year marks the 10 year anniversary of the cruelest April Fools occurrence. We had just closed on the new house. We were doing some maintenance and prepping for our move. I went to the old house to get some dinner when the phone rang. It was the doctor, Jessie's GI doctor to be precise. She asked if we were both available by phone. I explained what was going on. She said it was important that we all be able to speak together. You see, Jessica had been undergoing a medical evaluation. She had an enlarged liver and spleen, was experiencing delays in a number of areas even as a 10 month old and had been diagnosed with cirrhosis only months before. There had been numerous tests, among them a skin biopsy. The results of the biopsy came through, confirming that Jessica had Niemann-Pick Type C.

Lisa had researched quite a bit over the months of testing. The internet provided a lot of information on a lot of the possibilities. She wrestled with what to share with me and what to put aside. She had shown me the printout on NP-C and I chose to put it in a drawer and not deal with it or worry about it. I wasn't in denial; I just didn't see anything productive in cluttering the issues surrounding our daughter's health. (I still feel that way some of the time.)

Now, I had to go to the new house to break the news to Lisa. Jessie was on the floor playing. I leaned over and kissed her on her head and held her for a moment and told her that I love her. I then left.

The new house was only a few minutes away. It was a long drive, though. When I arrived, Lisa was in the lower bedroom, cleaning or painting. I said, "It is 'C'." and she didn't hear me. I said, "The doctor called and it is Niemann Pick Type C." She lashed out in fear and we held each other, wanting to be holding our baby and willing her to good health.

We returned home to take the doctor's call later that evening, starting to strategize and game plan on what we had to do. We have been in a reaction/response mode ever since.

Thus the journey started on a day usually reserved for practical jokes. It was the cruelest of April Fools Days.

Anthony Leoni ~

Dad to Jessica Nicole (NPC ~ age 11 years)

### BARBARA VORPAHL NNPDF Board Chair



If you ask any of the families whose child was diagnosed with Niemann-Pick Disease, like the Leoni family, they can tell you the exact time, the exact date, the exact moment, because life as they knew it had changed in that instant.

Even though our daughter Stacey's diagnosis was over 20 years ago, I can remember the diagnosis as if it were yesterday. We were thrust into a world of the unknown... seizures, feeding tubes, medical specialists, therapists, special equipment. At that time, there was no one knocking at your door saying, "we heard you have a sick child, can we help." We spent countless hours on the phone trying to find anyone who had even heard of this disease.

We depleted every local resource trying to find someone who could

help us with the relentless progression of symptoms our daughter faced as she battled each phase of this disease.

Today, thanks to your continued support we can now say the NNPDF is that "knock on the door".

### WE ARE HERE TO HELP!

In this issue, we highlight the many services now available to families facing a diagnosis of NP. Our hope is that one day a diagnosis of NP will not have such devastating effects.

Until then, what we can do is provide information and emotional support to families so they do not have to face the journey alone.

Barb Vorpahl ~ NNPDF Chair

# "Family Services ~ Activity Updates"

## NEW FAMILY SERVICES DATABASE TO BE IMPLEMENTED

As new families find their way to the NNPfD and as we maintain ongoing contact with existing families, the Foundation Staff is constantly challenged to be sure that everyone is benefiting from all of the resources available through Foundation Programs and Services. To help keep track of what has been sent to each family, and to alert us of possible needs as time passes, we are launching a new computer-based tracking system. This Family Services Database can be run using existing software in the office, and will allow entry

of each item that is sent to families as well as entry of a catalog of available resources. It will also be possible to create a reminder system so that we are prompted to reconnect with families at various times to see what more we can do to be of assistance. While this will help keep the staff on track, remember that you are always welcome to call to request any of the available materials and services, or to simply talk to a supportive and caring staff member!

## FAMILY SERVICES PROGRAM ([http://www.nnpdf.org/familyservices\\_01.html](http://www.nnpdf.org/familyservices_01.html))

We offer assistance in the following areas:

1. Making supportive connections with individuals and families affected by NPD
2. Education: for the family, the health care provider and for school personnel
3. Awareness and advocacy: brochures, videos, media guide, fundraising manual, etc.
4. Referral for medical care, assistance with negotiating through the health care system, and supportive and crisis counseling
5. Others, as needed or requested!

## NNPDF EQUIPMENT EXCHANGE

The NNPfD Equipment Exchange has been launched! Details about how to list an available item and how to request an item can be found at [http://www.nnpdf.org/familyservices\\_09.html](http://www.nnpdf.org/familyservices_09.html).

It is possible to request an item from the list of available supplies, as well as to place an item that you need on our "Wish List." We encourage everyone to occasionally view these lists, as you never know when you might have something in your closets or attic that could be put to use by another member of the NNPfD family, or when you might find something that will help with a new challenge you are facing.

We have created a list of items that you may have at your homes which other families may find of use (please see sidebar).

Many thanks to the Lisa and Jami Chavez Family and the W & A Distribution Company for their gracious support of this program!

### EQUIPMENT EXCHANGE ITEMS

Wheel chair	Protective gear
Stand up walker	IV poles
Walker with seat	Portable Feeding Pump and supplies
Percussive vest airway clearance system	Trach equipment – Concha, compressor, Corrugated EVA Tubing
Hospital bed	Oxygen concentrator and tubing
Computer assist devices (picture keypads, keyboards)	Portable and regular pulse/oxygen machines
Shower adaptations	Diapers/depends, underpads
Assistive Lifts and slings	Suction Machine, canister and catheters
Step lift	
Vehicle ramp	

Please consider donating or requesting any of these items or others as the need arises.

## MEMORIALS

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

Corynne Michelle Bouchard (NPC) ~ 20 months  
 Cathryn Elizabeth Bouchard (NPC) ~ 22 months  
 Damon Eli Cardinal (NPA) ~ 16 months  
 Tamara De Boey (NPC) ~ 30 years  
 Ibrahim Hamdan (NPC) ~ 4 years

Hisn ul Islam (NPC) ~ 4 years  
 Michael Landry (NPC) ~ 25 years  
 Sofia Mezzacapa (NPA) ~ 2 ½ years  
 Elizabeth "Hunter" Ozmer (NPC) ~ 22 years  
 Anthony Paul Sanchez (NPC) ~ 22 years

*Our hearts go out to their families and friends.*

*Time is Merely A  
 Treasure Chest,  
 Keeping Memories Of You  
 Safely In Our Hearts,  
 Until We Can Hold You In  
 Our Arms Once Again.."*

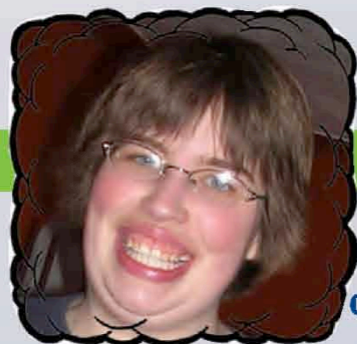
# Making Our Voices Heard

"The NNPDF ~ Where Success Begins with You!"

*It is important to know that words don't move mountains...  
Work, exacting work, moves mountains.*



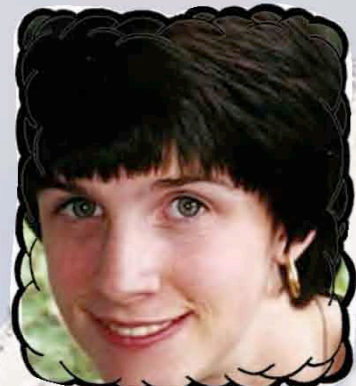
Adam Recke



Callie Traxler



Hisan ul Islam



Erin Roberts

Corynne & Cathryn Bouchard



Steve Daitch atop Mt. Everest in 2008



Jacob Jarrett



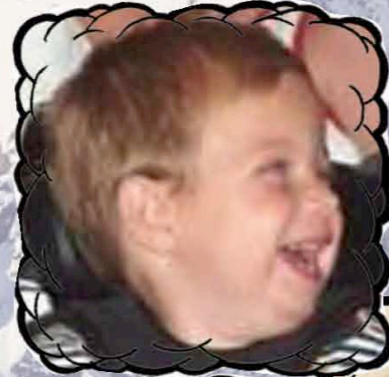
Hunter Ozmer



Graham Kehoe



Braden Smith



Chase Jackson



Naomi Tyrrell



Nathan Archuleta



Bryanna Scribner DeSouza



Brooke Thomas



Damon Cardinal



Addi & Cassi Hempel



Ty Quandt



Brianna & Bailey Patterson



John Taft IV



Stacey Vorpahl



Dillon Papier



Emma Shaffer



Heldamarie Ramos



Breann Chavez



Kevin Eadie



Kathryn Taft



Michelle Trombley



Katlyn Rose Sumbad

We, the families of the children affected by Niemann-Pick Disease thank you for joining us as we persevere in our quest to find a cure.

# Research Highlights

With the help of our Scientific Advisory Board, and under the expert guidance of Janet Ward-Pease, the NNPfD Research Committee is currently hard at work updating our Research Strategic Plan. Although the pace of Niemann-Pick research often seems agonizingly slow, it does keep changing. We need to adapt to these changes when funding research projects to ensure that we are effective in enabling good progress to be made - and in making the best use of our hard-earned research dollars!

In developing our new Research Strategic Plan, we will define where we can best make a positive difference. A clear Foundation research strategy will help those applying for Grants to assess whether their research falls within our financial guidelines. It will also enable the Scientific Advisory Board to give us appropriate advice to ensure that we make good decisions on which projects to fund and will provide a clear focus for the work of the Foundation. Watch this space for developments!

For details about recently completed NNPfD-funded research and ongoing NNPfD-funded research, please see [www.nnpdf.org/](http://www.nnpdf.org/) and select the "Research Update" from the "Research News and Announcements" section.

We are grateful to all of the researchers who work so diligently to better understand the Niemann-Pick diseases and to seek to develop potential therapies for our children.

*Respectfully submitted by Susan Green, Research Committee Chair*

## SCIENTIFIC ADVISORY BOARD MEMBERS

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Emeritus Scientist

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**Yiannis A. Ioannou, Ph.D.**

Mount Sinai School of Medicine

**Jean E. Vance, Ph.D.**

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Dir de Recher Batiment

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Mayo Clinic Rochester, MN

**Steven U. Walkley, DVM, Ph.D.**

Albert Einstein  
College of Medicine

## MEET YOUR NNPfD BOARD AND STAFF



(FROM LEFT TO RIGHT)

**Tammy Vaughan - Secretary and Canadian NNPfD Representative**  
*Mother of Alex and Laura NPC, Procurement Executive, Ontario, Canada*

**Nadine M. Hill - Director of Family Support Services**  
*NNPfD Central Office ~ Fort Atkinson, WI*

**Cate Walsh Vockley, MS, CGC - Coordinator of Education, Referral and Advocacy**  
*Senior Genetic Counselor - Med Genetics Children's Hospital of Pittsburgh, Pittsburgh, PA*

**Carl "Wayne" Moore - Treasurer**  
*Uncle of Andrew and Catie Limer NPC, CPA, Fort Worth, TX*

**Susan Green - Research Committee Chair**  
*Mother of Roy and Murray NPC, Founder of NPfD Group UK, Scotland*

**Barbara Vorpahl - NNPfD Chair**  
*Mother of Stacey NPC, Teacher, Fort Atkinson, Wisconsin*

**Rhonda Brown-Kehoe - Director-at-Large**  
*Mother of Graham NPC, ICU Nurse, Chepachet, Rhode Island*

**Melissa King - Director-at-Large**  
*Mother of Lee NPC, Student/Homemaker, Eufaula, Alabama*

**Holly Roberts - Director-at-Large**  
*Mother of Erin NPC, Travel Agent, Hanover, Maryland*

**Nancy Sullivan - Director-at-Large**  
*Mother of Karen NPC, Nurse, St. Louis, Missouri*

**Karen Quandt - Director-at-Large**  
*Mother of Ty NPC, Nurse, Edgewood, Washington*

(NOT PICTURED)  
**Doug Pease - Vice Chair**  
*Uncle of Adam and Amanda Ward NPC, Computer Consultant, New Orleans, Louisiana*

**Hunt Ozmer - Director-at-Large**  
*Father of Hunter NPC, Sales Representative, Roanoke, Virginia*

As you can see, most members of our board have experienced first hand the devastating effects of a diagnosis of Niemann-Pick Disease. They are a dedicated group of individuals who have reached past their own pain and struggles to help others facing a diagnosis of NPD.

This picture was taken at the 2008 Annual Board Meeting in February where the Board met to set clear direction, focus and new initiatives for our programs of Research and Family Support.

We would like to offer a special welcome to our newest Board Members Karen Quandt and Susan Green.

Thanks to your support and the fundraising efforts of NPD families, 2007 was a "banner year" for the NNPfD. We continue to forge ahead with a commitment to making 2008 an even stronger year.

## NNPDF OFFICE CONTACT INFORMATION:

NADINE M. HILL,  
Director of Family Services  
E-mail: [nhill@nnpdf.org](mailto:nhill@nnpdf.org)

PHONE: 1-920-563-0930  
TOLL FREE: 1-877-287-3672  
401 Madison Avenue, Suite B  
P.O. Box 49  
Fort Atkinson, WI 53538-0049  
Web site: [www.nnpdf.org](http://www.nnpdf.org)

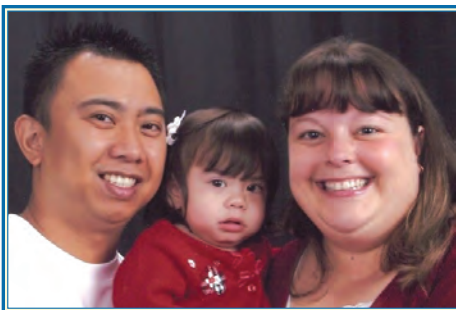
CATE WALSH-VOCKLEY,  
Coordinator of Education,  
Referral, and Advocacy  
E-mail: [Catherine.walshvockley@chp.edu](mailto:Catherine.walshvockley@chp.edu)  
Phone: 412-692-7349  
Children's Hospital of Pittsburgh ~  
Medical Genetics

CONTRIBUTIONS & MEMBERSHIPS TO:  
NNPDF, Inc.  
Post Office Box 310  
Fort Atkinson, WI 53538-0310

NNPDF BOARD OF DIRECTORS:  
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Susan Green Holly Roberts  
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Hunt Ozmer

## 2007 ~ 2008 FUND RAISER CONTEST WINNERS



Meet our "First Time"  
Fund Raiser Contest  
Winners for 2007 ~ 2008!!

The Genesis and Jennifer  
Sumbad Family,  
in honor of their baby  
daughter Katlyn ~  
NPD Type B.

The Sumbad family let little time pass between receiving the diagnosis of NPB for Katlyn and starting their efforts to host a fund raising event to support continued research into NPD. Shortly after learning of their baby's diagnosis the family made plans to host a local golf outing and to attend the 15th Annual NNPFD Family Support and Medical Conference in Milwaukee, Wisconsin.

After the conference they returned home to Modesto, California, gathered their family and friends and began work in earnest on their 1st fund raising

event. On September 8, 2007 family and friends took part in a golf outing, silent auction and dinner ~ AND ~ their efforts paid off!

As this year's winners they have won complimentary deluxe accommodations at the Georgian Terrace in Atlanta, Georgia during this year's family conference... not to be outdone... the Sumbad family have already scheduled a follow-up golf outing to be held on October 4th, 2008.

The NNPFD hosts this contest annually. Please contact the foundation offices for more information.

# NNPDF 16<sup>th</sup> Annual Family Support & Medical Conference

was held in Atlanta, Georgia  
July 31<sup>st</sup> thru August 3<sup>rd</sup>



2008 Family Conference Hosts:  
**Melissa and Jimmy King**  
in honor of their son Lee (NPC)

Refer to the NNPFD website  
for more details

CHECK OUT OUR NEW WEBSITE



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

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.

National Niemann-Pick Disease Foundation  
P.O. Box 49  
Fort Atkinson, WI 53538-0049

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