

In commemoration of this year's 10<sup>th</sup> Annual October Niemann-Pick Disease Awareness Month, families across the United States and Canada are working in their communities to raise awareness and funds in support of the work of the National Niemann-Pick Disease Foundation. Some have asked their state's governor or their city's mayor to proclaim October as NPD Awareness Month, some are scheduling elaborate fundraising events, and some are planning simple awareness projects.

**As a Foundation we have found that awareness drives donations, donations drive research, and it is this essential research that will ultimately lead to effective treatments and a cure for Niemann-Pick Disease!**

In this mailing, we are highlighting two young adults who are valiantly fighting the battle against the ravages of Niemann-Pick Disease Type C. It is through their courage and perseverance that family and friends garner strength and hope to journey on until a cure is found. Please consider what you can do to help raise awareness and funding during October Niemann-Pick Disease Awareness Month!

### *The Promise of Youth!*

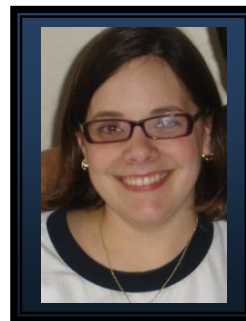
Oh, to be 21 again! Out of high school, a “real” adult, so many options in front of you, the world as your oyster!

Plans to lay...decisions to make....furthering your education, finding a job, striving to make your mark in today's world...

Sadly, for an unfortunate few, the hopes, dreams and challenges of a life unfolding are overshadowed by creeping problems that at first seem strange and annoying, but as difficulties mount and multiply, become truly frightening.

Put yourself in the shoes of Mitchell Brubaker, age 22, of California, or those of 32-year-old Kelly Thompson of Connecticut. Ripe with youth's potential, you are driving happily full-speed ahead, preparing for and creating the life of which you've always dreamed.

Then, a few seemingly inexplicable problems show up. You begin having difficulty moving your eyes up and down. Your gait becomes unsteady. You stumble and fall for no apparent reason. You have difficulty enunciating words properly. Your short term memory begins to falter.



## Life Derailed

You wonder what is happening to you, and why. You try to shake it off and go on with your life, attending school, socializing with friends, thinking about a future, a career, perhaps a family and a home of your own. Unfortunately, ignoring your nagging difficulties does not make them go away, and in fact, more symptoms surface while existing issues worsen.

Mitchell and Kelly, and others like them, have experienced this nightmare firsthand. They struggled with growing lists of physical and neurological symptoms, along with the anxiety and fear of what was happening to them, until finally they received the dreaded diagnosis of Niemann-Pick Disease Type C (NPC).

## Niemann-Pick Disease Type C (NPC)

NPC is a terminal neurodegenerative disease with no known cure. Though highly variable in its onset and progression, typically, NPC makes its presence known during childhood.

However, a growing number of young adults, like Mitch and Kelly, are being diagnosed with NPC. Mitchell was diagnosed with NPC at age 19 and Kelly at age 25.

## Mitchell's Story

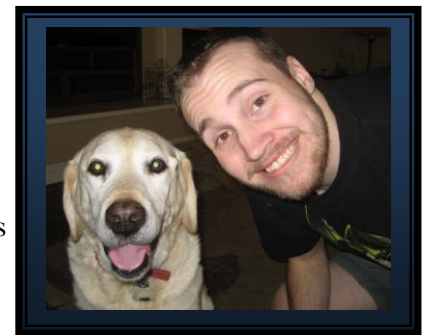
A very attractive young man, Mitchell once had dreams of becoming an actor or working as a voice-over actor, but due to NPC, his speech is now slurred and labored. In retrospect, Mitchell's parents, Terri and Mark Brubaker, report that Mitch first started showing a few symptoms around age 13. (Early symptoms of NPC may first be diagnosed as a learning disability, "clumsiness," delayed motor skill development, etc.)

Now, due to NPC, Mitch's short term memory, cognitive functions and motivation have all declined. Mitchell has an infectious laugh, but due to NPC-related cataplexy, he tries not to laugh too much anymore. (Cataplexy is an abrupt loss of muscle tone, often brought on by sudden emotion, causing a person to collapse to the floor.)

Today, unable to attend school or hold a job, Mitch spends his days watching tv or reading rather than socializing with friends whose lives have moved on.

Niemann-Pick Disease can take another form called Acid Sphingomyelinase Deficiency (ASMD), or Type A or Type B, depending on the severity. Children with Type A usually die by age 3 or 4, while individuals with Type B may live fairly full lives, barring complications.

The NNPfD works to raise funds for research into all types of Niemann-Pick Disease. For more information, visit [www.nnpdf.org](http://www.nnpdf.org)



*Mitchell and his pal, Riley*



*Kelly and her "Running4Kelly" Marathon Team*

## Kelly's Story

Through childhood, Kelly maintained above-average academic standing and participated in soccer, softball and cheerleading. Known for her boundless energy and positive outlook, Kelly graduated high school and enrolled in Eastern Connecticut State University.

During college Kelly's motor skills, speech and swallowing abilities declined noticeably. Despite her struggles, Kelly worked hard to compensate for what was believed to be an underlying learning disability, and she graduated in 2002.

NPC's assault on neurological function has cruelly robbed Kelly of her love of reading and her ability to concentrate. Kelly now attends an adult day-center where she helps out with daily activities. While Kelly still brightens the day of everyone she meets, this is not the life anyone would have envisioned for her ten years ago.

## **The Heavy Toll of Niemann-Pick Disease**

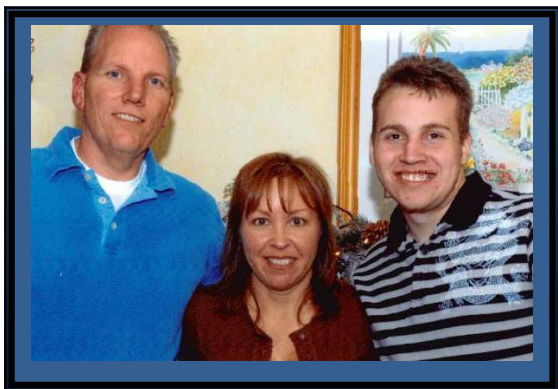
Niemann-Pick Disease Type C is a rare genetic lysosomal storage disease. It strikes through accumulation of excess fats in the cells of the liver, spleen and brain. When brain cell function is blocked, one loses coordination, stumbles, falls, and eventually needs a wheelchair, hospital bed and other adaptive equipment.

As the disease worsens, other devastating symptoms develop, including loss of the ability to speak and swallow, and seizures may occur. The health of one with NPC declines until the disease claims another life.

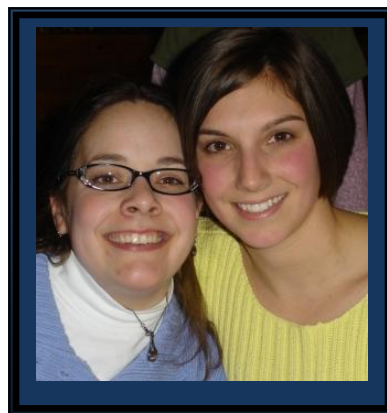
**Without a treatment or cure, NPC will inevitably rob Mitch and Kelly of all their abilities, and ultimately, their lives.**

## **Our Quest for a Cure**

NPC is the cruel killer that has dramatically altered the course of these two promising lives. NPC is the cruel killer that will take the life of Mark and Terri Brubaker's only child, Mitchell, unless we find a treatment or a cure. NPC is the cruel killer that will rob Becky McGuire of Kelly, her beloved cousin who she views more as a sister, unless we beat this terrible disease first.



*Mark and Terri Brubaker with their son, Mitchell*



*Kelly and her "sister" (cousin) Becky McGuire*

**NPC is the cruel killer that will continue to devastate families UNTIL WE FIND EFFECTIVE THERAPIES AND A CURE.**

Finding those therapies and a cure is the mission of the National Niemann-Pick Disease Foundation (NNPDF) and our member families -- families like those of Mitchell and Kelly -- who are working hard in their communities to raise awareness and funding for research.

In the year following Mitchell's diagnosis, his family and friends rallied to raise \$8,000 for research. Several of Kelly's family and friends recently ran a marathon and raised over \$10,000. In the NNPDF's 19-year existence, families like these have helped to raise almost \$5 million dollars for NPC research!

## **Research Progress**

The good news is that research is making progress! The gene responsible for Niemann-Pick Disease has been identified and an experimental drug shows some promise for some NPC patients. The NNPDF is currently funding six studies by researchers who are working diligently to unlock the mysteries of NPC.

We are more hopeful than ever in this Quest for a Cure, but time is fleeting. We must PERSEVERE and drive research forward to save precious lives such as Kelly's and Mitchell's, and so many more around the country and into the future.

**Please, will you help us, and families like the Brubakers and Thompsons, find effective treatments and a cure for Niemann-Pick Disease?**

**Your Donation Will Help!**

Please look into your heart and consider making a gift in honor of Mitch and Kelly and their battle against NPD.

Your donation will make a difference and give hope to the families of all who struggle with this horrific disease. Even modest donations, when added together, drive research forward and hasten the date when we can celebrate victory over Niemann-Pick Disease!

In these difficult economic times, we want you to know that we are doing everything in our power to make your gift go farther and do more. You can be assured your gift to the NNPFD will be greatly appreciated and faithfully stewarded to bring the greatest benefit to families impacted by Niemann-Pick Disease.

Please don't hesitate to contact us if you have any questions or if we can provide further information about the National Niemann-Pick Disease Foundation, or the research we support.

**Thank you for your support!**

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

**The important work of the National Niemann-Pick Disease Foundation includes:**

- Family Services such as informational packets, referrals to medical professionals, telephone consultation, brochures, newsletters, a comprehensive Web site, listserv groups, updates on research and news, etc.
- The Annual NNPFD Family Support and Medical Conference where families affected by NPD can hear the latest news and research directly from the experts, and make connections with other families who also face the challenges of Niemann-Pick Disease and who truly understand and care.
- A total of nearly \$5 million dedicated to essential Niemann-Pick Disease research since the foundation's inception in 1992.
- Fundraising and awareness-raising support for families in their communities, including assistance with handout materials, press releases, posters, requests for official proclamations, "best practices," etc.
- Information and support for medical professionals, educators, caregivers, social workers, etc., who may be encountering Niemann-Pick Disease for the first time in a client.
- And much, much more! Please visit [www.nnpdf.org](http://www.nnpdf.org) for more information about Niemann-Pick Disease and the NNPFD.

**About the National Niemann-Pick Disease Foundation (NNPFD)**

The NNPFD, established in 1992, is an international, voluntary, non-profit organization comprised of parents, relatives and friends committed to finding a cure for Niemann-Pick Disease. For more information about the NNPFD and Niemann-Pick Disease of all types, please visit [www.nnpdf.org](http://www.nnpdf.org).

The NNPFD is a 501(c)(3) corporation and your gift is fully tax-deductible. We also work with businesses, employers and community organizations to verify donations for qualified matching funds. Please contact us for information about making a planned gift or bequest to the NNPFD.

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