

NF NEWS

NEUROFIBROMATOSIS NORTHEAST

Promoting Research, Awareness and Advocacy since 1988

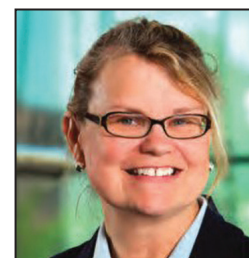
Volume 35, No. 2

Winter 2023

Happy Winter!

As we look forward to another prosperous year of fundraising, education, and therapies for NF, the word IMPACT stands out for me. What will my IMPACT on NF be in 2023? While 2022 is a celebration of so many accomplishments for NF Northeast, we take this opportunity to reflect on where else we can make a difference in the NF community.

From raising funds through our many NF Northeast events, including walking, running, hiking, and golfing, to celebrating wins playing the many games at Committed to Curing NF, we continue to move the needle forward, advancing research through the NF investigators we fund. We are so pleased to expand our research opportunities to encourage more applicants studying NF. Our refreshed website allows investigators to apply online and get more detailed information about what NF Northeast currently funds.



*Sharon Klein
Executive Director*

In the advocacy world, we are thrilled to announce that, thanks to your ongoing efforts, President Biden signed the bill to secure \$25 million for NF research in late December. This is a \$5-million increase over previous funding levels from the Congressionally Directed Medical Research Programs' (CDMRP) Neurofibromatosis Research Program. And we just attended our Advocacy Days event on Capitol Hill in person for the first time in 3 years! Volunteer advocates from across our country came together to urge their senators and representatives to support NF research funding through CDMRP and the National Institutes of Health (NIH). We once again asked for funding to support critical NF research. The advocacy program has generated over \$500 million dedicated to NF research; this would not have happened without the voice of our advocates. Thank you!

Please read on—this newsletter is full of great information, celebrating milestones and looking ahead to what's possible in the world of NF. And please take a minute to think about what your IMPACT will be for NF patients in 2023!

Best,

A handwritten signature in black ink that reads "Sharon Klein". The signature is fluid and cursive, written in a professional style.

Sharon Klein
Executive Director
NF Northeast

Our Vision: A world where the burden of neurofibromatosis does not exist.

Our Mission: To bring hope to those affected by neurofibromatosis and allied disorders.

Patients and families are at the heart of NF Northeast's mission. We provide advocacy, support, and awareness, while promoting research and enhanced clinical care.

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Dear NF Community,

*“HOPE sees the invisible, feels the intangible,
 and achieves the impossible.”*

Katie Doucette

As we move into 2023, NF Northeast’s mission to bring hope to those affected by neurofibromatosis and schwannomatosis is even more important.

I am honored to have been on the NF Northeast Board of Directors for 16 years—many of those years as the Board Secretary and Board Chair. Having led the organization through a lot of change, particularly the COVID-19 pandemic and the transition to a new Executive Director, I am grateful for our impressive Board, Karen Peluso, and NF Northeast founders, as well as our committed staff, volunteers, and supporters over the years, who have collectively made the organization such an important advocate, resource, and place of hope for the NF community.

I am excited that John Manth, a longstanding NF Northeast Board and community member, was elected the next Board Chair. John will be a passionate advocate and leader of NF Northeast. I also want to recognize and thank Bob Ryan, who has transitioned off the Board as Treasurer after 15 years, for his efforts and dedication. We are very fortunate to have Andrés Lessing move into the Treasurer role.

My family and I are grateful to have been part of this incredible community for so many years. To be involved in the expansion of the organization geographically and in the valuable programs and services that NF Northeast provides is heartwarming. Additionally, it has been very promising to see the progress of clinical care and research in the NF field.

Of course, I will remain actively involved in NF Northeast and hope to see you at some upcoming events!

Wishing you all a Happy New Year!

Sincerely,

Lori Ryan



*Lori Ryan
 RN, MS, Chair Emerita*



Business Friends Corner

Eryn (aka Erin Dawson) passed away in 2016 due to a brain tumor as a result of NF. As a way to celebrate Eryn’s life, Trish Mahoney-Diefenbach honors her daughter’s legacy through her work as a real estate agent. She donates 10% of her commissions to NF Northeast. If you would like information on how to become a part of Eryn’s Angels, contact Trish directly.



Trish Mahoney-Diefenbach
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 500 Cummings Center, Suite 1550
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 Email: trish@dalbon.com

From Our Board Chair...



*John Manth,
Board Chair*

For those who may not know me, my name is John Manth. My connection to NF Northeast (NFNE) began with a phone call 16 years ago when our 7-year-old daughter, Leah, had just been diagnosed with NF2. My wife Linda and I were searching the Internet for information, and NF Northeast Founder and then Executive Director Karen Peluso answered our call. From that moment, our lives changed. Karen listened, but also provided hope and a path forward for us. Her compassionate words and the mission of NFNE have stuck with us and guided us ever since. We have dedicated ourselves to helping families dealing with NF by working to find meaningful treatments, spreading awareness, and advocating for federal research funding.

I am extremely honored to be named Chair of the Board of Directors. This is the first time a Chairman comes from outside the Boston area; my family and I live in Buffalo, NY. I want to thank Lori Ryan for her many years of dedication to the Board. She is a tireless volunteer leader for NFNE, and it has been an honor serving with and learning from her. Lori led this organization during the pandemic, the Executive Director search process, as well as the transition period since Sharon Klein has filled that role.

I have three goals for my tenure as Chair.

- First, continued outreach from NFNE to NF patients and families as well as NF clinics in the 9-state region that we serve;
- Second, build relationships with our network of patients and families, and engage them more in the organization; I would like us to involve more volunteers in serving on committees, planning new events, and participating in our current events;
- Lastly, work with Sharon and the Board to expand our team of paid employees, as we are still rebuilding since the pandemic. It is imperative that we assemble the best team possible and bring in additional employees who have expertise and experience in the nonprofit world.

I plan to update you on the progress of these goals in subsequent newsletters and emails. In the meantime, please feel free to contact me with any questions, concerns, or to just chat. My cell is (716) 930-4585 and my email is jmanth7028@aol.com.

Sincerely,
John Manth

NF Northeast Welcomes New Board Members



Marybeth Shaw has been an avid supporter of NF Northeast since 2007 when her then 7-year-old niece was diagnosed with NF2. She has been an active member of the Run4Leah/CureNF2 Team, participating in the Buffalo Marathon, raising funds for NF2

research, and creating awareness. A long-time patient advocate on Capitol Hill, Marybeth now serves on the NF Network Advocacy Committee as a mentor to new advocates. Marybeth holds a bachelor's degree from Boston College and is retired from the consumer products testing and inspection industry where she spent over 20 years helping manufacturers and retailers comply with U.S. and international consumer product safety regulations.



Ben Buttrick, Managing Director, Global Client Relationship Management for Cambridge Associates, LLC, has been affiliated with NF Northeast for close to 10 years as a Coast to the Cure cycle committee member and rider.

In addition to his volunteer work with NF Northeast, Ben is a member of the CFA Institute and the Boston Security Analyst Society, and serves on the Town of Essex Finance Committee. He received a bachelor's degree from Boston University. Ben is an avid runner and cyclist, and has a strong interest in music—giving occasional piano and vocal performances.

A Personal Reflection

By Stephanie-Kaye Powers

Imagine, if you will, all the passions and skills that you have honed and learned to love from the time you were a child—triumphs, pride, confidence, personality traits—developed on your way to learning about and becoming your true self: the lover of long-distance running, track and field, figure skating at an earlier age, beloved days at the rink, pond hockey and mastering crossovers, rollerblading, skiing, dare devil, dancer, ballerina, first one in the ocean, try anything, beat you to the top of the hill? Dare me to.

And now imagine all of that slowly being taken away bit by bit and, in its place, a new scar, more complications, another loss, and fragments of yourself scattered on the floor. Imagine being left with only one choice—to pick up whatever's left, reconstruct yourself, and hope that the next hit doesn't take more (but it always does). You keep trying to live a semblance of a "normal" existence for as long as you possibly can, building and maintaining relationships, fulfilling lifelong dreams, dreams common to most people your age—fall in love, start a family—the ability to achieve them taken for granted. Now your dreams must constantly change.

You did not choose this change. You try to push away the overwhelming feeling that somehow you are being punished for something you cannot control, but still this incredibly cruel punishment tests you. Although you're immensely grateful for the ridiculously strong support of family and friends, who stick by you day in and day out, time after time, you terribly and heartbreakingly miss the things and people you loved but, in the end, lost because it was all just too much.

The stress from NF2 complications and the weight of emotional and mental strains caused by having this disease—one that does not have a cure, one that will continue inflicting "loss" until one is found—all takes too much of a toll. For a moment you feel that you might obtain

your dreams after all. But again, the NF2 hits keep coming and are too big. Your dreams have to change, emotions and mentality change, and more scars form. But this time there are no fragments, just dust, and the aspirations that held you together and helped you pick up the pieces over and over are now gone.

So, you just go on, trying not to feel completely destitute, and you set new goals: travel, work life, higher education, independent living, and, yes, even adopting a dog—appreciating those who support you—until NF2 hits you with a new series of tidal waves. Many of those new goals that you set and achieved must be deleted from yet another new life that NF2 forced you to create. Independent living gone. Work life gone. Travel opportunities gone. Walks and trips to the park with your dog gone. That car you wanted and eventually obtained gone—Because of NF2.



Stephanie, left, at Stonehenge with friends Samantha and Jessica in 1999 studying abroad.

Each time you think, "okay this was the worst," and pick yourself up, again, you are pummeled... another hit. Another potential surgery, drug trial, scan, test, scar. Another piece of whatever is left of you, taken away... and then another, all while struggling with pain and aftereffects, crippling NF2 complications every single day. That is living with NF2. Relentless hits. Constantly getting back up. Realizing this is something you always must do. Until there is a cure. There needs to be a cure.

Stephanie was diagnosed with NF2 during the spring of 2000, when she was a junior in college. After spending the

fall semester studying in London and traveling throughout Europe, she began experiencing pain in the lower lumbar spine. Taking into consideration her family history with NF2, Stephanie made an appointment with Dr. Mia McCullin at the NF clinic at MGH. An MRI and a blood test confirmed NF2. Her first spinal surgery took place during the summer.

Stephanie returned to college and completed her degree, graduating in the spring of 2001. She was very fortunate to experience a pain-free senior year and participate in all of the "normal" activities, such as spring break and graduation. She began her teaching career in the fall, but by the spring was suffering from severe headaches and vision problems, which led to a hydrocephalus diagnosis and, in turn, a VP shunt placement, followed by several surgical procedures to redirect the shunt.

After returning to teaching, Stephanie began experiencing significant decline in the hearing in her right ear, and a decision was made to remove as much of the acoustic neuroma on that side as possible. Despite the efforts at MGH, she lost all hearing in her right ear, however, was fortunate to eventually regain full facial functions.

Stephanie continues to undergo routine and otherwise not so routine MRI's, CT scans, hearing tests, and eye exams. She has lost track of the number and timeline of surgical procedures and other treatments over the past 20+ years since her initial diagnosis. So much in her life has changed since then, but NF2 has remained the same—ever present, unpredictable, and without a cure or end in sight. Using a walker 24/7 has become her reality with NF.

Stephanie is so grateful to her amazing support system—family, friends, the medical and administrative staff who have dedicated their careers to finding cures for NF. Recently, her college friends participated in Hike for NF to raise funds for NF Northeast. They climbed Mt. Monadnock in New Hampshire, where they had hiked as seniors.

Message from the Friends Who Hiked

Samantha Galli, Kristen McLaughlin, Jessica VanDenToorn, Karen Riggio

We met in 1997 as freshmen at Franklin Pierce College in Rindge, New Hampshire, all five of us searching for our place in this new adventure. As we got to know each other over the next four years, our lives as we knew them would forever change.

The “Condo 5 Cuties” were born and so many incredible memories made. The unwavering bond and friendship we shared would continue after college to this very day. Five unique women with completely different lives still finding a way to come together to share their experiences and nurture their friendship. We still meet up every few months, especially around the holidays, to visit and relive those fantastic, mostly hysterical, memories, all while creating so many more. We appreciate the uniqueness of this friendship and never take it for granted, supporting each other through life’s ups and downs, and cherishing the moments we have together.

During our junior year at Franklin Pierce, we learned of Steph’s battle with NF2. At the time, her diagnosis didn’t hold her back in any way. In fact, most of our friends didn’t realize that she was silently fighting this disease at all.

Following college, we supported Steph as she fought back from countless brain and spinal surgeries to remove her “gremlins,” as she affectionately refers to them. Even after the most intrusive of surgeries, she never let NF2 be the focus. She regained her strength, and she was always there and always the Stephanie we know and love.

We will forever stand by our dearest friend Stephanie through her battle with NF and would move mountains for her, if we could. Instead, this past fall we did what we could do, climb a mountain in her honor—Mt. Monadnock—the very mountain we all climbed together our senior year of college. We carried her with us in our hearts, and, as we reached the top, we reveled in the beauty and reflected on her strength and resilience.

The climb we completed was the one Stephanie makes daily. She is one of the strongest women we know, and we are so very proud to call her our friend.



Top of Mt. Monadnock senior year at Franklin Pierce, 2001



Stephanie (left) with her friends in 2017



Samantha, Kristen, Jessica, and Karen about to hike Pack Monadnock in the Mt. Monadnock Region of New Hampshire, in honor of their great friend Steph!

Raising NF Awareness through Geocaching!

Joseph “Joe” Brayman, from Oneonta, NY, started geocaching back in June 2006 and, since that time, has found 6,745 caches. Geocaching is a global treasure hunt in which people look for hidden objects using GPS devices and online clues.

Joe, who has neurofibromatosis type 1, is an avid hiker and often geocaches during his hikes. “At one point I had 11 active caches but am now down to 6, as I lost some of my stats for this year since getting sick,” he explains. “It is a fun game to play and a fun way to raise awareness for NF at the same time.” One of Joe’s trackables is an NF Northeast blue bracelet! Whoever finds it has an opportunity to learn about NF and then gets to hide the bracelet in another location for someone else to find as part of geocaching.

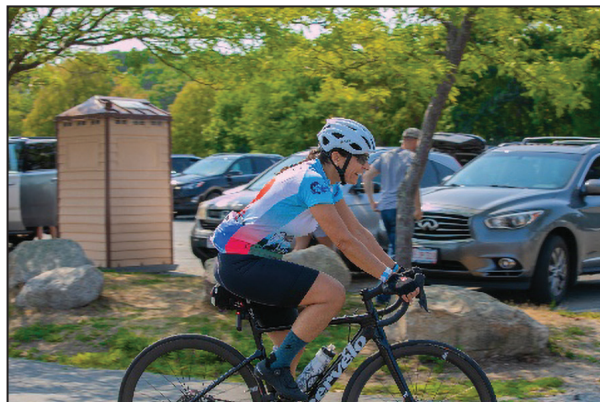
You can follow the bracelet’s travels by going to www.geocaching.com. Create a free account, select “Trackables” from the “Play” menu, and enter the tracking code: Tb8jnn. Be sure to post a comment and a picture of where you found it!

NF Bracelet stats to date:

- First cached February 19, 2019, in Verona, NY
- Miles traveled throughout the U.S., Brazil, and Canada: 11,747
- Currently in Rochester, NY



Celebrating 2022's Fantastic Fall Season of Events!



11th Annual Coast to the Cure
September 10 • Stage Fort Park, Gloucester, MA

Special thanks to Alexion Pharmaceuticals, Mission Sponsor



23rd Annual Fall Golf Classic
September 19
Four Oaks Country Club, Dracut, MA



Committed to Curing NF

November 12
Anthony's of Malden, Malden, MA

Congratulations to Melissa and Mike Malerba whose event raised nearly \$90,000 for NF Northeast. Guests enjoyed an Italian buffet while visiting multiple raffle tables, casino games, and even a photo opportunity with Santa!



Philadelphia "Philly" Marathon

November 18-19

Runners participated in the Philadelphia Marathon Weekend. Special thanks to Coach Scot Dedeo (far right) and Mentor Beth Hanlon (third from left) for their hard work and dedication.



Cynthia Cassandro, Alexion Pharmaceuticals; Dr. Frank Buono, PhD; and Cam Bean, after walking the stadium at Dunkin Donuts Park.

Steps2Cure NF Hartford

October 2 • Dunkin Donuts Park, Hartford, CT

Special thanks to event Mission Sponsor Alexion Pharmaceuticals and Silver Sponsor SpringWorks



36th Annual Golf Tournament to benefit NF Northeast

October 3
Worcester Country Club, Worcester, MA

Presenting Sponsor Grand Circle Travel



Hike to Cure NF

September 24

Hikers climbed Pack Monadnock in the Mt. Monadnock region of NH.

Special thanks to REI, our hike guide partner.



From the Director of Patient and Clinic Outreach...

By Anne Patterson

Supporting NF patients and providing hope and resources is at the heart of NF Northeast. Our **Matched for Hope Program** is designed to confidentially pair a patient/parent with another NF patient whose life is also affected by NF1, NF2, or schwannomatosis. Please email Anne Patterson at apatterson@nfnortheast.org if you would like to learn more about this program. For a more casual community-building gathering, join us for our monthly **NF Virtual Chat!** These chats are fantastic opportunities to meet others touched by NF and to have genuine conversations about day-to-day concerns and questions that may arise. The registration link is located on our website www.nfnortheast.org under “Events Calendar,” and on the Neurofibromatosis Northeast Facebook events page.

Do you have a child (ages 5-16) who could benefit from the social interaction and empowerment of connecting with a collegiate athletic team? NF Northeast would love to refer your family to **Team IMPACT**. Team IMPACT is a nonprofit organization that matches children facing serious illness and disability with college sports teams, **creating a long-term, life-changing experience** for everyone involved! There are colleges in the northeastern U.S. currently looking for children to join their teams. Please email Anne Patterson at apatterson@nfnortheast.org for more information.

NF Northeast is excited to announce **Tuesday Take Aways**, a four-part lunch and learn series beginning in February! These one-hour lunchtime sessions brief participants about updates and information relevant to our NF community. Please join us at any or all of the following sessions at 12:00 noon (ET). Registration links can be found on the NFNE website under “Events Calendar.”

- **February 28:** *Navigating clinicaltrials.gov and the Network Edge*—Vanessa Merker, PhD
- **March 28:** *Basic Science Investigations of NF1 Tumors*—Rebecca Dodd, PhD
- **April 25:** *Common Sleep Disturbances in Patients Affected by NF*—James Walker, PhD
- **May 30:** *Pain in Neurofibromatosis (NF), NF2-related Schwannomatosis (SWN), and SWN*—Frank Buono, PhD, and Dale Berg, patient advocate

There are many new developments in the medical and science community for neurofibromatosis research/studies. Here are some research studies that NF Northeast has recently shared*:

NF2 Study—*Doxycycline Injection of Cutaneous Schwannoma in Neurofibromatosis Type 2*, Dr. Brad Welling. To learn more about this study, please visit: <https://www.nfnortheast.org/wp-content/uploads/2022/11/NFNE-Welling-Research.pdf>

NF1 Family Planning Survey—*Survey of Family Planning Perspectives and Practices of Adults with Neurofibromatosis Type 1*, the NYU Langone Health Comprehensive Neurofibromatosis Center.

For more information, please call (212) 263-7744 or email apatterson@nfnortheast.org.

NF2 Recursion Study—*Efficacy and Safety of REC-2282 in Patients with Progressive Neurofibromatosis Type 2 (NF2) Mutated Meningiomas (POPLAR-NF2)*. If you are 12 years of age or older and are interested in participating, please visit clinicaltrials.gov or poplarnf2.com.

**Neurofibromatosis Northeast (NFNE) provides research notices as an information service to its community. NFNE does not conduct these studies. This information does not represent an endorsement by NFNE or its members, but rather makes you aware that clinical trials are available for your participation, if you so choose. You should discuss studies with your medical team before altering your current treatment program. NFNE is not responsible for any adverse outcomes.*

Moving the Needle Forward in Research

The Medical and Scientific Committee is thrilled to announce the current NF Northeast funded projects.

D. Bradley Welling, MD, PhD, FACS Massachusetts Eye & Ear Infirmary

Doxycycline Injection of Cutaneous Schwannoma in Neurofibromatosis Type 2. Dr. Welling is examining the effect of direct subcutaneous tumor injection with doxycycline, an antibiotic known to cause regression of other tumors and malformations.

Vanessa Merker, PhD Massachusetts General Hospital The Family Center for Neurofibromatosis

Expanding Patient-Centered Outcome Assessment in an NF Consortium Trial of Selumetinib Plus Cabozantinib for Growing or Symptomatic Plexiform Neurofibromas: A Qualitative Interview Study. The Food and Drug Administration (FDA) has issued guidance on collecting interview data in trials, and will even consider this kind of qualitative (non-numeric) data when it decides whether or not to approve new drugs.

**Bonnie Klein Tasman, PhD
University of Wisconsin, Milwaukee
Child Neurodevelopment Research Labs
In Partnership with NF Midwest, NF Upper Midwest, and NF North Central, University of Wisconsin, Milwaukee**
Effectiveness of a Telehealth Group for Improving Peer Relationships for Adolescents with Neurofibromatosis Type 1: Feasibility of Multisite Collaboration Supporting Academic and Emotional Functioning in Teens with NF1.

**James Walker, PhD
Assistant Professor of Neurology, Harvard Medical School—Center for Genomic Medicine at Massachusetts General Hospital**

Training fellowship for Francisco Fernandez in the Walker Lab to develop a gene-targeted approach to upregulate NF1 expression using “CRISPR activation” (CRISPRa) to treat NF1 haploinsufficiency.

return service requested

Does your teen have Neurofibromatosis?

Are their symptoms hard to manage,
stressful, or bothersome?
Could they cope better with stress or anxiety?



ONLINE HARVARD STUDY

Learn stress and symptom management
skills, earn \$30 for participating, and help
advance scientific research!



Want to learn more?

Email the Study Coordinator,
Julie Brewer
jrbrewer@mgh.harvard.edu
or the Study PI,
Dr. Ana-Maria Vranceanu
avranceanu@mgh.harvard.edu

Join an NF community!
Virtually meet a group
of teens who have NF
from around the world!



Who can participate?

- Ages 12-17 years old
- Have a diagnosis of neurofibromatosis type 1 (NF1), neurofibromatosis type 2 (NF2), or schwannomatosis

What does it involve?

Participate in 8 virtual group
sessions (45 minutes) weekly
over Zoom, which are led by a
Harvard / MGH clinical
psychologist and expert in NF!

Complete questionnaires at the
beginning, end of the program,
6 and 12 months after program

***No costs, no medication,
and no travel**

Looking Forward

Mark your calendars for upcoming events!

Sunday, April 30

Steps2Cure NF Walk Dunkin Donuts Park,
Hartford CT

Saturday, May 6

Hats, Horses, and Hope Wright-Locke Farm,
Kentucky Derby Celebration Winchester, MA

Sunday, May 7

Steps2Cure Walk and 5K Run Central Park,
Schenectady, NY

Saturday, May 13

Steps2Cure Walk and 5K Run Lake Quannapowitt,
Wakefield, MA

Steps2Cure NF Walk RiverWorks, Buffalo, NY

Steps2Cure NF Walk Forest Park, Camden, NY

Saturday, May 20

Steps2Cure NF Walk Overpeck Park,
Ridgefield Park, NJ

Monday, June 12

37th Annual Golf Tournament Ferncroft Country Club,
Middleton, MA

Sunday, August 20

ASICS Falmouth Road Race Falmouth, MA

Saturday, September 9

**12th Annual Coast to the
Cure cycle event** Stage Fort Park,
Gloucester, MA

Monday, September 18

24th Annual Fall Golf Classic Four Oaks Country Club,
Dracut, MA

Sunday, October 1

Maine Marathon Portland, ME

Saturday, November 4

**Committed to Curing NF
dinner and casino night** Anthony's of Malden,
Malden, MA

*hosted by volunteers
Melissa and Mike Malerba*

Have You Checked Our New NF Northeast Website?

Find information about NF and schwannomatosis,
along with educational programs and more.

Visit us at www.nfnortheast.org.



Special thanks to Fourth Idea for partnering
with us to boost our NF Northeast brand!