



NF NEWS

NEUROFIBROMATOSIS NORTHEAST

Promoting Research, Awareness and Advocacy since 1988

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Summer 2018



Austin Gimbrone

Austin's Family is Taking Steps2Cure NF

Written by Cindy Gimbrone, Austin's Grandmother

Eight-year-old Austin Gimbrone is a smart, funny, and very outgoing soon-to-be third grader. He enjoys swimming, riding 4-wheelers or tractors, playing with his Nerf toys, and lately, riding his bicycle around the yard. He is a great conversationalist who will tell you a story or a joke that is usually bigger than life, as he loves to be the center of attention. Nothing can stop him from challenging anyone to a good game of "name that tractor or truck" - and we are always wrong.

During one of his regular pediatrician visits at the age of eighteen months, we were deeply concerned that Austin was still not walking and just learning to crawl. The doctor noticed he had more than six café-au-lait spots on his body. With a couple being on the larger side, it was recommended he see a genetic doctor for further evaluation which confirmed the diagnosis of neurofibromatosis type 1. That is when our family first heard of neurofibromatosis.

With Austin being the only one in our family to have NF, we were at a loss as to what his outlook would be and where we needed to go first. We were advised that he should see an ophthalmologist for his vision, who to this date he sees on a regular basis. He will tell you he has "super hero" eyes, which is excellent vision in his words. He has continuous MRIs to monitor his optic glioma and we have been very pleased that it has remained stable. He also is being monitored by an ENT (Ear, Nose and Throat) doctor for a papilloma in his nasal cavity which will eventually need to be surgically removed. This summer will be his first visit to a dermatologist to check his café-au-lait spots which have become more numerous throughout his body.

Austin's family is planning the first Steps2Cure NF walk in South Wales, NY on Saturday, September 29th

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2018 Scholarship Recipients

NF Northeast's Scholarship program was established with funds raised at **Steps2Cure NF** walks and road races and by the generosity of the **Staples Foundation**. The scholarships assist people with NF to continue their education after high school and pursue undergraduate degrees. Congratulations and good luck to these students!



Rosa Bailey

from New Hampshire, will attend Saint Joseph's College of Maine as a freshman



Ethan Guertin

from Massachusetts, will attend University of Maine as a freshman



Leah Manth

from New York, will attend Rochester Institute of Technology as a freshman



Haley Thompson

from New York, will attend The College at Brockport as a freshman

The mission of

Neurofibromatosis Northeast is to find treatments and the cure for neurofibromatosis by promoting scientific research, creating awareness, and providing hope and support to those who are affected by NF.

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#The word is
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A Message From the Chair of the Board of Directors



Lori Ryan, R.N., M.S.

Dear Friends,

This newsletter highlights many NF advocates and the importance of advocating for NF1, NF2 and schwannomatosis. Patients, family members and caregivers can provide unique and valuable perspectives about the disease.

The past several years have brought a greater focus on the patient voice in drug development. In a draft guidance issued in June, the U.S. Food and Drug Administration (FDA) details how drugmakers can gather comprehensive and representative input from patients. FDA Commissioner Scott Gottlieb stated, "FDA has made it a priority to work with companies and other stakeholders on gathering information from patients about their views and needs, and on building the tools that are needed to capture patient input in a way that provides meaningful data."

Patient organizations, like NF Northeast, are partnering with researchers to ensure that the goals of patients are kept in the forefront, to communicate the patient's needs and treatment options, and to participate in clinical trial design and planning.

There are many ways to be an NF advocate, such as educating others about NF, attending a NF Northeast event, hosting a fundraising event, and contacting state and federal representatives to support NF research through the National Institutes of Health (NIH) and Department of Defense's NF research programs.

Please contact us if you have any questions, comments or are interested in becoming more involved in our NF community. As always, many thanks to our staff, volunteers and supporters. Together we are strong!

Patient Engagement in Research

Patient engagement in research is increasingly recognized as an important component of the research process and promises to accelerate the development of new treatments by focusing researchers on real-life issues that they may not have otherwise considered.

NF Northeast board member Alex Powers Cellucci has NF2 and was a speaker at the MEDAFFAIRS LEADERS FORUM USA, a three day conference held recently in Cambridge, MA. Senior executives, leading experts and key stakeholders from the Life Sciences industry met to share ideas, achieve critical insights and discover practical solutions to enhance medical innovation for patient benefit. By working more closely with patients living with a disease, pharma can learn how to target endpoints that are appropriately responsive to patient concerns and design protocols that encourage trial participation.

By working more closely with patients living with a disease, pharma can learn how to target endpoints that are appropriately responsive to patient concerns...

This past November, Alex was selected as a patient representative member of the Response Evaluation in Neurofibromatosis and Schwannomatosis (REiNS) International Collaboration. REiNS is an international effort to develop new standardized response criteria for determining treatment response in patients with NF1, NF2, and schwannomatosis.

In addition to her patient advocacy work, Alex is also the chair of the NF Northeast Families and Community Committee.



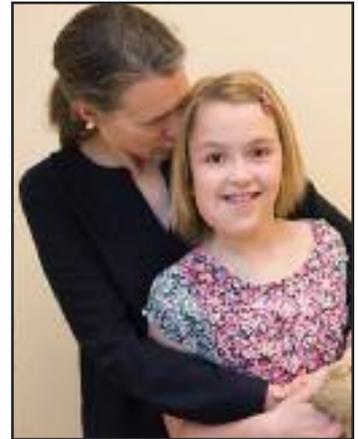
Alex Powers Cellucci

FDA grants orphan drug status to selumetinib for neurofibromatosis type 1 (NF1) treatment

In February, AstraZeneca and Merck announced that the U.S. Food and Drug Administration (FDA) granted Orphan-Drug Designation (ODD) for selumetinib, a MEK 1/2 inhibitor, for the treatment of neurofibromatosis type 1 (NF1).

Orphan drug status is given to therapies that treat, prevent or diagnose rare diseases. Although this designation does not necessarily mean that the treatment will be approved by the FDA or reach patients faster, it enables the drug's developers to receive incentives such as tax credits and provides them exclusive rights for seven years to further the treatment's potential for that condition.

Jane, shown on the right with her mom, Kristy, is 11 years old and has NF1. The primary manifestation of Jane's NF is a large plexiform neurofibroma on the right side of her face, jaw, and neck. Jane has been on the selumetinib trial for 4 years and initially had a 33% reduction in the tumor size. Her tumor has been stable overall in the past year, but some areas have shown some recent growth, and now it is at 27% of its original size. For this reason, and because Jane has grown so much herself in the past year, her doctors are increasing the dose of her medication over the next several months.



Kristy and Jane

Many of the clinical trials for MEK inhibitors have been done through the NF Clinical Trial Consortium, which is funded through the Department of Defense's Congressionally Directed Medical Research Program (CDMRP). So far, over 70% of participants in this MEK clinical trial are showing reductions of 20-50% in plexiform tumor size.

Research and clinical trials on MEK inhibitors have been possible in great part due to the advocacy work of NF Northeast and other NF organizations. Every year, we travel to Washington, DC with the NF Advocacy Network during Appropriations and we push for federal funding of NF research through the CDMRP, as well as language encouraging neurofibromatosis research through the NIH.

Resiliency for NF Study - A research project to enhance stress and symptom management

Are you feeling stressed? Are your NF symptoms bothersome?

The Family Center for Neurofibromatosis and the Integrated Brain Health Clinical and Research Program (IBHCRP) at the Massachusetts General Hospital are conducting a research study to compare 2 programs that teach stress and symptom management skills for adults with NF1, NF2, or schwannomatosis.

The Department of Defense is paying for this study.

What does the study involve?

8 group sessions led by an MGH psychologist with expertise in NF. Completion of questionnaires on your own computer at the beginning, end, 6 months and 12 months after the sessions. All sessions will take place from your home, using Vidyo, a secure software program you can use with a computer and a webcam to have live video-conferences, that we will help you install for free. Sessions are 1.5 hours long and held once a week.

This study DOES NOT involve taking medication. This study DOES NOT involve travel to MGH.

Who can participate?

We are looking for people ages 18 and older who have a diagnosis of NF1, NF2 or schwannomatosis.

What are the benefits?

If you participate in this research study, you will learn stress and symptom management skills that you can continue to use after the study. You will also interact with other adults with NF like yourself. There is no cost to participate in this study and your decision to participate will not affect your medical care.

How do I join?

If you would like to learn more about this research study, please contact the study coordinator, Sofia DiStefano, sdistefano1@mgh.harvard.edu, or the principal investigator, Ana-Maria Vranceanu, avranceanu@mgh.harvard.edu.

Camp New Friends - Generous donors make camp possible for children and teens with NF



NF Camp offers children and teens with neurofibromatosis a week of summer fun and social connections. It serves to reduce the stigma and isolation that is often associated with NF, and can be a life-changing experience for campers who have never met anyone else who lives with the condition.

We send a big THANK YOU to: **DCU for Kids** – a non-profit charitable foundation for children and families founded by Digital Federal Credit Union – for their

\$10,000 commitment to NF Northeast to support Camp New Friends, The **Boston Bruins Foundation**, who has been partnering with NF Northeast since 2011 to support the camp program, for their donation of \$2,500. And, last but not least, the **golfers at The International Golf Tournament** who responded to John Driscoll's challenge and donated \$42,000 for NF Camp!



Increasing Awareness of Neurofibromatosis

May is NF Awareness Month, and Governors and legislative leaders throughout the Northeast met with patients and families and signed official Proclamations.



In Albany, New York, Senator Jim Tedisco presented a Legislative Resolution to Susan Benjamin (l) and Rebecca and Vito Grasso



LT. Governor Karyn Polito of Massachusetts met with NF families and presented the Proclamation signed by her and Governor Baker

Rare Disease Day - February 28, 2018

Patient and Clinic Outreach Coordinator, Anne Patterson, was invited to represent NF Northeast at Metropolitan Hospital's Rare Disease Event in **New York City**. Dr. Maryam Banikazemi, Clinical and Biochemical Geneticist at the hospital, spear-headed the event. Anne and patient representative, Alwyn Dias (l), spoke with medical students, medical residents and staff about neurofibromatosis. This was an ideal opportunity to educate medical personnel in training and help them understand the different manifestations of neurofibromatosis and how it affects people's lives.



neurofibromatosis and how it affects people's lives.

In Boston, Michelle Hirsch Donovan, M.D., Ph.D. (below) was guest speaker at the Rare Disease Day program at the University of Massachusetts Club. Over 500 attendees heard Michelle talk about how neurofibromatosis has affected her daughter, Riley, as well as how other NF patients have been impacted. Michelle was recently featured on the Congressionally Directed Medical Research Program (CDMRP) website where she was recognized for having tackled NF from every possible angle: a fundraiser, advocate, spokesperson and most importantly, as Riley's mother.



NF Family Symposium at Massachusetts General Hospital



NFNE was proud to be a sponsor of the NF Symposium held at MGH on March 24th. Our thanks to Dr. Justin Jordan for coordinating a full day of informative presentations, research updates and breakout sessions. Attendees were also able to take the always popular lab tour to see NF research in action.

Photo: Dr. Justin Jordan (l) from the NF Clinic at MGH, Dr. David Miller from the NF Clinic at Boston Children's



Two friends named Emma from two different states display their Proclamations! Emma Soos (l) from New Hampshire and Emma Lee (r) from Massachusetts



Spreading the word all over Buffalo, NY via the Niagara Frontier Transit Authority
Shown at the rear of one of the busses are l to r: Linda, Leah and John Manth, Anne Patterson and "Dasher"

**#The word is
NEURO-FIBROMA-TOSIS**

Austin continued from page 1 For the 2017-2018 school year Austin was finally set up with an I.E.P. (Individual Education Program) and 504 education plans to stay on track with his academics. He receives occupational and physical therapy along with counseling during the school year that has helped him tremendously. He has increased his strength and is now able to ride a bicycle without training wheels. We are extremely proud of his achievements this year.

Recently our family became involved with a special organization: NF Northeast. While doing research on NF when my grandson was diagnosed, I happened to come across NFNE on a social media page and was pleased at the resources they had listed on their website. After further reading, I noticed that in the fall of 2017 they were planning to start a group for the Buffalo and Western New York areas. That was about the same time our new Oishei Children's Hospital of Buffalo was getting ready to open the new NF clinic. The clinic is fantastic, as when you go for an appointment you see multiple doctors at one visit instead of having multiple office visits. My family and I knew we had to get involved with this organization for Austin and other kids like him.



Austin and his Grandmother

Overjoyed with excitement over our newfound wealth of education and resources that we did not have before, our family felt very strongly about helping NF Northeast. Austin and our family were able to attend a Buffalo Bandits lacrosse game with other kids being treated at the Oishei NF clinic. We also got to enjoy a family night picnic at a local park where we met another NF family and had some discussion about our very first Buffalo and WNY "Steps2Cure NF" walk. As a member of the committee, we are pleased to report we have secured Saturday, September 29th, 2018 at Emery Park, South Wales, NY. We are so excited for this and we hope that all of our family and friends will be able to join us in this fun activity!

My grandson Austin has such a loving personality which I hope never changes. With him having NF1 we know it will bring him some challenges in his lifetime. I can only hope that the knowledge and resources we are giving him by being proactive will give him HOPE and COURAGE to overcome any of these challenges. I love you Austin William to the moon and back, your Grandmother Cindy.

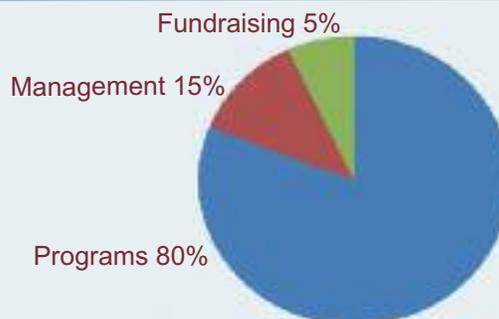
Development of NF Clinics

Being followed at an NF Clinic is beneficial to all patients who have neurofibromatosis, because it is a comprehensive team approach that facilitates continuity of care. This is more convenient for patients, who can have several doctor visits within one appointment. NF Northeast is dedicated to developing more NF Clinics throughout the northeast and has made this a priority. In New York, Anne Patterson, Patient and Clinic Outreach Coordinator, is a resource to the clinical staff and their patients at both the **Melodies Center at Albany Medical Center** and **John R. Oishei Children's Hospital in Buffalo**. She is available to meet with NF families and individuals after their appointments to see how NF Northeast can be a resource to them. Many families have benefitted by becoming involved in advocacy, increasing awareness of NF, meeting other families and participating in NF Community Events.

**When you give to NFNE
this is where your
support goes**

The Programs of NF Northeast are:

- Promoting and funding NF research
- Creating awareness of NF
- Providing hope and support to patients and families



Source - NF Northeast Audited Financials



*For more information about:
Programs, Management,
Governance and Financials of
NF Northeast
visit the Giving Common at
www.givingcommon.org*

Leave a Legacy



You can make a simple, yet transformational gift to Neurofibromatosis Northeast that costs you nothing in your lifetime by including Neurofibromatosis Northeast in your will or estate plans.

All it takes is a quick phone call to your lawyer to add a charitable gift to your will.

Contact Jillian Black at jblack@nfnortheast.org or (781) 272-9936 to learn more.

Become a Sustaining Giver

Join others who make a regular monthly donation to NFNE through their credit card.

It is convenient, and a little each month adds up to a lot in a year!

Making a difference in the fight to cure NF...



Carol Flaherty



Shawn Feeney

Runners, fans, and volunteers braved 35 mph winds, pouring rain, and a below-freezing wind chill during the 122nd **Boston Marathon**. But our NF Team, Shawn Feeney of Melrose, MA and Carol Flaherty of East Aurora, NY didn't let the conditions bother them and finished strong - all for NF!

On April 11th, NF Northeast celebrated the 19th Annual **Table for TEN** dining event in Boston with Honorary Co-Chairs Dan Andelman, host of Phantom Gourmet and Alex Pineda, Executive Sous Chef at Scampo. Following dinner at 25 of Boston's best restaurants, diners closed out the night at the University of Massachusetts Club. Live music was provided by Ancient Mysteries, and fabulous desserts were donated by Montilio's Bakery. We thank everyone that came out, especially Presenting Sponsor **Apex New England Construction**.



Alex Pineda and Dan Andelman

Awareness is Key! Bill and Kim Palmer are owners of Palmer Lock and Key and have an ongoing campaign to create awareness of NF. On Saturday, March 10th they



hosted their 4th Annual Comedy Night in Raynham, MA. Attendees had a great dinner followed by hilarious comedians and topped the evening off with a delicious birthday cake in celebration of NF Northeast's 30th anniversary!



On May 27th, the **Manth Family and Run4Leah** once again partnered with the Buffalo Marathon to raise funds and awareness of NF2. Leah Manth is the inspiration behind this great effort.



The 5th Annual Steps2Cure NF Walk in Camden, NY took place on Saturday, May 19th. Amanda Metott founded the walk that has grown to include many New York NF families.



3rd Annual Hats, Horses & Hope - NF Mom Melissa White (center above) put on yet another amazing party! Over one hundred well-dressed and stylish supporters of NF Northeast bet on their favorite horse in the Kentucky Derby and came together to support the race to cure NF.

On June 20th, the **33rd Annual NF Golf Tournament** took place at the International Golf Club. The success of this event would not be possible without the longstanding support and generosity of Presenting Sponsor **Grand Circle Corporation**, the many corporate sponsors, volunteers and the dedicated golf committee led by Chairperson Steve Picardi.



Upcoming Events!

NF Northeast is thrilled to have 27 runners participating in the **New Balance Falmouth Road Race on August 19th**. This is our largest team ever and includes



World Record Endurance Marathoner **Becca Pizzi** (l) who will also be sporting the NF Northeast red, white and blue singlet! If you are in the Falmouth area, please cheer on all of our runners who are doing so much for NFNE!



The **8th Annual Coast to the Cure NF Bike Ride** starts and finishes at Stage Fort Park in Gloucester, MA on **Saturday, September 8th** and offers four route options - 24, 40, 66 or 100 miles to accommodate all levels of riders! Register today at nfnortheast.org.

4th Annual Clubbing for Caden



NF Mom, Amy Spirito is once again organizing a golf tournament in honor of her son, Caden at the Brockton Country Club on **Saturday, September 15th**.

For information contact Amy at spirito2892@gmail.com.



Cruising with Motown for NF Saturday, August 25th, 2:30PM

Join Catherine Doherty and Loretta DeGrazia and cruise towards a cure in honor of their son, Miguel. Motown costumes are encouraged as you cruise around Marina Bay in Quincy, MA. For more information contact Loretta at oilgirl01@comcast.net!



Miguel



20th Annual Fall Golf Classic will be held on **Monday, September 24th** at the Four Oaks Country Club in Dracut, MA. We thank **Dassault Systemes** for generously sponsoring this milestone event. This will sell out! Go to nfnortheast.org to register!



Melissa and Mike Malerba are back with the **9th Annual Committed to Curing NF** event on **Saturday, November 3rd** in honor of their son, Jonathan. Join the fun with raffles, great food and casino tables at Anthony's of Malden, MA. Contact the Malerbabs at sheamelissa@hotmail.com.

Steps2Cure NF

There are 4 walks coming up soon (see back cover). If there is not one in your area, perhaps you would like to start one!

Stock up on your NF Northeast gear! visit the new Merchandise shop at nfnortheast.org

Get T-shirts, Sweatshirts, Hats, Socks and more!

The NF Families and Community Committee's mission

is to create local communities of peer support among NF patients and their families throughout the northeast. The next gathering will be a playgroup organized by Lauren Soos in **southern New Hampshire on September 29th**. Watch the NFNE website and Facebook for information about this and other informal get-togethers, or contact Alex Cellucci, Committee Chair at Apowers54@gmail.com.



The **Annual Sponsor Program** allows companies to support and participate in many of NFNE's activities by making one annual donation. It is a great way to spend your charitable dollars and get a lot of exposure. Plus, you will help fight NF. Why not give it a try!



Rob Ninkovich will be Honorary Chairperson of the **34th Annual NF Golf Tournament** at the International on June 19th, 2019. A retired outside linebacker, Rob played for eight seasons with the New England Patriots and won two Super Bowl titles with the team. The NF community is thrilled to have Rob on our team!



Parents! YOU can help us spread awareness of neurofibromatosis and NF Northeast – right in your own town! This year, please consider sponsoring you child's sports team on behalf of NF Northeast. You can get neurofibromatosis and NF Northeast on the t-shirts of an entire team for a donation to their individual league. If your child is playing any sort of sport this year please consider helping us spread the word by getting NF Northeast on teams all over the Northeast (and beyond!).



Let's make neurofibromatosis a household word!



9 Bedford Street
Burlington, MA 01803

30 YEARS | 1988-2018

return service requested

Taking Steps2Cure NF

Loudonville, NY
September 15th

Western New York
September 29th

Windham County, CT
October 20th

Wakefield, MA
October 21st

