

NF Tennessee Newsletter

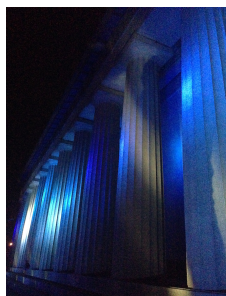


**Creating a Community
for Patients and Families**

Our Mission Statement:

NF Tennessee's mission is to create a community for patients and families with NF through support, education, and advocacy.

Shine a Light on NF:

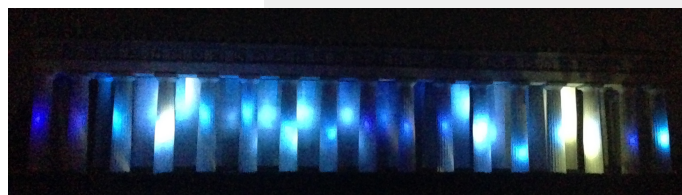


World NF Awareness Day is May 17th. This year, Nashville participated by lighting the Parthenon and the Korean Veterans Bridge to help raise awareness of Neurofibromatosis.



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The Nashville Parthenon at day versus at night (when illuminated with blue and white to Shine a Light on NF).

Visit our website at www.nftennessee.org and email us with any comments, suggestions, or ideas at contact@nftennessee.org.

NF Registries News:

Children's Tumor Foundation Patient Registry:

www.ctf.org/understanding-nf/nf-registry

CTF sponsors a registry for patients with NF1, NF2, and Schwannomatosis. The enrollment is now over 8000 patients. The aim is for 10,000. This registry collects information on symptoms and problems of individual patients. When combined this information helps researchers identify key problems and rare problems that deserve special attention. It also provides registered patients with information about potential clinical trials for some NF related problems. So joining the Registry helps all NF patients, and also can help the patient who is registering. We encourage our patients to look at the CTF website, and read about and join their NF Registry.

(ClinicalTrials.gov listing: NCT01885767)

Clinical Trials News:

Vitamin D Supplementation for Adults With Neurofibromatosis Type 1 (NF1) (VitDBoneNF1): A Phase II Trial on the Effect of Low-Dose Versus High-Dose Vitamin D Supplementation on Bone Mass in Adults With Neurofibromatosis Type 1 (NF1)

In addition to the problems of bone development that occur with NF1 like scoliosis and impaired bone growth (pseudo-arthritis), the bone mineral content of adults with NF1 is often low. This is called osteopenia and osteoporosis, and it can result in an increased risk of fractures and poor healing. Many adults with NF1 have low Vitamin D levels and that is known to cause osteopenia and osteoporosis. This international study is testing whether Vitamin D supplements, at two different doses, can prevent bone mineral loss in NF1 patients whose Vitamin D levels are low. The study is available at NF Clinics at the University of Utah and at the University of Cincinnati as well as sites in Canada and Europe. This study is of special interest because so many patients wonder if diet, nutritional supplements, or vitamins are helpful for neurofibromatosis. This study is an example of how that question is being addressed in a scientific way.

(ClinicalTrials.gov listing: NCT01968590)

Washington University in St. Louis Patient Registry Initiative:

Congratulations to the Washington University in St. Louis NF Clinic! Their NF1 Patient Registry completed enrollment in July 2017. Since opening in 2011, a total of 2,391 patients enrolled. Patients of all ages were included. To date seven articles have been published utilizing the NF1 patient registry.* Topics have included factors like racial and ethnic background, allergies, pregnancy-related issues, and parental age in relation to the development of pediatric brain tumors in NF1 patients. Also, the use of registries as a means of collecting data about rare diseases has been analyzed. Research using the registry information will go on for a long time. (Clinical Trials.gov listing: NCT01410006)



Photodynamic Therapy for Benign Dermal Neurofibromas: Topical Photodynamic Therapy (PDT) With Levulan® Kerastick® for Benign Dermal Neurofibromas Phase II

Treatment for neurofibromas on the skin has not received nearly as much attention as plexiform neurofibromas and malignant peripheral nerve sheath tumors. Yet for many NF1 patients, the cutaneous neurofibromas contribute a lot to the psychological and social effects of NF1. In this trial by Dr. Harry Whelan at the Medical College of Wisconsin, NF1 patients have several skin tumors treated with Levulan or with just a topical ointment without the Levulan (placebo). The Levulan makes tumors cells sensitive to light. The day after application the tumors are exposed to a special light. The growth of those tumors will then be measured over the next three years. Understanding of cutaneous neurofibromas was the subject of an extensive discussion at the recent Children's Tumor Foundation conference, and this study highlights the increase in efforts to treat this very common part of NF1.

(ClinicalTrials.gov listing: NCT02728388)

*See: Sharkey, EK, et al. "Validity of participant-reported diagnoses in an online patient registry: a report from the NF1 Patient Registry Initiative." *Contemp Clin Trials*. 2015 Jan, 40: 212-17.

And: Johnson, KJ, et al. "Evaluation of participant recruitment methods to a rare disease online registry." *Am J Med Genet A*. 2014 Jul, 164A(7): 1686-94.

Children's Tumor Foundation NF Conference 2017 Recap

The annual CTF NF Conference took place in Washington, D.C., on June 10 – 13, 2017. This was the largest meeting of NF researchers and clinicians ever, with over 350 individuals attending. Participants came from around the world, which has been a tradition at the CTF meetings.

The 2017 Friedrich Von Recklinghausen Award was presented to Dr. Karen Cichowski, Professor of Medicine at Harvard Medical School. Her research has focused on the development of tumors related to NF1 and have served as the foundation for clinical trials for the treatment of malignant peripheral nerve sheath tumors (MPNSTs), which are one of the most serious complications of NF1. The keynote speaker for the conference was Mr. Greg Simon, Executive Director of the Biden Foundation and former Executive Director of the Whitehouse Cancer Moonshot Initiative. From both a personal experience with cancer and from a career involved in accelerating the movement of cancer research strategies into clinical use for patients, he delivered a very inspiring and positive message on what the future holds for conditions like NF.

Among the research efforts that were highlighted at the CTF conference were analysis of the genes involved with NF1, NF2, and Schwannomatosis. A number of researchers, including Dr. Bruce Korf from the NF Clinic and Department of Genetics at the University of Alabama-Birmingham, discussed gene restoration therapy. The ability to turn genes on and off and the ability to correct the “read out” (i.e.

transcription and translation) of abnormal genes is starting to move out of the research laboratory and into patient treatment for some disorders. For example, this type of treatment has already been approved by the FDA for cystic fibrosis and spinal muscular atrophy. These types of treatments have created a huge interest in the NF

The Capitol Building in Washington, D.C. >>



community and really get to the heart of the CTF goal of “ending NF through research”.

Two areas of clinical concern that drew a great deal of attention were: 1) skin (dermal) neurofibromas, and 2) NF1 related problems in older adults. Neither of these areas has received a lot of research or analysis over the years. It is remarkable that while almost every NF1 patient will have some dermal neurofibromas, why the number varies so widely from one person to the next has never been understood. Why they tend to get more frequent in adults when other NF tumors like optic gliomas only occur in children is another mystery. Dermal neurofibromas do seem to have hormone sensitivity and have long been known to increase during pregnancy for some but not all women. Surgery for dermal neurofibromas that cause pain or other symptoms has been the traditional treatment, but the vast majority of these benign tumors are left untreated. Efforts to find better treatments, looking at medications instead of surgery, is one of the great needs of NF patients that has not been fully addressed yet.

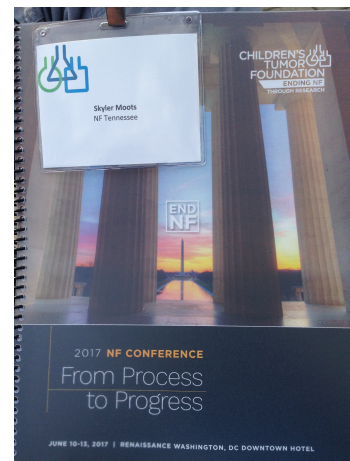
The realization that most medical problems for adults with NF1 have not been thoroughly studied is year-by-year becoming more widely emphasized at the CTF meeting. There already is an ongoing effort to establish guidelines for monitoring adults with NF1, NF2 and Schwannomatosis. The number of speakers at this year's meeting who addressed adult issues seemed to have increased noticeably. An entire afternoon was devoted to talks on “Learning, Memory and Behavior through the Ages in NF.” Dr. Eric Legius, from the University of Leuven, Belgium, highlighted the fact that memory and cognitive problems of children with NF1 continue into adulthood. He showed research results indicating...

[Continued on page 4]



[Continued from page 3] ...that some medicines used to target NF1 related tumors could also influence how nerves develop. This was followed by talks covering how children with NF1 learn social skills including how facial recognition is learned differently by NF1 children, how features of autism are different for NF1 children than other children with autism, how nerve networks in the brain are organized in children with NF1, and what animal models of NF1, in this case the fruit-fly, show about learning and memory in NF1. A common theme coming from the speakers in this session was the lack of research on what happens in older adults with NF1 in terms of memory and cognitive skills.

The CTF meetings really are an international event. One global NF effort is the International Neurofibromatosis Autism Consortium Team (INFACT) that meets in conjunction with the CTF meeting. To carry that idea even further, the 2018 Children's Tumor Foundation NF Conference will be held in Paris, France, next October.



NF Tennessee's Mission:

NF Tennessee's mission is to create a community for patients and families with NF through support, education, and advocacy.

How do we fulfill our mission?

- **Support**
 - Establish and maintain community support groups for NF patients and their families
 - Host a patient and family symposium to connect with the community and share information on the latest developments in NF research and treatments
- **Education**
 - Provide education about NF to patients, families, and caregivers
 - Publish a newsletter with educational information, as well as news, interviews, and events
- **Advocacy**
 - Advocate for NF patients and families in schools, workplaces, and civic institutions
 - Spread awareness of NF and participate in local events
- We are also committed to promoting the efforts of national NF-related organizations, such as the Children's Tumor Foundation and the NF-Network, in our local community and state.



About Us:

NF Tennessee Board of Directors:

- Dr. Paul L. Moots, M.D., President
- Ms. Elizabeth M. Wright, Vice President and Treasurer
- Mrs. Pennie Brooks, Secretary
- Mr. Theodore Goodman, J.D.
- Mr. Mitchell Korn

NF Tennessee Staff:

- Mr. Skyler Moots, Chief Operating Officer and Newsletter Editor

Consultants:

- Thrive Creative Group (Clarksville, TN)
- Mr. Mark Moots, Jr., C.P.A.

Non-Profit Status:

NF Tennessee is working to develop an active infrastructure to support neurofibromatosis patients, families, and caregivers in the Tennessee Valley region. We are in the process of applying for 501(c)(3) non-profit status, although that has not established yet.

Disclaimer:

NF Tennessee is an independent organization and is not affiliated with the Children's Tumor Foundation, the Vanderbilt-Ingram Cancer Center NF Clinic, or Vanderbilt University Medical Center, although some NF Tennessee board members are employed by Vanderbilt University (EMW, MK) or Vanderbilt University Medical Center (PLM).

Q&A: Healthcare Perspective with Dr. Sheryl Rimrodt-Frierson

Q: What is your work with NF?

A: Dr. Cutting and I moved to Vanderbilt in 2009 to continue our research. In 2015, Dr. Cutting, Dr. Barquero and I completed and published a study showing that students with NFI and reading deficits (NF+RD for short) benefitted from an intense burst of phonologically-based (teaching how letters represent certain speech sounds), multi-sensory reading instruction (using sight, sound, and touch to learn).^{*} In 2016, the National Institutes of Health funded our current clinical trial that builds on that previous study. I also have a clinic at Monroe Carell Jr. Children's Hospital where I care for school-age children, including some with NFI, who are struggling academically.

Q: What are the biggest academic hurdles for children with NFI?

A: The most common reasons that children with NFI come to my clinic are because they are having difficulty at school due to reading problems or Attention Deficit Hyperactivity Disorder (ADHD), or both.

Q: What would you want teachers to know about children with NFI?

A: I would want teachers to know that NFI is more than the birthmarks that you can see; it also affects the central nervous system (CNS). Two of the most common CNS effects of NFI are reading difficulties and ADHD. In fact, children with NFI are four times more likely to have learning disabilities than other children. I would want teachers to know that most children with NFI are trying to do their best in school, but they may need extra help.

Q: Can you tell us about your current clinical trial?

A: Yes, it builds on our 2015 study. Children with NFI have a deficiency of neurofibromin - a protein important in a signaling pathway that regulates learning and memory. Our previous work demonstrated that school-age children with NF+RD can respond to standard phonologically-based reading tutoring. We now propose to examine the synergistic (combined) effects of medication plus reading tutoring. More information can also be found on www.clinicaltrials.gov website (Identifier number NCT02964884).

-Here is a flyer with all of the information. >>>

^{*}See: Barquero, LA, et al. "Teaching reading to children with neurofibromatosis type 1: a clinical trial with random assignment to different approaches." Developmental Medicine & Child Neurology. 2015 Apr, 57: 1150-56. (You can find this journal article at DOI: 10.1111/dmcn.12769).



Because her father was in the Air Force, Sheryl grew up in many places, including Maine, Michigan, Illinois, England, Alaska, Alabama and California. She stayed in California for most of her education, including

college at Stanford University, medical school at the University of California, San Diego, and pediatrics training at Children's Hospital Oakland. She practiced general pediatrics for 7 years before moving to Baltimore to study Neurodevelopmental Disabilities at the Kennedy Krieger Institute (KKI). At KKI, she worked with Dr. Martha Denckla and Dr. Laurie Cutting doing research to better understand children with reading disorders, ADHD, and NFI.

ATTENTION FAMILIES OF CHILDREN WITH NEUROFIBROMATOSIS TYPE 1

We are currently looking for children and adolescents ages 10 to 17 with Neurofibromatosis Type 1 to participate in a research study. We are trying to learn more about the best ways to teach people to read.

WHAT WILL BE INVOLVED:

- ❖ Comprehensive evaluation of reading-related skills at Vanderbilt University
- ❖ 4 Magnetic Resonance Imaging (MRI) scans of the brain
- ❖ If your child meets our study criteria for having problems with reading, he/she may be invited to participate.
 - Children will be randomly assigned to one of three different groups.
 - 1: Reading tutoring program and a medication called Lovastatin
 - 2: Reading tutoring program and no Lovastatin (placebo)
 - 3: No reading (sham) tutoring and LovastatinLovastatin is not approved by the Food and Drug Administration for the treatment of reading trouble due to Neurofibromatosis Type 1.
- ❖ Parent and teacher questionnaires

WHAT YOU WILL RECEIVE FOR PARTICIPATING:

- ❖ Check for \$100 for the first visit, \$150 for the second visit, and \$100 for the last visit.
- ❖ Travel reimbursement available for NF-1 families living outside middle TN area

FOR MORE INFORMATION CONTACT:

Clinical Trials Center
Vanderbilt University
(615)-343-8010
clinicaltrialscenter@vanderbilt.edu

Laurie E. Cutting, Ph.D., Principal Investigator
Vanderbilt University, Department of Special Education
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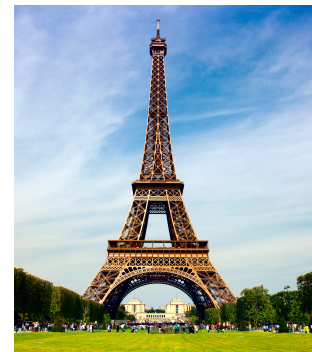


VANDERBILT UNIVERSITY
Institutional Review Board



Calendar: Upcoming Events in the Local and National NF Community

- **Cupid's Undie Run**
 - February 10, 2018
 - Nashville, TN
 - Benefiting CTF, this fun run (approximately one mile) is mostly a social event/excuse not to wear pants all for a good cause.
 - www.cupidsundierun.org
- **Rare Disease Day**
 - February 28, 2018
 - Nashville, TN – Tennessee State Legislative Plaza
 - Organized by the National Organization for Rare Disorders (NORD), this is a day to educate Tennessee lawmakers on rare diseases, such as NF, and advocate for beneficial policies.
 - www.rarediseases.org
- **World NF Awareness Day**
 - May 17, 2018
 - Worldwide
 - Last year in Nashville, the Parthenon and Korean Veterans' Bridge were lit up in blue and white for Shine a Light on NF. Be on the lookout for local events or ways that you can raise awareness.
 - www.ctf.org/get-involved/nf-awareness-month
- **2018 CTF NF Conference and Forum**
 - October 19-23, 2018
 - Paris, France
 - Maison de la Chimie
 - The annual CTF Conference and Forum will be international next year in the "city of light".
 - www.ctf.org/get-involved/nf-conference



<< The Eiffel Tower in Paris, France, the site of the 2018 CTF NF Conference and Forum

Get Involved! Interested in helping a non-profit start-up devoted to the NF community? How can you help?

- Volunteer to help organize events like the NF Tennessee Patient Symposium or a local support group
- Contact us about being interviewed for a future newsletter or being featured on our website
- Give us feedback on events or activities that you would like to see
- Spread the word about NF Tennessee to friends and family

Please email us at contact@nftennessee.org if you are interested!