

# NF Tennessee Newsletter

## Our Mission Statement:

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



## Introducing NF Tennessee:

We are a new non-profit organization, and this is our first newsletter. As we develop, we need input from the community to help us achieve our mission. We plan on publishing newsletters semi-annually, and our first major event will be the NF Tennessee Patient Symposium on June 24<sup>th</sup> in Nashville, Tennessee.

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.

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Visit our website at [www.nftennessee.org](http://www.nftennessee.org) and email us with any comments, suggestions, or ideas at [contact@nftennessee.org](mailto:contact@nftennessee.org).

## NF National News:

### NF Registries:

There are two major registries that are actively collecting information on NF patients: the CTF Patient Registry and the Washington University in St. Louis Patient Registry Initiative.

#### -Children's Tumor Foundation (CTF) Patient Registry:

[www.ctf.org/understanding-nf/nf-registry](http://www.ctf.org/understanding-nf/nf-registry)

CTF sponsors a Registry for patients with NF1, NF2 and Schwannomatosis that now includes over 7,000 patients. 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 while joining the Registry helps all NF patients, it can also help the patient who is registering. We encourage everyone to look at the CTF website, read about, and join their NF Registry.

#### -Washington University in St. Louis Patient Registry Initiative:

[nflregistry.wustl.edu](http://nflregistry.wustl.edu)

Another very valuable patient registry is being collected at the Washington University in St. Louis NF Center. This is for patients with NF1. This NF Center, which sponsors many clinical trials, is collecting information to understand the risks of developing cancer and other health problems in NF1 patients and to develop individualized treatments for those problems. Again, we encourage everyone to read about the Washington University in St. Louis NF1 Patient Registry Initiative (NPRI) and register.

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



### NF Worldwide:

In any small community and even in some big ones, it is easy for patients with NF to feel that nobody in the community knows much about NF. However, it is helpful to realize that NF is a worldwide concern, and there is a worldwide NF community. A good place to see that is on the NF-Network website\* which provides a list of NF organizations around the world. As another indication of the international efforts on behalf of NF patients, the Children's Tumor Foundation Conference in 2018 will be held in Paris, France.

\*Visit: [www.neurofibromatosis-network.org/en/](http://www.neurofibromatosis-network.org/en/)

## Ongoing Clinical Trials for NF-Related Issues:

The following clinical trials and other NF-related trials can be found at [ClinicalTrials.gov](http://ClinicalTrials.gov) by searching "neurofibromatosis". While some of the information is very technical, the basic information on the purpose of the trial is usually straightforward. There is detailed information on who is eligible and at what locations the trial is open.

#### -Phase II Trial of the MEK1/2 Inhibitor Selumetinib (AZD6244 Hydrogen Sulfate) in Adults With Neurofibromatosis Type 1 (NF1) and Inoperable Plexiform Neurofibromas

The connection between neurofibromatosis, tumors, and cancer treatment is strong, and this has made clinical trials for NF-related tumors a major part of ongoing research in NF. New medications that block the growth of cells that lack the NF1 protein or the NF2 protein have been discovered and are useful in some cancers that are not part of NF. There is a particularly promising set of medications that are being tested for progressive plexiform neurofibromas of NF1. Dr. Brigitte Widemann at the National Cancer Institute and collaborators at many NF Clinics are currently testing "MEK inhibitors" for progressing plexiform neurofibromas.

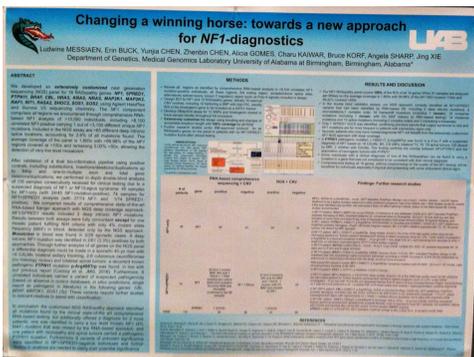
#### -Acceptance and Commitment Training for Adolescents and Young Adults With Neurofibromatosis Type 1, Plexiform Neurofibromas, and Chronic Pain

Clinical trials that address other features of NF are also very important. This trial, sponsored by the National Cancer Institute, is a good example that involves dealing with pain management for NF patients with tumors.

# Children’s Tumor Foundation NF Conference and Forum 2016 Recap

The annual CTF NF Conference and Forum took place in Austin, Texas June 18-21, 2016. Over 250 researchers including many from England, Europe, and around the world attended. Dr. David Viskochil, one of the researchers who helped identify the NF1 gene, received the Freidrich von Recklinghausen Award. Dr. Viskochil, who is a researcher and clinician at the University of Utah, is an active member of the CTF national organization and is also active in the CTF NF Camp for teens and young adults with NF that takes place every July in Salt Lake City, Utah.

Among the many excellent presentations of research and clinical topics related to NF, some of the most notable were efforts to develop guidelines for monitoring of adult NF1 patients. Guidelines laying out the frequency of visits and types of monitoring for children with NF1 were formulated many years ago and have been very valuable. Unfortunately, there are not similar best practice guidelines for adults, and this is widely viewed as a big need. These guidelines will include monitoring for tumors and cancer, blood pressure and heart issues, hormone problems and more.



*A presentation poster from the University of Alabama at Birmingham at the 2016 NF Conference <<*



*The Texas State Capitol Building in Austin, home of the 2016 NF Conference and Forum >>*

Other topics that were highlighted included the transition of plexiform neurofibromas to malignant peripheral nerve sheath tumors (MPNSTs), studies on optic gliomas, new medications undergoing clinical trials for tumors in NF1, NF2, and schwannomatosis, and investigations on the psychological and social aspects of NF1.

CTF provides invaluable support for high quality research for patients with NF, and the annual meeting is the premier event in NF research. CTF also fosters collaboration among researchers across the spectrum from laboratory science to everyday patient care.

The NF Forum took place concurrently with the scientific meeting and provided a great opportunity for patients to learn from specialists and from each other. Ms. Pennie Brooks, whose daughter was born with NF, is the Tennessee representative to the CTF Volunteer Leadership Council and attended the CTF Forum in Austin. (Look for more information about her experience in the Q&A section on page 5.) Overall, the Forum is an inclusive, educational, and social atmosphere.

The 2017 CTF NF Conference will be held in Washington, D.C., June 10-13. Online registration will begin soon on the CTF website ([www.ctf.org](http://www.ctf.org)).

## 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
- Provide support with skills specific to the non-profit business sector (Accounting, Administration, etc.)
- 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](mailto:contact@nftennessee.org) if you are interested!

## Q&A: Healthcare Perspective with Dr. Jennifer Brault

### Q: How did you become interested/involved in NF care?

A: Neurofibromatosis offers the perfect combination for my interests in genetics and neurology. While I was completing my residency, I had the opportunity to work in the NF Clinic at Riley Hospital for Children at Indiana University Health. I enjoyed seeing individuals and families with NF. I enjoy the follow-up care of the children throughout their childhood and adolescence.

### Q: What are the most important questions for parents of children with NF to ask a doctor?

A: Receiving a new diagnosis of NF can be scary and overwhelming. I think it is important to get educated, so ask for resources so that you will know what to expect. With NF, it is important to have ongoing screening during childhood for optic nerve tumors, scoliosis, other skeletal issues, hypertension, learning disabilities, etc. Ask about the screening that needs to take place, at what age it starts and how often it needs to happen. Largely children with NF do very well, but it is important to be vigilant and monitor for any changes. Issues like visual loss, new onset seizure, headaches, weakness, chronic pain, unintentional weight loss, change in stooling, or voiding problems need to be carefully assessed; let your doctor know if there are any new issues.

Ask about the risk for tumors. One of the scariest things to deal with in NF is the risk of tumors. In childhood, the biggest concern is for optic nerve gliomas (OPGs). Routine surveillance should be done in young children with NF as soon as they are old enough to cooperate. Recognize that most children with NF do not get optic nerve gliomas (only 1 to 2 out of every 10 children with NF are believed to have OPGs). Even in children who have OPGs, many will not require treatment as the tumors are typically slow growing and benign.

We often get questions about genetic testing. Genetic testing is available for NF but often can be diagnosed clinically. There are some situations where genetic testing may be considered to help clarify the clinical picture. Talk to your doctor about the testing options.

Finally, remember that children with NF are still children. Routine illnesses can be treated in the usual fashion. It is important to remember that common childhood illnesses are (by definition) *common* and can be treated in the same manner as children without NF. Also, children with NF should receive all of their childhood vaccines.

### Q: How can people get involved?

A: Ongoing research and the support of that research are critical for the development of treatments and hopefully a cure. Get involved in the local support groups. Participate in fundraisers and activities on the local and national level. Check out the Children's Tumor Foundation website ([www.ctf.org](http://www.ctf.org)). You can register your child (and any other affected family members) at this site and report symptoms. This is a national registry of patients so when new research studies are under development you can learn about them and decide if there are studies that are applicable to your family and if the studies are of interest to you.

[continued on page 5]



Dr. Brault

Dr. Jennifer Brault recently joined the Vanderbilt Children's Hospital and Vanderbilt's Neurofibromatosis Clinic. She is originally from northern Ohio and decided in elementary school that she wanted to be a pediatrician. As her education and training progressed, she specialized in pediatric neurology with a special interest in neuro-genetics. She attended Hillsdale College in Michigan as an undergraduate, before receiving a Master's Degree in Chemistry from the University of Toledo. She began medical school at the University of Cincinnati and then transferred to Indiana University after two years so that she could live and study in the same city as her husband. After graduating, she remained at Indiana University for residencies in pediatrics and child neurology. During this time, she was drawn to patients with underlying genetic disorders, which prompted her to pursue additional training in Child Neurology and Genetics. During this training, she had the opportunity to work in the Neurofibromatosis Clinic.

When asked about her family's move to Nashville, she said, "I was pleased to join the NF Clinic at Vanderbilt and help coordinate the care of children with NF in Tennessee. I have a wonderful, supportive husband and two beautiful daughters. We are delighted to call Nashville our home, and we are enjoying making new friends and exploring our new hilly and hot surroundings!"

[continued from page 4, Q&A with Dr. Brault]

**Q: What kinds of NF related problems in children tend to get overlooked?**

A: One of the biggest concerns in pediatric care is academic achievement. Children with NF are at risk for learning disabilities; frank intellectual disability can occur but is less common. Behavioral issues such as ADHD are also more common, which can make schooling challenging as well. Detailed neuropsychiatric testing can be a great tool for schools and families to assess their child's needs. Not all children with NF have learning issues, so many will not need testing. However, in those who do, testing is important. The testing should help demonstrate where a learner's talents are, where they may be struggling, and most importantly how they learn best. Having this information allows the schools, teachers, therapists, and families to create an individualized education plan (IEP) to help maximize their achievements in and out of school.

**Q: What do you see on the horizon for NF care?**

A: At the end of October, I attended the Child Neurology Society meeting. The first day of the conference was dedicated to the care of children with NF. At the meeting, many clinical trials were discussed, but the advances in medications aimed at the treatment of plexiform neurofibromas and malignant peripheral nerve sheath tumors were not discussed. More research needs to be done, but I am excited that there are prospective medications in the pipeline that may help treat symptoms and perhaps help find a cure one day.

## Q&A: Family Perspective with Pennie Brooks

*Pennie Brooks is a local NF advocate, activist, and organizer. She is a nurse who lives in Nashville and has a daughter with NF1.*

**Q: What roles do you play in the NF community?**

A: I am a member of the Volunteer Leadership Council for the Children's Tumor Foundation. We are dedicated volunteers who work all year and are passionate about Ending NF. Council members are the public ambassadors for patient education and community outreach within the Foundation. We are often the first contact that a family has when learning of a diagnosis. Council members learn how to better advocate for NF with legislators, learn about fundraising, support NF clinics, and raise awareness about NF in their local community.

Also, I am on the Advocacy Task Force for CTF, and we are working on a big project. Once the new administration begins in January, we will know better how to proceed with the Congressionally Directed Medical Research Programs. For NF, this program has been a major lifeline. I will also set up an NF Awareness Day again this year at the TN Legislative body when they are back in session.

**Q: What motivated you to attend the 2016 NF Forum?**

A: My daughter was born with NF, and I look forward to attending the Forum each year. The NF Forum is also for the NF community. It allows those living with NF and their families to connect, support, and learn from each other while attending seminars on relevant topics pertaining to neurofibromatosis. NF experts from around

the world cover the most current information on NF, including how to manage the symptoms of NF and updates on treatment strategies. There are fun activities, meals, and social time with friends and family.

**Q: What were the highlights of the NF Forum?**

A: Researchers and clinicians from all over the world displayed their research posters for our review, and many were available to answer my questions. I saw research going on from as far away as Japan, various countries in Europe, and Brazil in South America to name a few. There is a global concern about NF.

Texas Governor Greg Abbott was the keynote speaker for this largest NF gathering in the world. He spoke inspiring words about ending NF, saying, "the cavalry is coming and nothing can stop us!"

There was also exciting news related to the MEK Phase II clinical trial and the positive results of a child who had a reduction in his inoperable tumors. This is why we continue to raise funds to further potential treatments to manage the growth of these tumors.

**Q: Besides the annual NF Forum, what's an event that you are looking forward to in 2017?**

A: There's a lot to look forward to, but don't forget about Cupid's Undie Run on February 18<sup>th</sup>! The fundraising goal here in Nashville is \$175,000 to benefit CTF.

# Calendar: Upcoming Events in the Local and National NF Community

- **Cupid's Undie Run**
  - February 18, 2017, Noon – 4:00 p.m.
  - Nashville, TN – Dick's Last Resort
  - 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](http://www.cupidsundierun.org)
  
- **Rare Disease Day**
  - February 28, 2017
  - 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](http://www.rarediseases.org)
  
- **World NF Awareness Day**
  - May 17, 2017
  - Worldwide
  - Last year, the Parthenon in Nashville was lit up in blue and green 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](http://www.ctf.org/get-involved/nf-awareness-month)
  
- **CTF NF Conference and Forum**
  - June 10-13, 2017
  - Washington, D.C. – Renaissance Downtown Hotel
  - [www.ctf.org/get-involved/nf-conference](http://www.ctf.org/get-involved/nf-conference)
  
- **NF Tennessee Patient Symposium**
  - June 24, 2017
  - Nashville, TN – Vanderbilt University Medical Center
  - Join us for our first annual NF Tennessee Patient Symposium. At this event, experts will provide information on the latest NF treatments and discuss issues relevant to the NF community. This will also be an opportunity to connect and get involved in the local effort to support those with NF.
  - [www.nftennessee.org/events](http://www.nftennessee.org/events)
  
- **2018 CTF NF Conference and Forum**
  - October 19-23, 2018
  - Paris, France – Maison de la Chimie
  - [www.ctf.org/get-involved/nf-conference](http://www.ctf.org/get-involved/nf-conference)