

THE WASHINGTON UNIVERSITY NEUROFIBROMATOSIS (NF) CENTER

Exceptional Care through Groundbreaking Research

WELCOME!

The Washington University Neurofibromatosis (NF) Center is composed of clinicians and laboratory scientists focused on accelerating the pace of scientific discovery and its application to the care of individuals with NF.

Our mission is to galvanize and promote research on NF, achieving significant breakthroughs in the diagnosis and treatment of this condition. We believe that these breakthroughs are possible when researchers, medical professionals, and families partner together.

The Washington University Neurofibromatosis (NF) Center comprehensive care team offers detailed patient evaluations and assessments. They work seamlessly with families, referring physicians, allied health professionals and other agencies to deliver the most advanced medical services available to children and adults affected by NF.



THE 2014 WASHINGTON UNIVERSITY NF CENTER RESEARCH SYMPOSIUM

Please join us on May 16, 2014 for the second Washington University NF Center Research Symposium.

Our keynote speakers are:

Dr. Sean Morrison, Mary McDermott Cook Chair in Pediatric Genetics
Director, Children's Medical Center Research Institute at the
University of Texas—Southwestern
Investigator, Howard Hughes Medical Institute

Dr. Jonathan A. Epstein, William Wikoff Smith Professor of Medicine
Chair, Department of Cell and Developmental Biology
Scientific Director, Penn Cardiovascular Institute at the
Perelman School of Medicine at the University of Pennsylvania



In addition, there will be presentations on multiple aspects of NF1 ranging from a molecular understanding of specific NF1-associated features to how to provide meaningful care to individuals with NF1. We anticipate that this will be an engaging and informative experience.

There is no fee for attending, but registration is required.

For details, please contact Kirsten Brouillet at brouilletk@neuro.wustl.edu

SAVE THE DATE!

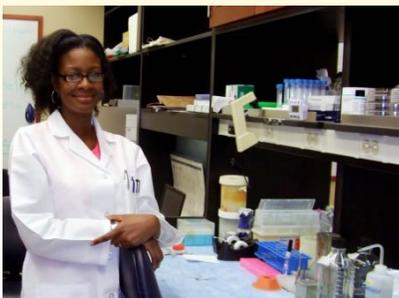
<p>CLUB NF APRIL 5, 2014</p> <p>Join us for rock climbing at Upper Limits!</p>	<p>SYMPOSIUM MAY 16, 2014</p> <p>Register now for the 2014 research symposium!</p>	<p>CLUB NF JUNE 7, 2013</p> <p>Join us at the St. Louis Zoo for a scavenger hunt!</p>
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RESEARCH AT THE WASHINGTON UNIVERSITY NF CENTER

GENDER INFLUENCES SYMPTOMS OF NF1

The following is a segment of an article that originally appeared in the *Record* on February 6, 2014 and was written by Michael C. Purdy

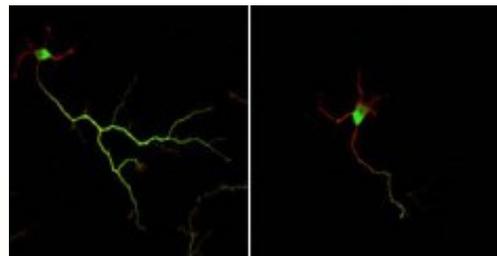
Researchers at Washington University School of Medicine in St. Louis have identified a patient's gender as a clear and simple guidepost to help health-care providers anticipate some of the effects of NF1. The scientists report that girls with NF1 are at greater risk of vision loss from brain tumors. They also identified gender-linked differences in male mice that may help explain why boys with NF1 are more vulnerable to learning disabilities.



Kelly Diggs-Andrews, PhD, a postdoctoral research associate in Gutmann's laboratory, reviewed NF1 patient data collected at the Washington University NF Center. In her initial assessment, Diggs-Andrews found that the number of boys and girls was almost equal in a group of nearly 100 NF1 patients who had developed brain tumors known as optic gliomas. But vision loss occurred three times more often in girls with these tumors.

With help from David Wozniak, PhD, research professor of psychiatry, the scientists looked for an explanation in *Nf1* mice. They found that more nerve cells died in the eyes of female mice, and they linked the increased cell death to low levels of cyclic AMP, a chemical messenger that plays important roles in nerve function and health in the brain. In addition, Wozniak discovered that only female *Nf1* mice had reduced vision, paralleling what was observed in children with NF1.

Two previous studies have shown that boys with NF1 are at higher risk of learning disorders than girls, including spatial learning and memory problems. To look for the causes of this gender-related difference, the scientists first confirmed that *Nf1* mice had learning problems by testing the ability of the mice to find a hidden platform after training. After multiple trials, female *Nf1* mice quickly found the hidden platform. In striking contrast, the male *Nf1* mice did not, revealing that they had deficits in spatial learning and memory.



When the researchers examined the brain regions involved in learning and memory in the *Nf1* mice, they identified biochemical abnormalities in the males but not in the females.

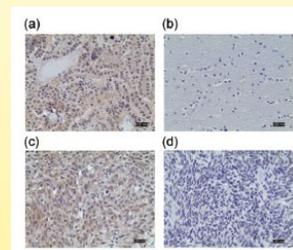
If hormones are responsible for these gender-linked distinctions in NF1, treatments that block hormonal function may be an option for use in patients with NF1.

NEW MUTATION IN MALIGNANT PERIPHERAL NERVE SHEATH TUMORS IDENTIFIED

Angela C. Hirbe, MD, PhD and her colleagues at the Washington University NF Center and the University of California – San Francisco have recently identified a new mutation in malignant peripheral nerve sheath tumors (MPNSTs). They discovered mutations in the *BRAF* gene – a gene previously implicated in skin cancer (melanoma).

In this new study, Dr. Hirbe demonstrated that *BRAF* mutations occurred in 20% of non-NF1-associated MPNSTs and in 2.7% of MPNSTs arising in people with NF1. This exciting finding suggests that future therapies might block *BRAF* activity, as has been used for other cancers.

Dr. Hirbe is a Hematology/Oncology fellow currently performing her post-doctoral research in the laboratory of Dr. David Gutmann.



MAKING A DIFFERENCE TODAY!

WASHINGTON UNIVERSITY NF CENTER TODDLERS SHOW PROGRESS AFTER BEAT NF



Children with NF1 can experience a range of developmental delays, including problems with gross motor skills and trouble learning how to appropriately socialize. A recent study showed that these delays can affect the development of children with NF1 as young as the age of three. Studies like this, and those currently taking place at the Washington University NF Center, highlight the need for early intervention.

Beat NF is a collaborative music therapy program co-developed by the Washington University NF Center, Jazz St. Louis and the Maryville Music Therapy Department. The goal of the program is to help toddlers with NF1 strengthen gross motor skills, improve social skills, and focus their attention through jazz music. After the completion of the first session, it has become clear that positive effects were emerging.

All toddlers who attended Beat NF were screened by Washington University NF Center physical therapist Courtney Dunn, PT DPT, for motor skills on the first and last days of the event. "Every child had a better motor screen after the final event and could preform skills they were unable to preform during the first screen," said Dunn. Some of those skills included the ability to leap, catch a ball and walk sideways.

One child, Sophia age 4, even showed growth in unexpected areas. "Her speech therapist and family members were amazed when they noticed a sudden change in her speech," said Dori, Sophia's mother, "The only change in what we were doing was having Sophia attend Beat NF and listen to the Beat NF music on days in between sessions."

Sophia, who was recently diagnosed with a brain tumor, became a leader during Beat NF sessions, encouraging her peers to follow the movements and sing along. "I think being with other children her age was definitely the highlight for Sophia. The chance to experience a group dynamic was huge for her," said Dori.



The Washington University NF Center and our collaborators are thrilled with the positive outcomes from our first session of Beat NF. Beat NF plans to launch new sessions in May 2014 so stay tuned for details!

CLUB NF CREATES ART



On February 1, 2014, the Washington University NF Center in collaboration with the St. Louis Children's Hospital Foundation hosted their first Club NF Event of 2014! The February program focused on self-expression and fine motor skills through the creation of visual art.

Club NF met at the St. Louis Artists' Guild in Clayton, MO. Families had the opportunity to explore multiple mediums including watercolor and acrylic paints, clay and mixed media. Families worked together to create joint pieces as well as developed individual canvases and sculptures. Local professional artist Ben Guffee worked with the families on understanding acrylic paint and different ways to integrate it into pieces of art, including traditional paint brushes and the less traditional tossing of balloons. Vicki Fried, the director of the new Arts as Healing Foundation in St. Louis, helped families develop a joint watercolor piece using fun techniques, such as blowing watercolor bubbles across the canvas.



All artwork created by our families will be displayed at the 2014 Washington University NF Center Research Symposium. We hope you will join us on May 16, 2014 for a chance to learn about research advances in the field of NF as well as to see the art created by our families.

GET INVOLVED IN RESEARCH!

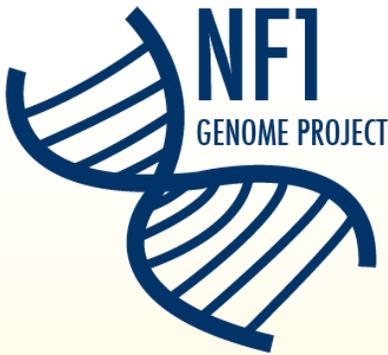
SOCIAL AND BEHAVIORAL HEALTH IN NF1



Researchers in the Washington University NF Center are currently recruiting both adults and children with NF1 to better understand the social and behavioral problems sometimes experienced by individuals with NF1. Recent studies have suggested that people with NF1 often have features seen in individuals with autism spectrum disorder. To better understand this possible connection, Dr. John Constantino and his colleagues have initiated a detailed study as an initial step towards uncovering potential genetic causes for this problem in children with NF1.

The study involves completing a few questionnaires. You will receive \$25 for your participation. For more details, please contact Alicia Vallorani at vallorania@neuro.wustl.edu.

THE NF1 GENOME PROJECT REACHES 350 PARTICIPANTS



In late December 2010, the Washington University NF Center established the NF1 Genome Project (NF1GP). Now, three years later, the NF1GP has over 350 participants. The Washington University NF Center extends its gratitude to the individuals with NF1 who have donated their DNA to this initiative. These samples will allow Washington University NF Center investigators to determine how changes in the DNA of people with NF1 predispose affected individuals to specific medical problems.

Although the NF1GP has reached a significant benchmark in its history, it is still vitally important for those with NF1 to continue to provide DNA samples to this project. We have set a goal of 1,000 specimens over the next several years.

With exciting advances in the field of genomic science, now is the time to work together—researchers, clinicians and families alike—to find the best possible treatments for people with NF1.

If you are interested in participating or in learning more about this project, please contact Alicia Vallorani at vallorania@neuro.wustl.edu.

STAY CONNECTED!

WEBSITE



Washington University
Neurofibromatosis Center



FACEBOOK



YOUTUBE



We want to hear what you think! For questions or to submit ideas for future Washington University NF Center newsletters, please email:

Alicia Vallorani at vallorania@neuro.wustl.edu