NF RESEARCHERS DISCOVER ROLE FOR NF1 IN CIRCADIAN RHYTHM FUNCTION

Investigators from the Perelman School of Medicine at the University of Pennsylvania, led by Dr. Amita Sehgal, found that the NF1 gene controls the brain’s perception of time of day signals.

Working in flies, her team previously reported that the Nf1 gene is important for establishing and maintaining time of day (circadian) rhythms. However, the circadian clock was normal in Nf1 mutant flies, suggesting that the Nf1 gene might be important for relaying the signals from the clock to other regions of the brain involved in sleep-wake cycling.

In their new study, they demonstrate that the Nf1 gene in flies is critical for the function of multiple different cell types involved in rest/activity rhythms. Moreover, they worked with investigators in Dr. David H. Gutmann’s laboratory to show that the Nf1 gene also controls these daily rhythms in mouse brain cells.

These exciting findings suggest that the Nf1 gene is a key regulator of daily rhythms in fly and mammalian cells relevant to the sleep disturbances common in children and adults with NF1.

Dr. Sehgal’s findings were published in the journal Cell Reports.
PATIENT SPOTLIGHT: LEXI’S JOURNEY WITH NF1

Forward Strides 4NF: Walking for awareness, Racing towards a cure! Forward Strides 4NF is a charity that I started in 2016 in honor of two very special people who both deal with Neurofibromatosis (NF) on a daily basis - my husband, Alex, and my daughter, Lexi.

Alex was diagnosed with Neurofibromatosis type 1 (NF1) when he was in the second grade. NF1 caused some learning difficulties that he was able to overcome with the help of some wonderful teachers at St. John’s Lutheran School. After learning of his NF1, he had a team of incredible doctors in St. Louis who monitored his condition on an annual basis. Alex had some minor surgeries to remove some of the neurofibromas on his body. He feels very fortunate that he has been able to live a normal, happy life. He has a passion for the outdoors, and enjoys hunting and fishing.

Lexi was born with a plexiform tumor on the bottom of her left foot and a leg length discrepancy. As a result, Lexi currently wears leg braces and inserts in her shoes, as well as a lift on the bottom of her right shoe. Since the age of 5 months, she has had several MRI scans and routine eye exams. She has also had an ankle surgery, where they placed a plastic implant to help with the stability of her ankle, and two staples to slow down the growth of her ankle bone.

Lexi receives both physical and speech therapy each week to help with her low muscle tone and communication skills. She is a very strong, independent four-year-old who doesn’t allow NF1 to limit her daily activities. She loves to swim, has participated in tumbling, and is currently taking dance lessons. Even with her braces and shoe lifts, she keeps up with the other children to the best of her ability.

NF1 is a worrisome genetic disorder because of the “unknown.” No one individual is the same. You don’t know what to expect. Alex has overcome minor hurdles with his NF1, but we still face hurdles with him. Education is key with any genetic disorder.

Staying abreast of all the research and medical advancements going on in the NF world is very important. We are very thankful for the progress that Lexi has made overcoming some obstacles that she has faced with already. We could not have done this without Dr. Gutmann and his amazing team. Since Lexi was 10 months old, she has been seeing Dr. Gutmann annually for her exams. We have also been so fortunate to have one of the best physical therapists, Dr. Courtney Dunn, who is also a part of the NF Center team. Since she was 18 months old, Lexi has attended the Beat NF music motor therapy program offered through the Washington University NF Center.

Trying to reach out in the St. Louis city community to see who else is affected with NF1 is challenging. In 2015, I helped organize a Great Steps 4NF walk in conjunction with NF Midwest. After raising over $16,000 for Great Steps, I decided that I wanted to help raise money that stayed here in the St. Louis community. In 2016, Forward Strides 4NF was formed. For the past 2 years, we have hosted a 2-mile family fun walk in Valley Park, MO. These events occur at the end of September each year, and 100% of our proceeds go to the Washington University NF Center.

Our walks have definitely been a huge success. We raised $19,351.00 in 2016 and $18,679.00 in 2017. We have had over 200 registered walkers both years, with over 30 volunteers. Entertainment at the event included an inflatable slide and bouncy house, bubble bus, face painting, Minnie Mouse and a clown that handed out balloon animals. In addition, the participants were greeted by Fred Bird and Louie at the finish line. We also hold a raffle, which included over 120 amazing items; this proved to be a HUGE attraction that alone generated over $4,300. Lastly, local restaurants donate beverages, snacks and food for all the participants after the walk is completed.

Trying to maintain a positive outlook given the diagnosis of NF1 is sometimes hard. There is a not a day that goes by when we don’t think about the NF community. However, we don’t let NF1 control our life, we try to run it ourselves. We keep up with our weekly therapy appointments, we are always learning more about NF1, and we stay up to date about new medical advancements. Importantly, we do our best to raise money to support the outstanding research at the Washington University NF Center. We are walking for awareness every day, and racing towards finding a cure in the future.

Mark your calendars for Saturday, September 29, 2018 for the Forward Strides 4NF Walk. Join our Facebook group as well: Forward Strides 4NF to keep up with quarterly fundraising events and updates regarding the walk.

-Written by Gina Wilburn (Mom)
2018 WASHINGTON UNIVERSITY NF CENTER RESEARCH SYMPOSIUM

We are pleased to announce that the fourth biennial NF Center Research Symposium, celebrating 25 years of NF at Washington University, will take place on Wednesday, September 5, 2018 at the Eric P. Newman Education Center (EPNEC) located on the campus of the Washington University School of Medicine.

Dr. David H. Gutmann, Director of the Washington University NF Center and the Donald O. Schnuck Professor of Neurology at the Washington University School of Medicine, is pleased to announce Eric Legius, MD, PhD from the University of Leuven, Belgium and Frank McCormick, PhD, FRS, DSc (Hon) from the University of California, San Francisco are the two keynote speakers. This day-long symposium will highlight advances in NF research and clinical care.

Dr. Legius is Professor of Human Genetics at the University of Leuven, Belgium. He and his colleagues have been leaders in characterizing NF1-like syndromes and in identifying genotype-phenotype correlations important for precision medicine.

Dr. McCormick is Professor of the UCSF Helen Diller Family Comprehensive Cancer Center and holds the David A. Wood Chair of Tumor Biology and Cancer Research at UCSF. He and his team were one of the first to define the role of RAS in human disease and to demonstrate that the NF1 protein is a key regulator of RAS.

To register for this event, please visit our website at http://nfcenter.wustl.edu.

THE NF1 MUTATION AS A RISK FACTOR FOR AUTISM IN INDIVIDUALS WITH NF1

Although every person with Neurofibromatosis type 1 (NF1) is born with a mutation (change) in one of their two NF1 genes, the problems arising in any one person can be quite different. In an attempt to better understand the relationship between specific NF1 gene mutation and autism, Dr. Stephanie Morris examined over 50 people with NF1.

Leveraging existing data generated in the Washington University NF Center, Dr. Morris, working with David H. Gutmann MD, PhD, found that people with mutations within the first half of the NF1 gene were less likely to have severe autism symptoms compared to people with mutations in the second half of the gene. This is the second study from the NF Center to demonstrate that the location of the mutation within the NF1 gene may be an important predictor of specific medical problems.

While these findings are compelling, they are currently not specific enough to allow doctors to predict which child will (or will not) develop autism symptoms. Future studies are currently underway to identify additional factors that may improve our ability to identify those children at greatest risk for autism and other neurodevelopmental problems.

This report was published in the journal, Neurology.
GARY GUO JOINS THE GUTMANN LABORATORY

Gary Guo, MD joined the research laboratory of NF Center director, David H. Gutmann, MD, PhD, as foreign graduate student. Dr. Guo received his medical training at Shandong University in Jinan China, and is currently a PhD candidate in the Department of Neurosurgery. Over the past several years, Dr. Guo has been studying malignant brain tumors. For his graduate studies, Gary will be using stem cells from low-grade brain tumors arising in Nf1 mutant mice with optic gliomas to find new treatments for children with NF1-associated brain tumors.

UPCOMING EVENTS

CLUB NF YOGA & SWIMMING
AUGUST 4, 2018, 10am - 12pm

NF CENTER RESEARCH SYMPOSIUM
SEPTEMBER 5, 2018, 9am - 4pm

For more details, or to RSVP, please visit our events website at:
https://nfcenter.wustl.edu/events/