Defining why people with NF1 have learning and memory deficits can be challenging in mice. For one, the human brain is far more complex than the mouse brain. In addition, it is not known whether brain nerve cells (neurons) from people with NF1 have the same abnormalities that have been reported in neurons from Nf1 mouse models.

However, a new technology has changed all of that. Scientists were awarded the Nobel Prize several years ago for pioneering work in which skin cells could be reprogrammed into stem cells capable of generating nearly every type of cell in the body.

Leveraging this method, Corina Anastasaki, PhD, a post-doctoral fellow in Dr. David Gutmann’s laboratory, recently converted skin cells from people with NF1 into neurons to determine what underlies some forms of learning and memory problems in children with NF1. Dr. Anastasaki found that these neurons had different levels of a neurotransmitter, called dopamine, which directly correlated with memory deficits observed in mice.

Her studies are the first to use reprogrammed neurons that model those found in the brains of people with NF1.
“Sports seem really hard for my child.” I often hear this comment from parents in the Washington University NF Center Clinical Program, and each time I feel frustrated that I don’t have a good recommendation to alleviate this concern. After observing the gross motor skills of over 100 children with NF1 in our clinic, I have seen several trends:

- They tend to be physically weaker than their peers.
- They typically have more difficulty with coordination and balance, making riding a bike and playing a variety of sports especially challenging.
- They frequently report high levels of fatigue with daily activities.

Despite these generally accepted developmental challenges in children with NF1, there have been very few research studies initiated to examine methods to improve the motor skills in these children. Currently, there is only one published outcome-based study, and it found that:

1. Children with NF1 can improve their strength through specific strengthening activities, and
2. Specific strengthening activities appear safe for children with NF1.

As a result, in the next several Courtney’s Corner blogs, I will be exploring various aspects of gross motor skills and providing simple ideas to improve motor skills in children with NF1. When looking at making changes in motor skills, we need to keep one thing in mind: change takes time. It reminds me of advice a friend gave me when I decided to train for a marathon. She explained to me that training for a marathon starts off really fun – everyone is excited for you, and it is exciting to talk about your new goals with other people. However, once you are finished talking and the true work begins – long runs alone in the rain, sore muscles and less time for completing other daily tasks, the training becomes less fun, and it can be very easy to lose sight of your end goal and give up.

I have often observed a similar experience when working on skills with children. When the team discovers an area of concern that is important to address, we get excited and are eager to “get to work.” However, much like increasing mileage when running, achieving noticeable progress takes time. To help your child avoid burnout, it is vital to find methods to incorporate activities into your daily routine. More specifically, I recommend sneaking activities into your day by reducing screen time! In fact, one of the best ways to support your child in achieving a new level of gross motor skills is by increasing the activity level in your entire household. Although sitting in front of a screen can be educational, relaxing, or just plain fun, reducing the time you and your children spend looking at screens can really change the dynamics of your day. Read more at [http://nfcenter.wustl.edu/courtneys-corner-improving-gross-motor-skills-in-children-with-nf1/](http://nfcenter.wustl.edu/courtneys-corner-improving-gross-motor-skills-in-children-with-nf1/).

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**CLUB NF LEARNS CHESS STRATEGY**

Everyone had a great time learning how to play chess and discovering new chess strategies at the June Club NF event! Chess experts from the Chess Club and Scholastic Center of St. Louis joined us for a two hour tutorial, teaching the children everything they needed to know about chess, from the rules of the game to optimal strategies for success. The event encouraged development of executive function and planning skills, which are often delayed in children with NF1.
RESEARCHERS USE WHOLE GENOME SEQUENCING TO IDENTIFY GENES INVOLVED IN MALIGNANT PERIPHERAL NERVE SHEATH TUMOR FORMATION IN NF1

Malignant peripheral nerve sheath tumors (MPNSTs) are extremely aggressive soft tissue sarcomas that occur more frequently in people with the Neurofibromatosis 1 (NF1) tumor predisposition syndrome. These cancers often arise from benign plexiform neurofibromas.

Over the last several years, since completion of the sequencing of the human genome, whole genome sequencing techniques have become an important tool to understand the genetic changes that occur in cancer.

Leveraging these whole genome sequencing methods, Angela C. Hirbe, MD, PhD, an oncology fellow and postdoctoral researcher in Dr. David Gutmann’s laboratory has performed the first-ever whole exome sequencing to identify the genetic changes that accompany the transition from benign to malignant peripheral nerve sheath tumor in a single individual with NF1 over a 14-year period.

In these studies, Dr. Hirbe discovered a new gene which is both mutated and overexpressed in MPNSTs. Importantly, she found that reducing the levels of this protein in mouse MPNST cells decreased tumor growth. Dr. Hirbe is now working to determine how this new gene controls cancer cell growth relevant to the identification of new therapies for these deadly malignancies.

RENEWED JAZZ MUSICIAN PLAYS FOR KIDS AT ST. LOUIS CHILDREN’S HOSPITAL

Considered one of the foremost jazz violinists of her generation, Regina Carter and accordionist, Will Holshouser, generously volunteered their time to play for in-patient children at St. Louis Children’s Hospital during a recent trip through St. Louis. Several members of the Washington University NF Center and Jazz St. Louis gathered with the pediatric patients and enjoyed an afternoon of great music.
SUMMER 2015 BEAT NF

Join us at the upcoming spring sessions of Beat NF, a toddler and parent jazz music therapy group focused on developing motor, social and attention skills in toddlers with NF1, while fostering parent child interactions and jazz appreciation.

WHO: Toddlers (2.5 - 5 years) with NF1 and their parents/caregivers

WHEN: Tuesday mornings June 16 - July 14, 9:30 - 10:45 AM

WHERE: The Harold and Dorothy Steward Center for Jazz, 3536 Washington Ave., St. Louis, MO 63103

MARK YOUR CALENDAR!

Interested in attending any of these events? RSVP to Kirsten Brouillet at brouilletk@neuro.wustl.edu today!

CLUB NF PLAYS AT THE LODGE!

Join us at the Des Peres Lodge for a morning of gym games and dance lessons, followed by an afternoon enjoying the indoor/outdoor pools and waterslides!

WHO: Children grades K-8 with NF1 and their siblings

WHEN: Saturday, August 1, 2015, 9:30 a.m. - noon

WHERE: The Lodge Des Peres
1050 Des Peres Rd., Des Peres, MO 63131

SATURDAY, OCTOBER 3 - CLUB NF HEADS TO FUNFEST!

STAY CONNECTED >> nfcneter.wustl.edu

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