Power in Numbers: NPRI Enrollment Worldwide

Approximately one in 3,000 or two million individuals worldwide have Neurofibromatosis Type 1 (NF1). NF1 is equally distributed among both sexes and has no particular racial, ethnic, or geographic distribution worldwide.

It can be difficult to assemble information from enough people for epidemiological research of rare diseases, like NF1. Having a large number of participants is important because it allows researchers to see patterns in the data. This could help them determine, for example, why some individuals with NF1 develop severe complications with NF1 while others do not. For example, what factors increase the risk of high blood pressure in NF1? The Washington University NF1 Patient Registry Initiative (NPRI) was developed to support NF1 research and to one day help doctors develop personalized treatment for individuals with NF1.

The NPRI is open to individuals with NF1 from across the globe. This online questionnaire collects information to help doctors and researchers better understand the medical and social issues experienced by children and adults with NF1. There are currently almost 1,600 people from 47 countries across the world participating in the registry!

Map of NPRI Participation

There is power in numbers. We are continuing to work to expand the number of individuals signed up for the registry but we need more participants. You can help by spreading the word.

If you know someone with NF1— or who has a child with NF1— encourage him/her to visit the registry (https://nf1registry.wustl.edu) to learn more about this important initiative and determine whether they are interested in participating. We greatly appreciate your contribution to the registry and look forward to expanding in the future.
Registry Requests

Frequent information requests from registry participants

Social Skills in Children with NF1

Children with NF1, like all children, differ in their social skills and abilities. While some have few challenges interacting with others and making friends, others find social interactions very difficult.1 Studies have found that, on average, children with NF1 have challenges making friends and are teased more often than children who do not have this condition. They may also be less involved in hobbies, clubs, and other out-of-school activities.2

There is no established theory to explain why children with NF1 experience more social problems than their peers. Researchers think these difficulties could relate to: (1) changes in brain structure and function due to having NF1, (2) complications associated with having NF1, and/or (3) effects of having a chronic disease.1

For example, children with NF1 may have difficulty in social interactions because of challenges with perception and interpretation of social cues, like facial expressions and emotions.2,3 Various complications associated with having NF1—such as increased risk for learning disabilities and attention deficit hyperactivity disorder (ADHD) and visible manifestation of lesions and neurofibromas—could also contribute to social challenges.3

Finally, just like any child with a chronic disease, children with NF1 are often absent from school due to medical appointments. This could make it hard for them to make friends.3

Since every child with NF1 is different, there is no universal solution to addressing the social problems he/she may experience. If you are concerned about your child’s social skills, it is important to talk with his/her pediatrician or NF specialist. Some steps that could be helpful in figuring out the best intervention include:

- having your child screened for social and emotional issues to determine if they should be seen by a clinical psychologist or psychiatrist, and/or
- having your child evaluated and, if necessary, treated for issues with speech, motor function, and cognitive ability (since these can all affect someone’s ability to interact with others).1

Even though we have learned a great deal about NF1 in the past few years, more research is needed to clarify why children with NF1 struggle with social interactions, and what treatments would be helpful in overcoming this issue. As an NPRI participant, you are contributing directly to this work. Along with medical information, the NPRI’s online questionnaire gathers data to better understand the social problems experienced by people with NF1. This could one day help doctors and researchers develop personalized treatments that improve quality of life for individuals with the condition.

References:


Nearly half of all children with NF1 have low tone (hypotonia). A recent study at the Washington University NF Center suggested that children with both NF1 and hypotonia may be at greater risk for developing a brain tumor. However, there are few objective methods for evaluating hypotonia.

To address this need, Courtney Dunn, PT DPT, led a study designed to establish criteria for assessing hypotonia in children with NF1. She found that hypotonia could be reliably diagnosed if a child’s head lags at or behind the shoulders when pulled up from a prone position (pull-to-sit-test) and the child has increased flexibility at the hip joint (hip range of motion).

Dr. Dunn is currently using these findings in a prospective study to determine whether hypotonia can be used to predict which children with NF1 are at greatest risk for developing brain tumors.


**Gender Influences Symptoms of NF1**

Researchers at Washington University School of Medicine in St. Louis have identified patient 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 and a brain tumor are at greater risk for vision loss. They also identified gender-linked differences in male mice that may help explain why boys with NF1 are more vulnerable to learning disabilities.

“This information will help us adjust our strategies for predicting the potential outcomes in patients with NF1 and recommending appropriate treatments.” said David H. Gutmann, MD, PhD, the Donal O. Schnuck Family Professor of Neurology, who treats NF1 patients at St. Louis Children’s Hospital. In addition, “further understanding of the interplay between sex and NF1 may change the way we manage individuals with this common brain tumor predisposition syndrome.”


To learn more about these and other clinical studies please visit:

http://nfcenter.wustl.edu/research/nf-clinical-trials/
Meet the Team

Dr. Kimberly Johnson is an Assistant Professor at Washington University. Dr. Johnson has spearheaded work at the Washington University NF Center to develop the international registry of individuals with Neurofibromatosis Type I. She serves as the principal investigator of the initiative. What Kim enjoys most about working with the registry is building a virtual community to conduct research that she hopes will ultimately improve the quality of life for individuals living with NF1.

Dr. David H. Gutmann is the Donald O. Schnuck Family Professor of Neurology. Dr. Gutmann was awarded the 2012 Friedrich von Recklinghausen Award and the Distinguished Researcher Award from Washington University School of Medicine in 2013 for his significant contributions to NF research and clinical care.

Nancy Zoellner is the manager for the registry. Nancy is from St. Louis, MO, and holds a Masters Degree in Public Health and a Bachelors degree in Psychology and Gender Studies from Washington University. What Nancy enjoys most about working with the registry is being able to communicate with people from all over the world and working to constantly improve the procedures of the research team to better accommodate all participant needs.

Qian Liu is a research assistant for the NPRI. Qian is from Hunan, China, and has a Bachelor in Medicine from XiangYa School of Medicine, China. What Qian enjoys most about working with the registry is using her data analysis skills to keep up with the ever changing participant information and create automated data solutions.

Evelyn Sharkey is a research assistant. Evelyn is from Orono, ME, and has a Bachelors in Biopsychology from Tufts University. What Evelyn enjoys most about working with the NPRI is getting feedback and questions from participants and working to address them in the quarterly newsletters.

Melody Schaeffer works as a research assistant. Melody is from West Plains, MO and has a Bachelors in Biochemistry from Rockhurst University. What Melody enjoys most about working with the NPRI is getting to talk with the participants and assist them with continuing their participation with the registry.

Claudia Landazabal works as a research assistant as part of her involvement with the MARC uSTAR pre-PhD program funded by the National Institutes of Health. What Claudia enjoys most about working with the NPRI is identifying any difficulties participants have encountered in order to improve participant experience.

Salma Abadin works as a research assistant. Salma is from Milwaukee, WI and has a Bachelor of Arts degree in Classics from Macalester College. What she enjoys most about working with the NPRI is having the opportunity to gain experience with primary data collection and management as well as interacting directly with participants.

We want to hear what you think about the NPRI Newsletter! For comments, questions, or to submit ideas for future NPRI newsletters please email us at: nf1registry@brownschool.wustl.edu. This and other newsletters can be viewed at https://nf1registry.wustl.edu/newsletter.aspx.