

What Is Neurofibromatosis Type 1?

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Neurofibromatosis (NF) is a common complex genetic condition. There are two types of NF: neurofibromatosis type 1 (NF1) and neurofibromatosis type 2 (NF2). NF1 is ten times more common than NF2. People with NF1 do not get NF2. These are two completely different conditions.

Neurofibromatosis Type 1 (NF1) is a common genetic condition. It affects approximately one in every 3000 people worldwide. That makes NF1 more common than cystic fibrosis and hereditary muscular dystrophy combined.

A diagnosis of NF1 is made following an extensive physical examination. Your NF doctors are looking for birthmarks (café-au-lait macules) and freckles in the armpits or groin areas. Other signs of NF1 include neurofibromas (benign tumors affecting nerves throughout the body), optic gliomas (brain tumors involving the optic nerve), and specific bone abnormalities.

Once a child or adult is diagnosed with NF1, your NF medical team closely monitors for specific signs and symptoms. Importantly, NF1 affects infants, school-age children, and adults differently. For this reason, young children and adolescents require yearly visits with an NF specialist. In adulthood, visits may become less frequent.

Although NF1 increases the risk of tumor growth, individuals are expected to generally live full and active lives. However, due to the complicated and unpredictable nature of NF1, you or your child should be cared for by a team of specialists, each an expert in the management of NF1. Every member of the team has a specific role. Together, your NF team aims to help you/your child obtain the highest level of health, function and quality of life.

My NF1

It is important that you have a clear understanding of NF1. You/your child have neurofibromatosis type 1 (NF1). There is another type of NF: neurofibromatosis type 2 (NF2). NF1 is ten times more common than NF2, and people with NF1 do not get NF2. These are two completely different conditions.

Your NF Team made the diagnosis of NF1 based on the following findings on your/your child's examination:

- Café-au-lait macules (birthmarks)
- Skin fold freckling (freckles in the armpits or groin areas)
- Neurofibromas (benign tumors affecting nerves)
- Plexiform neurofibroma (larger benign tumor affecting nerves)
- Lisch nodules (colored spots on the iris of the eye)
- Optic glioma (brain tumor affecting the nerve for vision)
- Specific bone problems
- A family member with NF1

In addition, we found:

- Learning or behavioral problems
- Attention problems
- Motor coordination problems
- Visual-motor delays
- Speech problems

Your Team Makes A Difference Today

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Your NF team is led by **Dr. David H. Gutmann**, an international specialist in NF with over twenty years of experience caring for children and adults with NF. Working closely with him is a nurse practitioner, Anne C. Albers.

As a scientist, Dr. Gutmann was one of the members of the research group that identified the entire NF1 gene. Dr. Gutmann also founded and leads the Washington University Neurofibromatosis Center (<http://nfcenter.wustl.edu>). The center includes doctors and laboratory scientists focused on accelerating the pace of scientific discovery and its application to the care of individuals with NF.

As a physician, Dr. Gutmann sees patients regularly at St. Louis Children's Hospital and actively trains new physicians. The focus of his practice is to support his patients in achieving the highest quality of life.

Ms. Anne Albers is a pediatric nurse practitioner with expertise in the care of individuals with NF. She works very closely with Dr. Gutmann to ensure that all possible services are provided to patients and their families. Anne also performs follow-up visits at more frequent intervals should the need arise.

Our goal is to provide excellent care that is:

- **Multi-disciplinary**
Many of your team members work in clinic with Dr. Gutmann. Most others are just down the hall. Although you may not need to see every specialist involved with the NF team, please know a wide range of experts are available to help you reach all your/your child's goals.
- **Compassionate**
We strive to provide support and resources to address all of your/your child's medical concerns.
- **Up To Date**
This unique physician and research center focuses on advancing knowledge and treatments for NF. All the members at SLCH and Washington University meet regularly to discuss current standards of care and exciting new developments. Through regular communication, our team is able to provide the most up to date and progressive treatment plans.
- **Family-centered**
You are your/your child's best advocate and champion. Do not hesitate to contact us with questions or concerns, no matter how large or small.

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As a part of our commitment to your care, we often provide you with referrals to specialists who are members of our multi-disciplinary team. To help you figure out what everybody on our team does, below is a list of our team members.

- **Pediatric Neurology Secretary** – When you call our main line, this is the person who will direct you to the appropriate resource or person.
- **Patient Care Coordinator** – This is your main contact at the St. Louis Children's Hospital NF Clinical Program. She works closely with Anne Albers to coordinate the complex care services for our patients and their families. During clinic hours, she meets with each family to provide screening materials and a summary of their individualized follow-up plan.
- **Physical Therapist** – We have a dedicated physical therapist. She assesses motor skills and identifies delays, allowing for prompt treatment. Additionally, she provides treatment and resources for our children, including outpatient therapy, school-based services and community-based services. You might also be referred to a physical therapist closer to home or through school.
- **Occupational Therapist** – These therapists assess how NF1 affects a person's ability to participate in age-appropriate daily activities. For example, a child with NF1 may have difficulty completing school work due to fine motor or attention deficits, getting ready in the morning (bathing, dressing, eating breakfast, getting to school/work) or enjoying a leisure or recreational activity (sports, reading, watching a movie).
- **Speech Therapist** – Children have important things to say, but sometimes they need extra help learning how to make sounds for certain letters or words. A speech therapist makes sure everyone can understand your child.
- **Neuropsychologist** – A neuropsychologist provides help understanding why your child might be having a hard time at school, keeping track of time, or sitting still like other kids. They will usually perform a battery of tests (in the form of games) and then recommend other services.
- **Psychologist** – Like the neuropsychologist, a psychologist might help understand where and why your child is having difficulties. They are also trained to provide services to help your child and your entire family with emotional support and counseling, individual or family therapy, behavioral techniques to manage pain, anxiety or reactions.

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- **Social Worker** – This team member provides emotional support and counseling to patients and families. They can also provide guidance and suggestions about community resources, financial assistance, lodging options, and/or transportation assistance.
- **Ophthalmologist** – Eye exams are an important part of routine care for children with NF1. Be sure to see an eye doctor we recommend.
- **Audiologist** – Ear exams are needed when your NF team suspects hearing or balance problems.
- **Plastic surgeons** – Some people choose to have their neurofibromas or plexiform neurofibromas removed. It is critical that you see a plastic surgeon with considerable expertise in the care of people with NF1.
- **Otolaryngologist** – This is a surgeon who specializes in problems affecting the ears, including hearing or balance problems.
- **Cardiologist** – These medical doctors care for people with heart problems.
- **Oncologist** – Although it is rare, some patients with NF1 get cancer. Oncologists are cancer doctors who deliver chemotherapy and treat cancer.
- **Surgical Oncologist** – These are doctors who specialize in removing tumors and cancers.
- **Neurosurgeons** – Neurosurgeons are surgeons who often work closely with oncologists to treat brain or spinal tumors.
- **Orthopedic Surgeon** – These are surgeons who perform operations to correct bone problems, like scoliosis or pseudarthrosis.
- **Endocrinologist** – The endocrine system is responsible for controlling the production of hormones in your body. An endocrinologist specializes in the treatment of children or adults with hormone deficiencies or excesses.
- **Pain Management** – These doctors manage pain using a combination of treatment methods.
- **Genetics** – A genetics counselor can talk to you about the chances of having children with NF1.

Making A Difference Today

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It was only in 1987 that the medical community agreed on how to reliably make a diagnosis of NF1. Three years later, a team of scientists found the gene responsible for NF1. Research continues to move forward at a rapid pace at the Washington University Neurofibromatosis Center (<http://nfcenter.wustl.edu>) and worldwide.

As a patient of a world-renowned clinical and research center, you have the opportunity to participate in one or several of our ongoing research projects. From genetics to behavioral studies, your contribution makes a real difference for the next generation of people with NF1.

If you are interested in participating, please let members of your NF team know.

Select Recent NF Center Publications

Banerjee S, Gianino SM, **Gao F**, Christians U, **Gutmann DH**. Interpreting mammalian target of rapamycin and cell growth inhibition in a genetically engineered mouse model of Nf1-deficient astrocytes. *Mol Cancer Ther.* 10:279-91, 2011.

Lee DY, Yeh TH, Emmett RJ, White CR, **Gutmann DH**. Neurofibromatosis-1 regulates neuroglial progenitor proliferation and glial differentiation in a brain region-specific manner. *Genes Dev.* 24:2317-29, 2010.

Dorward IG, Luo J, Perry A, **Gutmann DH**, Mansur DB, **Rubin JB**, **Leonard JR**. Postoperative imaging surveillance in pediatric pilocytic astrocytomas. *J Neurosurg Pediatr.* 6:346-52, 2010.

Brown JA, Emmett RJ, White CR, Yuede CM, Conyers SB, O'Malley KL, **Wozniak DF**, **Gutmann DH**. Reduced striatal dopamine underlies the attention system dysfunction in neurofibromatosis-1 mutant mice. *Hum Mol Genet.* 19:4515-28, 2010.

Warrington NM, Gianino SM, Jackson E, Goldhoff P, **Garbow JR**, **Piwnica-Worms D**, **Gutmann DH**, **Rubin JB**. Cyclic AMP suppression is sufficient

to induce gliomagenesis in a mouse model of neurofibromatosis-1. *Cancer Res.* 70:5717-27, 2010.

Brown JA, Gianino SM, **Gutmann DH**. Defective cAMP generation underlies the sensitivity of CNS neurons to neurofibromatosis-1 heterozygosity. *J Neurosci.* 30:5579-89, 2010.

Banerjee S, Byrd JN, Gianino SM, Harpstrite SE, Rodriguez FJ, Tuskan RG, Reilly KM, **Piwnica-Worms DR**, **Gutmann DH**. The neurofibromatosis type 1 tumor suppressor controls cell growth by regulating signal transducer and activator of transcription-3 activity in vitro and in vivo. *Cancer Res.* 70:1356-66, 2010.

Nunley KS, **Gao F**, **Albers AC**, Bayliss SJ, **Gutmann DH**. Predictive value of café au lait macules at initial consultation in the diagnosis of neurofibromatosis type 1. *Arch Dermatol.* 145:883-7, 2009.

Hegedus B, Hughes FW, **Garbow JR**, Gianino S, Banerjee D, Kim K, Ellisman MH, **Brantley MA Jr**, **Gutmann DH**. Optic nerve dysfunction in a mouse model of neurofibromatosis-1 optic glioma. *J Neuropathol Exp Neurol.* 68:542-51, 2009.

Albers AC, **Gutmann DH**. Gliomas in patients with neurofibromatosis type 1. *Expert Rev Neurother.* 9:535-9, 2009.

Yeh TH, Lee DY, Gianino SM, **Gutmann DH**. Microarray analyses reveal regional astrocyte heterogeneity with implications for neurofibromatosis type 1 (NF1)-regulated glial proliferation. *Glia.* 57:1239-49, 2009.

Houshmandi SS, Emmett RJ, Giovannini M, **Gutmann DH**. The neurofibromatosis 2 protein, merlin, regulates glial cell growth in an ErbB2- and Src-dependent manner. *Mol Cell Biol.* 29:1472-86, 2009.

Darken RS, Bogitch R, **Leonard J**, Perry A, **McKinstry RC**, **Gutmann DH**, **Rubin JB**. Brainstem glioma presenting as pruritus in children with neurofibromatosis-1. *J Pediatr Hematol Oncol.* 31:972-6, 2009.

Keeping Organized

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With so many people involved in caring for a child or adult with NF1, it is easy to become overwhelmed. It is also easy to lose sight of your family's goals and priorities. The purpose of this notebook is to help you organize your/your child's goals and aspirations, and to provide an invaluable resource to facilitate communication between your/your child's doctors, nurses, teachers, and therapists.

Things you can do to help keep your team organized:

- Make sure all medical releases are signed to allow your health care providers to talk about you/your child's health
- Request copies of medical records
 - Keep hospital discharge notes
 - Keep X-ray and MRI reports
 - Keep yearly NF Clinic notes
 - Keep vaccination records
- Keep therapy goals
- Keep copies of individual educational plans (IEPs)
- Keep running logs of medications and hospitalizations

Feel free to copy, add, delete, and reorganize the pages in this binder to make it work for you and your family. Electronic copies of these pages can be found online at <http://nfcenter.wustl.edu>.

Contacting My Team

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Please call at any time with any concerns you have about yourself/your child. You know yourself/your child best and can tell when something is “just not right.” Depending on your questions, you may want to talk to your/your child’s doctor or pediatric nurse practitioner. We are **all** here to help your child and family through treatment, both physically and emotionally. Please let us know what we can do to help!

When calling the doctor after hours, always identify:

1. yourself
2. your/your child’s name and age
3. your/your child’s condition (neurofibromatosis type 1, NF1)
4. your/your child’s symptoms or new problem

This information allows the doctor on-call to best address your needs in a timely manner.

Scheduling/General NF questions:

Monday through Friday	314-454-6120
<i>8 a.m.-4:30 p.m. CST</i>	
Evenings/Holidays/Weekends	314-454-6120
<i>Ask for the Pediatric Neurologist on call</i>	
Toll-free	888-503-2237
<i>Ask to be transferred to the NF Team in Pediatric Neurology</i>	

Contacting My Team

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NEUROLOGIST	David Gutmann, MD, PhD	phone	314-454-6120
fax	314-454-2523	address	One Children's Place, St. Louis, MO 63010
website	http://nfcenter.wustl.edu	e-mail	gutmann@neuro.wustl.edu
NURSE PRACTITIONER	Anne Albers, CPNP	phone	314-454-6120
fax	314-454-2523	address	One Children's Place, St. Louis, MO 63010
website	http://nfcenter.wustl.edu	e-mail	albersa@neuro.wustl.edu
NF COORDINATOR	Vicki Kuchnicki	phone	314-454-6120
fax	314-454-2523	address	One Children's Place, St. Louis, MO 63010
website	http://nfcenter.wustl.edu	e-mail	NFClinic@neuro.wustl.edu
EYE DOCTOR		phone	
fax		address	
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CANCER DOCTOR		phone	
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SKIN DOCTOR		phone	
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BONE DOCTOR		phone	
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NEURO- PSYCHOLOGIST		phone	
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PEDIATRICIAN		phone	
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OCCUPATIONAL THERAPIST		phone	
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Contacting My Team

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My Phone Calls

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Date call initiated	Question/concern	Recommendation/response
Contact person/facility		Date completed
Date call initiated	Question/concern	Recommendation/response
Contact person/facility		Date completed
Date call initiated	Question/concern	Recommendation/response
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Date call initiated	Question/concern	Recommendation/response
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