



NF Clinic Network (NFCN) Application Form*

Clinic Name:

Duke University Neurofibromatosis Clinic

Affiliated Hospital:

Duke University Medical Center

Affiliated University or Institution:

Duke University

Clinic Address:

Duke University Neurofibromatosis Clinic
DUMC 3936
Durham, NC 27710

Clinic Director:

Fawn Leigh, MD

Clinic Coordinator Name:

Elizabeth Rende

**Note: Some non-public information has been removed from this application form.*



The Neurofibromatosis Clinic Network (NFCN)

FORM PART A: Affiliate Clinic Application

1. ABOUT YOUR NF CLINIC

a. Is your NF Clinic:

- Freestanding
- Hospital based
- In an academic center
- Other (please describe)

b. Describe overall your NF Clinic, when it meets and how it functions.

The Neurofibromatosis clinic at Duke provides multidisciplinary clinical care for pediatric and adult patients with neurofibromatosis. This clinic was established in January 2007 as the first multidisciplinary neurofibromatosis clinic at Duke University Medical Center. The clinic currently meets the third Thursday of each month, and we are in the process of expanding it to the first & third Thursday of the month due to increasing patient volume. The clinic is located in the McGovern-Davison Children's Health Center, a state-of-the-art facility adjacent to Duke Hospital and is in close proximity to imaging resources of the Medical Center, including computed tomography (CT), magnetic resonance imaging (MRI), and positron emission tomography (PET). The clinic is also in close proximity to the neurophysiology lab where

electroencephalograms (EEG), evoked potentials and electromyograms (EMG) are performed. The Duke Neurofibromatosis Clinic is dedicated to outpatient medical and surgical specialty care to individuals with neurofibromatosis, type 1 and type 2.

Each patient is individually assessed by a triaging process. Patients are seen in the appropriate specialties and are scheduled for tests as needed. Physicians in the Duke Neurofibromatosis Clinic are familiar with and practice in accordance with the recommended consensus guidelines for Neurofibromatosis Clinical Care. At the end of the clinic day, the Neurofibromatosis team consisting of physicians, clinic coordinator, social worker and physical therapist meet to discuss patients seen in the Duke Neurofibromatosis Clinic. The meeting is an opportunity for all providers to communicate with each other about their assessment and the information discussed with the patients during their clinic visit. The comprehensive medical plan for patients is formulated as a group during this team meeting. The coordinator plays an integral part in carrying out the plans as discussed, i.e. scheduling tests, clinic appointments, and additional referrals and serving as a liaison between the patient and providers.

2. CLINIC DIRECTOR and STAFF EXPERTISE

a. CLINIC DIRECTOR: Please describe:

i. Your experience to date with Neurofibromatosis care

I have cared for neurofibromatosis patients since 1995, while attending the Pritzker School of Medicine at the University of Chicago. I continued to participate in their care as a resident and fellow at the University of Chicago until June 2005 when I was recruited to the Division of Pediatric Neurology at Duke University Medical Center. At Duke University, I established the Duke Neurofibromatosis Clinic in January 2007.

I can vividly remember the first patient for whom I cared for with neurofibromatosis. I was impressed by the complexity of the condition and, I was intrigued by the unknowns. As I continued in my medical education and training at the University of Chicago, I found myself drawn to this patient population, primarily from a neurological perspective and because of the multi-system involvement. As a participating member of the Neurofibromatosis Clinic at the University of Chicago, I gained greater perspective of the genotypic and phenotypic variability in neurofibromatosis. As a medical student, I observed and listened carefully to each patient's story and was deeply touched by how this disease affected the lives of these patients and their families. As the resident on call, I was the first one to see these patients in the emergency room and to address their acute medical needs, which varied in complexity from seizures, to increased intracranial pressure, to weakness and pain. In my training in pediatric neurology, I also trained in adult neurology and served as the adult neurology resident on call, where I was the only person with

experience in taking care of patients with neurofibromatosis. I have personally received many “thank-you’s” from adult patients with neurofibromatosis, which has left an indelible impression in me. It was clear that there is a need to better serve adult patients. As a faculty member in the Department of Pediatrics and in the Department of Medicine at Duke, I am committed to teaching the evolving story about neurofibromatosis to medical students and house officers training in Pediatrics and in Medicine, providing them with experience in the medical and surgical management of neurofibromatosis through their participation in the Duke Neurofibromatosis Clinic at Duke.

Over the years, I have made it a priority to educate myself as well as others about neurofibromatosis. I have familiarized myself with the consensus guidelines for clinical care in neurofibromatosis as early as 1997 and have continued to follow the medical literature on neurofibromatosis. I have attended the International Neurofibromatosis Consortium Meetings in 2006 and 2007, which has been a venue to discuss and share ideas with the thought leaders in neurofibromatosis from around the world. Additionally, I have also attended regional meetings of Neurofibromatosis, Inc. I have given numerous lectures to patients, medical students, house officers, faculty and scientists about neurofibromatosis over the past ten years. More recently, I gave a Neurology Grand Rounds lecture on neurofibromatosis, type 1, focusing on issues affecting adult patients such as neuromuscular and cardiovascular complications. Other lectures have included topics relating to learning disabilities, central nervous system tumors, cranial nerve palsy, and genotypic and phenotypic variability in neurofibromatosis. As a faculty member in the Division of Pediatrics and in the Division of Medicine, I will continue to serve as an educator to others about neurofibromatosis. My audience has recently included scientists in neuroscience, in addition to clinical staff.

I began my medical career with a few questions about neurofibromatosis and I have, expanded that list of questions greatly over time. In many ways, neurofibromatosis has driven my clinical and research interests. The establishment of the Duke Neurofibromatosis Clinic is the first step in my quest to find answers and to better serve this patient population. I am deeply committed to building a neurofibromatosis program which provides excellent medical and surgical care to individuals with neurofibromatosis, type 1 and type 2.

- ii. Your past and current association with Neurofibromatosis clinical trials.

As a clinician-scientist early in my career development, I hope to build a neurofibromatosis program at Duke University Medical Center that will be a center for upcoming neurofibromatosis clinical trials. Many resources at Duke facilitate clinical research, including the Duke Center for Clinical Research. Also, I will be enrolling in the Masters Degree Program in Clinical Research at Duke, a curriculum that will further equip me with the skills and tools to be a successful clinical investigator, including directing clinical trials.

Currently, I am focused on building the infrastructure for the Duke Neurofibromatosis Clinic. A solid infrastructure will enable the neurofibromatosis program to provide quality care to patients and families with neurofibromatosis, thereby providing an environment for basic and clinical research as well as patient advocacy.

- iii. Your past and current association with other clinical trials e.g. oncology trials

Dr. Leigh is the Principal Investigator for two clinical trials for Topamax, an antiepileptic drug in its use for refractory seizures and for Tourette's Syndrome.

Dr. Gururangan (pediatric neuro-oncologist) and Dr. Rich (adult neuro-oncologist), two members of the Neurofibromatosis team, are faculty members of the Preston Robert Tisch Brain Tumor Center at Duke University Medical Center. This brain tumor center is one of the largest and most successful in the field and has received the highest rating of "outstanding" by a National Cancer Institute peer review group's evaluation of the Duke Comprehensive Cancer Center over the last ten years. Dedicated entirely to the treatment and cure of brain and spinal tumors in children and adults, the Brain Tumor Center at Duke University Medical Center combines the resources of a leading research hub with a commitment to the best in patient care. Dr. Gururangan's research interest focuses on finding novel chemotherapeutic strategies for the treatment of children and young adults with brain tumors. Since his tenure at the Brain Tumor Center, he has written several clinical protocols. Dr. Rich has clinical and research interests in new therapeutic strategies targeting signal transduction pathways present in primary brain tumors in adults.

- b. CLINIC DIRECTOR: Please provide information on:

- i. Present and past funding you have received for Neurofibromatosis research. Include funding source, date received, amount and project description.

As a clinician-scientist who is in early career development and has recently started a neurofibromatosis program at Duke, I am proud of the success in collaborations in clinical care. I have submitted a grant application, currently to use neuroproteomics as an approach to identify differentially expressed proteins from serum in patients with neurofibromatosis, type 1. Additionally, two other grant applications are in the development stage and will be submitted in the near future.

- ii. Your Neurofibromatosis-related clinical and scientific publications.
Include Journal, Citation and Title.

I am, currently, early in my career, but I am deeply committed to expanding my clinical and research efforts toward neurofibromatosis, efforts that stem from broad experience in taking care of patients with neurofibromatosis. The initiative to establish and build the Duke Neurofibromatosis Clinic is the first of many accomplishments to come in better understanding neurofibromatosis, and ultimately in better serving this patient population. I am committed to the advancement of clinical and scientific efforts to the identification of therapeutic targets, development of treatment options and development of biological markers for the condition, as well as its associated complications. Currently, the following manuscripts are in preparation:

1. Linear dynamics of neonatal seizures. (In collaboration with Dr. Kurt Hecox, Pediatric Neurology, University of Chicago)
2. Atypical clinical presentations of opsoclonus myoclonus. Multi-center case review. (In collaboration with Dr. Charles Maxfield, Dept of Radiology at Duke University).
3. Case report of a 7 year old girl with Neurofibromatosis1 with granular cell carcinoma.
4. Retrospective review of neurofibromatosis, type 1 patients who have spinal neurofibroma.
5. Case review of two neurofibromatosis, type 1 infants with aggressive optic gliomas.

c. Who are the key staff in your Neurofibromatosis clinic facility?
Provide Name; Title; Degree/Qualifications; Role in Clinic.

Please see table and narrative about the Duke Neurofibromatosis Clinic on the following page.

<i>Physician Name</i>	<i>Degree</i>	<i>Specialty</i>	<i>Areas of Expertise</i>	<i>Phone# Area code (919)</i>	<i>Address DukeUniversityMedicalCenter Box (see# below) Durham, NC 27710</i>
Fawn Leigh	MD	Neurology (pediatric & adult)	Seizures, paresthesias, weakness, developmental delay, headache	668-0477	3936
Elizabeth Rende	RN, MSN, CPNP.	Pediatric Neurology Nurse Practitioner	Clinic Coordinator	668-0477	3936
Ronald Kanter	MD	Cardiology	Vascular Dx, Congenital Heart Defect	681-6772	3090
Marie McDonald	MD	Genetics (pediatric & adult)	Cancer genetics	681-982	3582
Gordon Worley	MD	Neurodevelopment (pediatrics)	Autism	684-3173	3120
Jeremy Rich	MD	Neuro-Oncology (adult)	Brain tumors	681-1693	3624
Sridhuran Gururangan	MBBS, MRCP.	Neuro-Oncology (pediatric)	Brain tumors	684-3506	3624
Debara Tucci	MD	Neuro-Otology (pediatric & adult)	Acoustic Neuroma	684-6968	3805
Melanie Bonner	PhD	Neuropsychology	Learning Disabilities	416-2443	3527
Herb Fuchs	MD, PhD	Neurosurgery (pediatric)	CNS & PNS tumors	684-5013	3272
Gerry Grant	MD	Neurosurgery (pediatric & adult)	CNS & PNS tumors	684-5013	3272
Corinne Linardic	MD	Oncology	Sarcoma	684-3401	2916
Terri Young	MD	Ophthalmology (pediatric & adult)	Ophthalmic genetics	684-0584	3802
Jeff Marcus	MD	Plastic/Craniofacial (pediatric & adult)	Facial reanimation, craniofacial reconstruction,	668-3110	3974
Jan Fitch		Physical Therapy		684-3730	3120
Charles Maxfield	MD	Radiology		684-7585	3808
Sheila Rittgers		Social Work		684-4209	3037

d. Who within this core staff currently coordinates Neurofibromatosis patient services?

Describe this individual's Neurofibromatosis clinic related duties.

Elizabeth Rende is the Clinic Coordinator under my direction. She is a nurse practitioner in the division of pediatric neurology and has extensive experience in patient care. Liz serves as the liaison between the patient and the physicians. When a referral is received, Liz is the first person to contact the patient to assess the medical issues and for any acute medical needs. Once this communication has taken place, Liz facilitates the coordination of clinic appointments and tests in advance of the clinic visit. Prior medical records are obtained, including clinic notes, test results and/or copies of neuroimaging for further review with Dr. Leigh and Radiology. Liz is the first and constant line of communication with the patients and coordinates communication with the physicians. As patients are seen in clinic, Liz directs the flow of specialty evaluations by guiding patients to their designated appointment(s). The Neurofibromatosis team meets at the end of the clinic day to communicate and share information about the patients seen in clinic today. This meeting is a venue for all the providers to convey their assessments and recommendations. The comprehensive medical and/or surgical plans are formulated during the Neurofibromatosis team meeting. Liz is present during the Neurofibromatosis team meeting. She will summarize and communicate these plans with the patients and families as she facilitates the implementation of the recommendations provided by the Neurofibromatosis team. She keeps track of all tests results and communicates those findings with the patients. More importantly, Liz ensures that the medical plan is carried out as discussed.

Liz received her Master's Degree in Nursing from Duke University and has over 30 years experience as a nurse. She has functioned as a Pediatric Neurology Nurse Practitioner since 2000 and is a Clinical Faculty Associate in the Duke School of Nursing. Liz also has prior experience as coordinator for a clinical trial with Keppra, an antiepileptic drug at Duke and can help to facilitate patient enrollment in clinical trials. She is also a sub-PI in a clinical trial with Topamax (antiepileptic drug), where I am the Principal Investigator. We work closely together daily, thereby facilitating fluid communication in patient care issues.

At present, Liz serves in this role part-time due to limited funding. Upon receipt of funding for the Neurofibromatosis Clinic Coordinator, Liz will take a more active role on a daily basis. Protected time to the Neurofibromatosis clinic will enable further growth of the clinic and Neurofibromatosis related projects.

e. Describe any areas of Neurofibromatosis care in which your clinic has particular expertise (e.g. optic glioma, vestibular schwannoma, bone manifestations, learning disabilities etc.) and the clinic staff that provide this care.

Please refer to the table above.

There are many distinguishing features to the Duke Neurofibromatosis Clinic. The first of these is that this multidisciplinary clinic serves neurofibromatosis, type 1 and type 2 patients. Another key feature is that the members of the Neurofibromatosis team see pediatric and adult patients, hence our patients receive specialized medical care for Neurofibromatosis and its associated complications across the age continuum. The following details the expertise and experience of some of our physicians, who are familiar with the recommended consensus guidelines for neurofibromatosis clinical care and management. The Neurofibromatosis team is committed to providing appropriate care to the individuals with neurofibromatosis, using the current consensus guidelines and to contributing to the ongoing evolution of consensus guidelines as we learn more about the natural history of neurofibromatosis and its multi-system involvement.

Neurology:

Dr. Fawn Leigh is a pediatric neurologist with extensive clinical experience in the clinical care of patients with neurofibromatosis. Her expertise spans a wide spectrum of neurological issues, including seizures, developmental delay, headache, paresthesias, weakness, complications of tumors in the brain and spinal cord, and others. Dr. Leigh works in the multidisciplinary setting on a daily basis in the care of patients with neurological complications. With the neurofibromatosis patient population, she is committed to the advancement of better treatment options and clinical research. She collaborates with scientists at Duke University in studying biological markers in neurofibromatosis and its associated complications. She is a faculty member in the Department of Pediatrics and the Department of Medicine at Duke University Medical Center. Dr. Leigh sees pediatric and adult patients in the Duke Neurofibromatosis Clinic.

Genetics:

Dr. Marie McDonald is a clinical geneticist. Her research includes cancer genetics and she has followed many patients at Duke with neurocutaneous disorders. The division of genetics has pioneered enzyme replacement for the treatment of Pompe disease, a genetic disorder due to deficiency of an enzyme called acid α -glucosidase (GAA), resulting in progressively debilitating and often fatal muscle disorder. The collaboration led by YT Chen and Priya Khrishnani in the Department of Pediatrics has culminated in the successful development of the recombinant enzyme, Myozyme which has been shown in clinical trials to

reverse symptoms and was approved by the FDA in April 2006. This experience in drug development partnered with clinical care is an excellent model and resource for further investigations in neurofibromatosis. Dr. McDonald sees pediatric and adult patients in the Duke Neurofibromatosis Clinic.

Neuro-Oncology:

Dr. Jeremy Rich is a neuro-oncologist with a clinical interest in primary brain tumors in adults and with research interests in laboratory and clinical trial design of new therapeutic strategies targeting signal transduction pathways. Recent research in Dr. Rich's laboratory includes: 1) responses to transforming growth factor- β shift towards tumor suppression in the presence of the PTEN tumor suppressor gene, 2) targeting DNA damage checkpoint signaling in glioma radioresistance, and 3) the roles of secreted protein acidic and rich in cysteine (SPARC) in brain tumor invasion. Dr. Rich is a faculty member in the Preston Robert Tisch Brain Tumor Center at DUMC, which is one of the largest and most successful in the field. It has received the highest rating of "outstanding" by a National Cancer Institute peer review group's evaluation of the Duke Comprehensive Cancer Center over the past ten years. Dr. Rich sees adult patients with brain tumors.

Dr. Sridharan Gururangan is a pediatric neuro-oncologist. His research interest focuses on finding novel chemotherapeutic strategies for the treatment of children and young adults with brain tumors. He has significant experience in the treatment of optic gliomas with carboplatin in neurofibromatosis, type 1 patients. Since his tenure at the Brain Tumor Center, he has written several clinical protocols. Dr. Gururangan is also a member of the Pediatric Brain Tumor Consortium. Dr. Gururangan sees pediatric patients in the Children's Health Center.

ENT/Neuro-Otology:

Dr. Debara Tucci is a neuro-otologist with extensive experience in the otologic, neuro-otologic and skull base surgery of patients of all ages. Dr. Tucci is a highly skilled surgeon and has extensive experience in the removal of acoustic neuromas in Neurofibromatosis, Type 2. Her research interests includes studying issues associated with cochlear implant and defining the effects of hearing impairment on the development and function of the central auditory system.

Neuropsychology:

Dr. Melanie Bonner is a clinical child psychologist who has expertise in working with children with learning disabilities and ADHD. She is a member of the Department of Psychiatry and Behavioral health. Dr. Bonner and her team frequently perform a battery of tests to assess the cognitive and social function in pediatric patients. She has seen neurofibromatosis patients for many years.

Dr. Bonner works closely with psychiatry, particularly in the management of ADHD. Her current research efforts focus on psychological adjustment in children and families who deal with the stress and strain of chronic illness. Current projects include: 1) an NIH funded project aimed at assessment of biological substrates of learning impairments in pediatric cancer survivors and evaluation of a pharmacological intervention to ameliorate those impairments; 2) a project funded by the Duke Institute for Care at the End-of-Life which involves the development and validation of a new measure of coping and loss for parents of pediatric cancer patients; 3) evaluation of social functioning in pediatric cancer patients including assessment of social cognition and peer relations; and 4) evaluation of quality of life parameters in patients with sickle cell disease. Her experience in patients with chronic illness translates into benefits to neurofibromatosis patients.

Neurosurgery:

Dr. Gerald Grant is a pediatric neurosurgeon. His experience spans from brain tumors to spinal disorders. Dr. Grant sees pediatric and adult patients in the Duke Neurofibromatosis Clinic.

Dr. Herbert Fuchs is a pediatric neurosurgeon. His research interests include collaborations studying molecular genetics of childhood brain tumors and preclinical studies of new therapeutic agents with the neuro-oncology laboratory of Dr. Darrell Bigner at Duke University. Dr. Fuchs sees pediatric patients in the Duke Neurofibromatosis Clinic and will continue to follow these patients into adulthood.

Ophthalmology:

Dr. Terri Young is a pediatric ophthalmologist with expertise in ophthalmic genetics. She serves on the Association for Research in Vision and Ophthalmology (ARVO) Genetics Advisory Group and the American Academy of Ophthalmologic Pediatric Ophthalmology Program Committee. She is the Director of the Ophthalmic Genetics Pediatric Clinic. Dr. Young is an NIH-funded researcher for the “International Collaborative Twin Study of Refractive and Glaucoma Endophenotypes.” Dr. Young sees pediatric and adult patients in the Duke Neurofibromatosis Clinic.

Plastic & Reconstructive Surgery:

Dr. Jeffrey Marcus is the section chief of pediatric and reconstructive surgery. His experience spans from craniofacial conditions, reanimation of facial paralysis, and post-traumatic facial abnormalities. Dr. Marcus has experience with neurofibromatosis patients in the removal of dermal and subcutaneous plexiform neurofibromas. Dr. Marcus’ expertise in facial nerve reanimation will positively enhance services available to neurofibromatosis patients with facial

nerve paralysis. Dr. Marcus sees pediatric and adult patients in the Duke Neurofibromatosis Clinic.

3. PATIENT SCHEDULING and REFERRALS

- a. Provide the details of the 'typical' timeframe in which patients receive a response to a request for scheduling, are actually scheduled for an appointment, how patients are prioritized, etc.\

Patients are scheduled for appointments in the Duke Neurofibromatosis Clinic within 2-3 weeks of contacting the scheduling office. The neurofibromatosis team is committed to providing optimal care to this patient population and will see urgent cases immediately if needed, even if it is not on a dedicated Neurofibromatosis clinic day. Dr. Leigh works closely with all the specialists on a daily basis and, hence communication regarding medical management issues occurs fluidly. Typically, upon receipt of a request for an appointment, the Clinic Coordinator will contact the patient/parent and/or referring physician within 24 hours to further discuss the medical issues involved. We request prior medical records and studies (i.e. neuroimaging, EEG, EMG, general labs, neuropsychological assessments, etc) that have been performed to date for review. Following review of this information, any further testing needed is coordinated with the upcoming clinic visit. The Clinic Coordinator forwards all appropriate medical information to the physicians and communicates all plans with the patient. An information packet is mailed to patients. This packet includes all appointments, tests, names of physicians, driving directions, parking map and a Neurofibromatosis questionnaire.

- b. Provide details of those specialists to whom (either within or outside our own clinic facility) your clinic refers Neurofibromatosis patients for the following specialty care. **These should be individuals familiar and experienced with consensus guidelines for care of individuals with Neurofibromatosis** (Please provide information for PEDIATRIC CARE referrals in the first table and ADULT CARE in the second table).

Please refer to the table above and discussion of physician expertise as written above. The Duke Neurofibromatosis Clinic is designed to see both pediatric and adult patients. More specifically, Dr. Leigh (Neurology), Dr. Grant (Neurosurgery), Dr. McDonald (Genetics), Dr. Young (Ophthalmology), Dr. Marcus (Plastic & Reconstructive Surgery), and Dr. Tucci (Neuro-Otology) all see pediatric and adult patients. Dr. Gururangan sees pediatric brain tumor patients and Dr. Rich sees adult patients with brain tumors. Dr. Bonner (clinical child psychologist) will see children with cognitive and/or behavioral issues.

4. NUMBER OF Neurofibromatosis PATIENTS YOUR CLINIC SEES

a. How many Neurofibromatosis PATIENTS did you see in the past 12 months?

The first multidisciplinary Neurofibromatosis Clinic at Duke began Jan 18, 2007 and 62 Neurofibromatosis patients have been seen in the past 5 months.

b. How many of these were **NEW** patients to your clinic?

Approximately 50% of the patients seen in the Duke Neurofibromatosis Clinic in the past 5 months were new patients to this clinic.

	Neurofibromatosis1	Neurofibromatosis2	SCHWANNOMATOSIS	OTHER
NUMBER OF PATIENTS SEEN IN SINCE JANUARY 2007 (PAST 5 MONTHS)	60	2	0	0
NUMBER OF NEW PATIENTS SEEN IN SINCE JANUARY 2007 (PAST 5 MONTHS)	30	1	0	0
TOTAL	60	2	0	0

****There are 709 individual patients with Neurofibromatosis at DUMC. (See below) We have performed a de-identified internal query at Duke University Medical Center to assess the number of patients with a diagnosis of Neurofibromatosis by ICD 9 codes.**

The de-identified query in the pediatric database (i.e., ages 0-20) reveals 560 individual patients. The de-identified query in the adult neurology database (i.e., ages 21 and older) reveals 149 Neurofibromatosis patients. Dr. Leigh has a secondary appointment with the Department of Medicine in the Division of Neurology. These adult patients are seen in the Duke Neurofibromatosis Clinic with Dr. Leigh and the Neurofibromatosis team. The following chart shows the breakdown by age groups.

Patient Ages	# of Patients
0-2	53
3-5	93
6-8	88
9-11	82
12-14	68
15-17	121
18-20	55
21 & older	149
Grand Total	709

Hence, there are 709 patients in the Duke University Medical Center based on these recent de-identified searches. Efforts are in progress to recruit these patients to the Duke Neurofibromatosis Clinic. We are in the process of creating a web page and links to the Duke Neurofibromatosis Clinic. Also, we will direct efforts to recruiting patients from nearby regions in North Carolina, South Carolina, Virginia and Tennessee. Duke University Medical Center is in a strategic location to serve as the center of neurofibromatosis excellence in the southern states. Importantly, the expertise and resources to provide specialized care and to carry out state-of-the-art research can all happen at Duke.

- c. Overall what proportion of patients seen in the since January 2007 (over the past 5 months) were (give finite numbers if these are available, or estimate percentage):

62 patients have been seen in clinic over the past 5 months in the Duke Neurofibromatosis Clinic. Of these 62 patients, 52 patients were under 18 years of age and 10 patients were older than 18 years of age. The estimate percentage of patients under 18 years of age is 84% and the estimate percentage of patients over the age of 18 is 16%.

Patient Ages	# of Patients	% of Patients
Under 18	52	84
18+	10	16

5. TRANSITIONING PEDIATRIC TO ADULT Neurofibromatosis CARE

How does your clinic facilitate continuity of care for patients transiting from pediatric to adult care?

Explain how continuity of care is accomplished. Describe those partnering clinics with which you coordinate services, and explain any limitations:

The clinical core group for the Duke Neurofibromatosis Clinic sees adult and pediatric patients, thereby providing specialized care and continued care to all neurofibromatosis patients in the Duke Neurofibromatosis Clinic.

6. INTERNAL Neurofibromatosis CONFERENCES

Provide details on internal conferences in your institution which are related to Neurofibromatosis patient care in your clinic (e.g. Neurofibromatosis Clinic case management conference, etc.)

The Neurofibromatosis team meets at the end of the day to discuss the cases seen in the multidisciplinary clinic. This meeting is held in a conference room equipped with audio-visual equipment with access to the

patient medical record and database. This facility also enables the viewing of imaging studies and review of test results. The imaging studies, are reviewed with the guidance of Dr. Charles Maxfield, the radiologist in the neurofibromatosis program. The Clinic Coordinator participates in this clinic as the key liaison between the physicians and the patients. Importantly, the Clinic Coordinator facilitates the communication and implementation of the recommended treatment and investigative plans discussed during this meeting.

7. CLINICAL TRIALS

Our clinic is willing and able to provide our Neurofibromatosis patients with information on, and to facilitate their participation in, clinical trials for which Neurofibromatosis patients are eligible (check box)

Yes No

Do you currently refer patients to clinical trials?

Yes No

If 'yes', provide details of current clinical trial protocols in which you currently or have had patients involved in the past 5 years.

As a newly established Neurofibromatosis clinic which began seeing patients five months ago, there is one pediatric patient who is currently enrolled in the FTI clinical trial at NCI. Recently, two adult patients have been provided with information regarding the AZD2171 clinical trial at Mayo Clinic.

We anticipate that Duke will be a center for Neurofibromatosis clinical trials in the future. Dr. Leigh is committed to clinical research and will begin a Masters Degree program in clinical research next month. She will be trained in study design, statistics, and clinical trial management. Currently, Dr. Leigh is the Principal Investigator in two clinical trials with Topamax (an antiepileptic drug). Liz Rende, Clinic Coordinator, also has experience as a coordinator for a clinical trial in the recent past with Keppra, and she also is a sub-PI on the Topamax clinical trial with Dr. Leigh. Furthermore, Duke University, in particular the Duke Clinical Research Institute, has world class experience in clinical trials and the resources are available to Dr. Leigh and the Neurofibromatosis team. Collaborations with the Duke Cancer Center will also positively augment clinic trials at Duke.

8. PATIENT REGISTRY

Do you currently have an Neurofibromatosis specific patient database/registry?

Yes No

If 'yes', please describe.

Patients seen in the Duke Neurofibromatosis Clinic are enrolled in the Duke Neurofibromatosis Registry. This longitudinal database provides pertinent data points for further analysis in understanding the natural history, the frequency of expected complications, and the identification of under-recognized complications. Data points include information extrapolated from the history of present illness, review of systems, developmental history, intellectual/academic history, family history, socioeconomic information, and physical exam, with additional data points for laboratory and radiological findings. The Clinic Coordinator maintains this database.

Would you be willing to transfer this data to a centralized CTF Neurofibromatosis Database?

Yes No

We will transfer this data to the centralized CTF Neurofibromatosis Database in accordance with the guidelines and regulations of the Duke University Medical Center Ethics and Research Committee.

9. PUBLICATIONS and RESEARCH (IF APPLICABLE)

a. Please list any relevant Neurofibromatosis publications from your clinic in the past 5 years.

Include Journal, Citation and Title.

This is a newly established clinic that began seeing patients on January 18, 2007. Our database of patients is growing, and our collaborations will surely result in relevant Neurofibromatosis publications in the upcoming months.

b. Please provide information on Neurofibromatosis-related research ongoing in your clinic or performed by personnel affiliated with your clinic.

Dr. Leigh has collaborations with the Duke Neuroproteomics Lab and has written two grant proposals with the lead scientist of the Neuroproteomics lab to perform comparative analysis of paired samples in studying Neurofibromatosis-1, using two-dimensional difference in gel electrophoresis (2D-DIGE), which will provide quantitative protein analysis, combined with protein identification by tandem mass spectrometry.

10. PATIENT SUPPORT

Do you have an Neurofibromatosis patient support group that meets in association with your Neurofibromatosis Clinic?

If 'yes' provide details.

If 'no', are you interested in starting such a group?

In the near future, when the clinic has a full time coordinator and as our patient population grows, I think that a local group will serve an important need to our patient population regionally. At the moment, the focus is on building the infrastructure which provides an environment for quality patient care, research and advocacy.

What resources would help you to do this?

A Clinic Coordinator serving as a liaison between the Duke Neurofibromatosis Clinic and advocacy.

Funding for office space for the director of the local Neurofibromatosis group.

Funding for a neurofibromatosis education day directed to patients with Neurofibromatosis and their families. This will also serve as a venue for the community to come together and work towards building a local support network regionally.

Educational materials that can be mailed to patients.

11. OTHER INFORMATION

Please provide any additional information that is pertinent to your request to join the CTF Neurofibromatosis Clinic Network.

The Duke Neurofibromatosis Clinic is committed to providing excellent medical and surgical care to individuals with neurofibromatosis. It is designed to serve pediatric and adult patients in the same clinic. Patients in the Duke Neurofibromatosis Clinic receive comprehensive specialized and ongoing care. Furthermore, the building of an infrastructure to support quality patient care is paired with an environment for basic and clinical research of the utmost importance. The skill, experience, facilities and resources at Duke University strengthen the interdisciplinary efforts of the neurofibromatosis program. A CTF-Neurofibromatosis Clinic Network(CN) Clinic Coordinator, as an integral member of the Neurofibromatosis team, will further endorse the quality of care to our patients as well as positively augment the communication and interaction among patients, families, providers, other Neurofibromatosis clinics in the region, the Children's Tumor Foundation, and local advocacy groups.