



NF Clinic Network (NFCN) Application Form*

Clinic Name:

University of Utah NF Clinic

Affiliated Hospital:

Primary Children's Medical Center

Affiliated University or Institution:

University of Utah

Clinic Address:

University of Utah
2C412 School of Medicine
50 North Medical Drive
Salt Lake City, UT 84132

Clinic Director:

David Viskochil, MD, Ph.D.

Clinic Coordinator Name:

Carol Hsieh

**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.

Our NF Clinic meets on an ongoing basis at either Primary Childrens Medical Center (Tuesday Clinic) or the adjacent State Health Department Clinic (Wednesday Clinic). We have not set aside a specific day for NF Clinic, and we no longer have multiple subspecialists participate in a specific clinic day. We try to bring new, urgent cases into the regularly scheduled clinic times either on Tuesday or Wednesday morning. In general, patients with known NF or multiple café-au-lait spots will be seen by either Dr. Viskochil or Dr. Carey; however, if neither is available then they would be seen by one of the other clinical geneticists at the University of Utah who would then transfer care to one of the co-directors. Dr. David Stevenson also sees NF patients on a routine basis, however he has protected time to perform research related to NF1 bone health. In some instances, adolescent females prefer to be seen by Dr. Susan Lewin, one our clinical genetics colleagues. Follow-up cases are

scheduled approximately 1 year from previous consultation, and they are intermixed with our regular clinic. We have a number of selected subspecialists who are considered part of our NF Clinic team, and patients have priority scheduling in their respective clinic if a medical complication should arise for one of our patients. Otherwise, NF patients are seen routinely in their regularly scheduled clinics.

Routine encounters focus on age-related issues, but all include assessment of growth, blood pressure, updating medical history, review of school performance, assessment of development and behavior, review of systems, and physical examination. A genetic counselor attends almost all visits and will explore issues related to reproductive risk, inheritance pattern, and pedigree analysis of associated conditions in both affected and unaffected family members.

Letters regarding significant findings of the clinic encounter are sent to the patients' primary care provider and subspecialists involved in their respective care. The clinic coordinator does not participate in additional scheduling or coordination of multispecialty care, but she will maintain ongoing follow-up clinic appointments.

2. CLINIC DIRECTOR and STAFF EXPERTISE

a. CLINIC DIRECTOR: Please describe:

i. Your experience to date with NF care

Dr. Carey has been following NF patients in the Intermountain west since he came to the University of Utah in 1979. Dr. Viskochil began caring for NF families during his clinical fellowship at the University of Utah. The NF Clinic was developed in the early 1990's as a multidisciplinary clinic with discussion of cases after the clinic. This clinic changed to accommodate more patients, and relieve subspecialists of a dedicated time slot that did not correspond to encounters that encompassed comprehensive care for families who had different needs. Our team of clinical geneticists and genetic counselors have been following NF patients on an ongoing basis for over 20 years.

Dr. Carey was a member of the NNFF Clinical Care Advisory Board, and served as Co-Chair for a number of years. Likewise, Dr. Viskochil has served as Co-Chair of the NNFF CCAB and he now chairs the CTF CCAB. Dr. David Stevenson is a faculty member who has developed his academic career to investigating bone health of individuals with NF1, and he is restricted in clinical activity because of a K23 grant from NINDS to evaluate NF1-related bone issues. He has been involved in the care of patients with NF1 since his clinical genetics fellowship at the University of Utah beginning in 2002. He routinely integrates research studies as an extension of his clinical encounters.

ii. Your past and current association with NF clinical trials

The NF Clinic at the University of Utah has been involved in a number of clinical trials; however, it has not independently launched a phase II clinical trial. Dr. Carey was involved in a gene mapping trial in the 1980's, which led to the

mapping of the *NF1* gene to chromosome 17q11.2. Dr. Viskochil was involved in the identification and characterization of the *NF1* gene. As extensions of these studies, patients followed in the NF Clinic continue to engage in research studies conducted at the University of Utah. Dr. Stevenson is actively engaged in a number of clinical studies regarding bone health in NF1. Drs. Carey, Viskochil, and Stevenson are presently funded to carry out a number of studies related to tibial dysplasia, spine abnormalities, and bone health issues in NF1. The NF Clinic has enrolled 12 patients in a natural history study of plexiform neurofibroma, which was a DoD-funded longitudinal study awarded to Dr. Bruce Korf as principal investigator. Dr. Viskochil has been involved in a number of molecular studies of the *NF1* gene. There have been 2 clinical trials associated with learning issues in NF1 that have been conducted at the U of Utah. One was performed by Dr. Connie Dilts as a sib-pair study, and the other by Dr. Sally Ozonoff to evaluate executive function in adolescents with NF1.

Presently, the U of Utah NF Clinic is affiliated with the Clinical Genetics Research Program (CGRP), which serves as one of 9 sites participating in a NF Clinical Trials Consortium. This consortium is poised to begin a trial to treat NF1 patients with plexiform neurofibromas with rapamycin. Drs. Carey, Viskochil, and Stevenson are members of protocol committees that have developed potential trials reviewed for implementation through the NF Clinical Trials Consortium. Patients followed in the NF Clinic are enrolled in studies through the CGRP, which has 3 study coordinators, a research analyst, and a phlebotomist. It is under the supervision of Dr. Viskochil and is integrated with the NIH-funded General Clinic Research Center at the University of Utah. Multiple IRB-approved NF1-related protocols are conducted through this mechanism.

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

Each NF1 patient with optic pathway tumor is evaluated in our neuro-oncology clinic, and is enrolled in a CCG-protocol. Dr. Carol Bruggers has a protocol to treat these tumors with a carboplatin-only regimen, and she has compared this protocol results with those from a carboplatin-plus vincristine protocol. Our ophthalmology team routinely performs visual evoked potential studies on NF1 patients suspected of having optic nerve pathway tumors, and they collaborate with Dr. Bruggers in managing these tumors. Malignant peripheral nerve sheath tumors, NF1-related leukemia, and other sarcomas related to NF1 are treated per multi-center, IRB-approved protocols that can be considered clinical trials. The NF Clinic is tied to the oncology services of the childrens hospital (PCMC) and the Huntsman Cancer Institute on the U of Utah medical campus. Orthopedic protocols for tibial pseudarthrosis and dystrophic scoliosis are performed at PCMC and the Shriners Intermountain Hospital by surgeons who are affiliated with the NF Clinic.

- b. CLINIC DIRECTOR: Please provide information on:
- i. Present and past funding you have received for NF research. Include funding source, date received, amount and project description.

David Viskochil, MD, PhD

Young Investigator Award, National Neurofibromatosis Foundation (NNFF), 1988 - 1990, Sponsors: Ray White, PhD, Department of Human Genetics, and **John Carey**, MD Department of Pediatrics, University of Utah. Clonal analysis of neurofibromas.

Clinical Investigator Award, K08-N501492, Molecular Genetics of NF1, 1991 - 1994, National Institute of Neurological Disorders and Stroke, Sponsor - Ray White, PhD, Department of Human Genetics, University of Utah.

National Neurofibromatosis Foundation Grant, Structure/Function Analysis of NF1 Mutations, 1992-1993, **PI - David Viskochil**.

Rocky Mountain Center for the Biology of Development, Structure/Function Analysis of NF1 Mutations, 1992 - 1993, Institute of Child and Maternal Health, PI - Michael Simmons, MD, University of Utah, D Viskochil (recipient of funds).

Department of Defense/U.S. Army Medical Research Acquisition Activity, DAMD 17-933070, 1993-1996, Gene Structure and Somatic Mutation Analysis of NF1, **PI – D. Viskochil**.

Japan Society for the Promotion of Science and the Fogarty International Center, National Institutes of Health, Short-term Invitation Fellowship for Research in Japan. Identification of Deletions in Neurofibromatosis 1 Patients in Japan, **PI – David Viskochil**, Host Scientist - Professor Michihito Niimura, MD, Jikei University, Tokyo, Japan.

Department of Defense/U.S. Army Medical Research Acquisition Activity, DAMD NF960003, 1997 – 2000, Analysis of Phenotypic Variability in Neurofibromatosis Type 1, **Co-Investigator - David Viskochil**, PI - Jan Friedman, MD, PhD, University of British Columbia, Vancouver, Canada.

Department of Defense/U.S. Army Medical Research Acquisition Activity, DAMD NF980007, 1999 – present, Genetic Analysis of Peripheral Nerve Sheath Tumors in NF1. **PI – D Viskochil**.

Shriners Research Foundation, Tibial Pseudarthrosis in NF1. 2003-2006. **PI – D Viskochil**.

NIH-NINDS, R01 NS050509 Spinal Abnormalities in NF1. 2006-2011. **PI – D. Viskochil**.

Shriners Research Foundation, Multi-Center Study of Outcome of Tibial Dysplasia in NF1 Patients. 2004-2007, co-Investigator – D. Viskochil (**PI – John Carey**)

John Carey, MD

Project Number: 9165
Source of Funding: Shriners Research Foundation
Title of Project: Multi-Center Study of Outcome of Tibial Dysplasia
in NF1 Patients
Dates of Approval: 01/01/04 – 12/31/07
Role: PI

Project Number: K23 NS052500-01
Source of Funding: NIH-NINDS
Title of Project: Osseous Abnormalities in Neurofibromatosis Type 1
Dates of Approval: 08/15/2005 – 06/30/2009
Role: Co-Mentor for David Stevenson

Project Number: COSAB Grant F03-04-05
Source of Funding: Shriners Research Foundation
Title of Project: Spinal Abnormalities in Neurofibromatosis Type 1 (NF1)
Patients
Dates of Approval: 01/01/06-12/31/08
Role: Co-Investigator

David Stevenson, MD

01/01/2004 – 12/31/2007 Multicenter Research Grant – Clinical Outcomes Study
Title: Multi-Center Study of Tibial Dysplasia in NF1
Patients
Shriners Hospitals for Children
Role: Co-investigator

08/16/2005 – 03/31/2010 K23 Mentored Patient-Oriented Research Career
Development Award (5 K23 NS052500)
Title: Osseous Abnormalities in Neurofibromatosis
Type 1
NIH / NINDS
Role: Principal Investigator

11/15/2005 - 12/31/2007 CDMRP (#NF050133)
Title: Evaluation of the Reliability and Validity of the
Crawford Classification of Congenital Tibial Dysplasia
Department of Defense
Role: Co-investigator

01/01/2006 - 12/31/2009 Multi-Center Clinical Outcomes Study 9198
Title: Multi-Center Study of Spinal Abnormalities in
Neurofibromatosis Type 1 (NF1) Patients
Shriners Hospitals for Children
Role: Co-investigator

07/01/2006 – 03/31/2011	#1 R01 NS050509 Title: Spinal Abnormalities in Neurofibromatosis Type 1 NIH / NINDS Role: Co-investigator
10/15/2005-11/14/2006	CDMRP (#NF050159) Title: The University of Utah Clinical Genetics Research Program as an NF Consortium Site Department of Defense Role: Co-investigator
02/01/2003 - 06/30/2007	Innovative Research Grant Title: Skeletal Phenotyping and Mutation Screening in Neurofibromatosis Type 1 Primary Children's Medical Center Foundation Role: Principal Investigator
1996	University of Utah Student Summer Research Program Title: Mutational screening of 38 exons in 8 neurofibromatosis 1 cases for identification of a phenotype/genotype correlation of pseudarthrosis and neurofibromatosis 1. Role: student award under the supervision of Dr. David Viskochil
1997-1998	Alpha Omega Alpha Honor Society Research Fellowship Grant Title: Psychosocial impact of pseudarthrosis in patients with neurofibromatosis 1 Alpha Omega Alpha Honor Society Role: student award under supervision of Dr. Carey

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

Carey JC, Laub J, Hall BD. Variability and Penetrance of Neurofibromatosis: A genetic study of 60 families. Birth Defects: Original Article Series 1979, 15(5B): 271-281.

Dietz JN, Robbins T, Cannon LA, Schwartz CE, **Carey JC**, Johnson JP, Kivlin J and Skolnick MH. Linkage analysis of von Recklinghausen neurofibromatosis: chromosome 4 and 19. Gen Epidemiol 3:313-317, 1986.

Riccardi VM and **Carey JC**. von Recklinghausen neurofibromatosis genetic linkage studies: clinical considerations. J Med Gen 24:521, 1987.

Barker D, Wright E, Nguyen K, Cannon L, Fain P, Goldgar D, Bishop DT, **Carey JC**, et al. A genomic search for linkage of neurofibromatosis to RFLP. J Med Gen 24:506, 1987.

- Barker D, Wright E, Nguyen K, Cannon L, Fain P, Goldgar D, Bishop DT, **Carey JC**, et al. Localization of von Recklinghausen neurofibromatosis to the pericentromeric region of chromosome 17. *Science* 236:1100-1102, 1987.
- Fain PR, Barker DF, Goldgar DE, Wright E, Nguyen K, **Carey JC**, et al. Genetic analysis of NF-1: Identification of close flanking markers on chromosome 17. *Genomics* 1:340-345, 1987.
- White R, Nakanwa Y, O'Connell, **Carey JC**, et al. Linkage analysis of von Recklinghausen neurofibromatosis to DNA markers on chromosome 17. *Genomics* 1:364-367, 1987.
- O'Connell P, Leach RJ, **Carey JC**, et al. Fine structure DNA mapping studies of the chromosomal region harboring the genetic defect in NF-1. *Am J Hum Gen* 44:51-57, 1989.
- Ward K, O'Connell P, **Carey JC**, et al. Diagnosis of neurofibromatosis 1 using tightly linked, flanking DNA markers. *Am J Hum Gen* 46:943-949, 1990.
- Jadayel D, Fain P, Upadhyaya M, Ponder MA, Huson SM, **Carey J**, Fryer A, Matthew CGP, Barker DF, and Ponder BAJ. Paternal Origin of new mutations in von Recklinghausen neurofibromatosis. *Nature* 1990, 343:557-559.
- Viskochil D**, Buchberg AM, Xu G, Cawthon R, Culver M, Stevens J, Wolff R, Culver M, **Carey JC**, et al. Deletions and translocations interrupt a clonal gene at the neurofibromatosis type 1 locus. *Cell* 62:182-192, 1990.
- O'Connell P, **Viskochil D**, Buchberg AM, Fountain J, Cawthon RM, Culver M, Stevens J, Rich DC, Ledbetter DH, Wallace M, **Carey JC**, Jenkins NA, Copeland NG, Collins FS, and White R. The human homologue of murine Evi-2 lies between two von Recklinghausen neurofibromatosis translocations. *Genomics* 7:547-554, 1990.
- Viskochil D**, Cawthon R, O'Connell P, Xu G, Stevens J, Culver M, **Carey JC**, and White R. The gene encoding the oligodendrocyte myelin glycoprotein is embedded within the neurofibromatosis type 1 gene. *Mol Cell Biol* 11:906-912, 1991.
- Dilts CV, **Carey JC**, Leonard CO, Hoffman RO, Kircher JC, Ward K, Clark E and Creel D. Neuropsychological characteristics of children and adolescents with the Neurofibromatosis 1 gene. *J Beh Dev Peds* 17:229-239, 1996.
- Gutmann DH, Aylsworth A, **Carey JC**, Korf B, Marks J, Pyeritz RE, Rubenstein A, **Viskochil, D**. The Diagnostic Evaluation and Multidisciplinary Management of Neurofibromatosis 1 and Neurofibromatosis 2. *JAMA* 278: 51-57, 1997.

Martin G, **Viskochil D**, Bollag G, McCabe P, Crosier W, Haubruck H, Conroy L, Clark R, O'Connell P, Cawthon R, Innis M and McCormick F: The GAP-Related Domain of the Neurofibromatosis Type 1 Gene Product Interacts with ras p21. Cell, 1990, 63:843-849.

Xu G, O'Connell P, **Viskochil D**, Cawthon R, Robertson M, Culver M, Dunn D, Stevens J, et al.: The neurofibromatosis type 1 gene encodes a protein related to GAP. Cell, 1990, 62:599-608.

Cawthon R, Weiss R, Xu G, **Viskochil D**, Culver M, Stevens J, Robertson M, Dunn D, Gesteland R, O'Connell P and White R: A major segment of the neurofibromatosis type 1 gene: cDNA sequence, genomic structure, and point mutations. Cell, 1990, 62:193-201.

Jorde L, Watkins WS, **Viskochil D**, O'Connell P, and Ward K: Linkage Disequilibrium in the Neurofibromatosis 1 Region: Implications for Gene Mapping. Am J Hum Genet, 1993, 53:1038-1050.

Li Y, O'Connell P, Breidenbach Huntsman H, Cawthon R, Stevens J, Gangfeng X, Neil S, Robertson M, White R and **Viskochil D**. Genomic Organization of the Neurofibromatosis 1 Gene (*NF1*) , 1995, Genomics 25:9-18.

Purandare S, Huntsman-Breidenbach H, Li Y, Zhu X-L, Sawada S, Neil S, Brothman A, White R, Cawthon R and **Viskochil D**. Identification of Neurofibromatosis 1 (*NF1*) Homologous Loci by Direct Sequencing, Fluorescence In Situ Hybridization and PCR Amplification of Somatic Cell Hybrids, 1995, Genomics 30:476.

Sawada S, Florell S, Purandare S, Ota M, Stephens K and **Viskochil D**. Identification of *NF1* Mutations in Both Alleles of a Dermal Neurofibroma. Nature Genet, 1996, 14:110-112.

Purandare SM, Cawthon RM, Nelson LM, Sawada S, Watkins S, Ward K, Jorde L and **Viskochil DH**. Genotyping of PCR-based Polymorphisms and Linkage-Disequilibrium Analysis at the *NF1* Locus. Am. J Hum Genet, 1996, 59:159-166.

Viskochil D. In search of the Holy Grail: *NF1* mutation analysis and genotype-phenotype correlation. Genetics in Medicine 1;245-247, 1999.

Stevenson DA, Birch PH, Friedman JM, **Viskochil DH**, Balestrazzi P, Buske A, Korf BR, Niimura M, Pivnick E, Schorry E, Short P, Tenconi R, Tonsgard J, and **Carey JC**. Descriptive analysis of tibial pseudarthrosis in patients with neurofibromatosis 1. Am J Med Genet 1999;84:413-419.

DeClue JE, Heffelfinger S, Benvenuto G, Ling B, Li S, Rui W, Vass WC, **Viskochil D**, Ratner N. Epidermal growth factor receptor expression in neurofibromatosis type 1-related tumors and *NF1* animal models. J Clin Invest. 105:1233-41, 2000.

Costa RM, Yang T, Huynh DP, Pulst SM, **Viskochil DH**, Silva AJ, Brannan CI. Learning deficits, but normal development and tumor predisposition, in mice lacking exon 23a of *Nf1*. Nat Genet. 27:399-405, 2001.

Mukhopadhyay D, Anant S, Lee R, Kennedy S, **Viskochil D**, Davidson NO. C > U Editing of neurofibromatosis 1 mRNA occurs in tumors that express both the type II

transcript and apobec-1, the catalytic subunit of the apolipoprotein B mRNA-editing enzyme. Am J Hum Genet. 70:38-50, 2002.

Liew MA, Coffin CM, Fletcher JA, Hang MT, Tanito K, Niimura M, **Viskochil D**. Peripheral nerve sheath tumors from patients with neurofibromatosis type 1 do not have the chromosome translocation t(X:18). Pediatr Dev Pathol. 5:165-169. 2002.

Packer RJ, Gutmann DH, Rubenstein A, **Viskochil D**, Zimmerman RA, Vezina G, Small J, Korf B. Plexiform neurofibromas in NF1: toward biologic-based therapy. Neurology 58:1461-1470, 2002.

Viskochil D. Genetics of Neurofibromatosis 1 and the *NF1* Gene. J Child Neurology 17:562-570, 2002.

Zhou H, Coffin C, Perkins SL, Tripp SR, Liew M, **Viskochil DH**. Malignant Peripheral Nerve Sheath Tumor (MPNST): A comparison of grade, immunophenotype, and cell cycle/growth activation marker expression in sporadic and neurofibromatosis 1 (NF1)-related lesions. Am J Surg Pathol 27:1337-45, 2003.

Coffin C, Cassity J, **Viskochil D**, Randall RL, Albritton K. Non-neurogenic sarcomas in four children and young adults with neurofibromatosis type 1. Am J Med Genet. 127A:40-43, 2004.

Stevenson DA, Moyer-Mileur LJ, **Carey JC**, Maxwell S, Quick JL, Hoff CJ, **Viskochil DH**. Case-control study of the muscular compartments and osseous strength in neurofibromatosis type 1 using peripheral quantitative computed tomography. J Musculoskel Neuron Interact 2005;5:145-149.

Stevenson DA, **Viskochil DH**, Rope AF, **Carey JC**. Clinical and molecular aspects of an informative family with neurofibromatosis type 1 and Noonan phenotype. Clin Genet 2006;69:246-253.

Stevenson DA, Zhou H, Ashrafi S, Messiaen LM, **Carey JC**, D'Astous JL, Santora SD, **Viskochil DH**. Double inactivation of *NF1* in tibial pseudarthrosis. Am J Hum Genet 2006;79:143-148.

Stevenson DA, Moyer-Mileur LJ, Murray M, Slater H, Sheng X, **Carey JC**, Dube B, **Viskochil DH**. Bone mineral density in children and adolescents with neurofibromatosis type 1. J Pediatr 2007;150:83-88.

Upadhyaya M, Huson SM, Davies M, Thomas N, Chuzhanova N, Giovannini S, Evans DG, Howard E, Kerr B, Griffiths S, Consoli C, Side L, Adams D, Pierpoint M, Hachen R, Barnicoat A, Li H, Wallace P, **Stevenson D**, **Viskochil D**, Baralle D, Haan E, Turnpenny P, Riccardi V, Lizaro C, Messiaen L. An absence of cutaneous neurofibroma associated with a 3-bp in-frame deletion in exon 17 of the *NF1* gene (c.2970_2972 delAAT): Evidence of a clinically significant *NF1* genotype-phenotype correlation. Am J Hum Genet 2007;80:140-51.

Stevenson DA, **Viskochil DH**, Schorry EK, Crawford AH, D'Astous J, Murray KA, Friedman JM, Armstrong L, **Carey JC**. The use of anterolateral bowing of the lower leg in the diagnostic criteria for neurofibromatosis type 1. Genet Med 2007;9:409-412.

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

David Viskochil; Professor of Pediatrics; MD/PhD; Co-Director
John Carey; Professor of Pediatrics; MD; Co-Director
David Stevenson; Assistant Professor of Pediatrics; MD; Clinician
Janice Palumbos; Certified Genetic Counselor; MS; Genetic Counseling
Karin Dent; Certified Genetic Counselor; MS; Genetic Counseling
Natalee Pihl; Clinic Coordinator; BS; Nursing and Clinic Coordination
Carol Hsieh; Clinic Coordinator; BS; Clinic Scheduling

- d. Who within this core staff currently coordinates NF patient services?
Describe this individual's NF clinic related duties.

There is a joint effort. Ms. Hsieh takes initial phone or email correspondences from families and primary care providers. She contacts one of the co-directors or Janice Palumbos about the urgency of scheduling. Once scheduled the clinic encounter leads to a number of tests and referrals that are coordinated by Natalee Pihl and/or the genetic counselors.

- e. Describe any areas of NF 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.

Our clinical expertise is related to NF1. We focus on optic pathway tumors, peripheral nerve sheath tumors, and orthopedic complications of NF1. Clinic staff includes the physicians and genetic counselors.

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.

Contact by family or primary care provider leads to a call back within 2 days. The clinical geneticist on call is available for urgent problems. If the concern is not urgent, the clinic coordinator sends a scheduling packet through the mail to obtain records and demographic information. Once the packet is returned, a clinic date is scheduled with priority placed on urgency, new versus follow-up evaluation, distance from Salt Lake City, and the need for co-scheduling with other clinics. Urgent visits are scheduled directly with the clinical geneticist who is seeing the patient, and determined by room availability at the outpatient clinic.

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

PEDIATRIC CARE

SPECIALTY	DOCTOR	CLINIC ADDRESS	PHONE	EMAIL (if available)
Genetics	David Viskochil John Carey David Stevenson Susan Lewin Alan Rope	Primary Childrens Medical Center 60 N Medical Dr Salt Lake City Utah 84132	801 581-8943 Div. of Medical Genetics) 801 588-2000 (Hospital Operator)	Dave.viskochil@hsc.utah.edu
Neurology	James Bale Susan Benedict	PCMC	801 587-7575	
Orthopedics	Jacques D'Astous John Smith	Shriners PCMC	801 536-3500	
Developmental pediatrics/learning disabilities	Nancy Cantor	PCMC		
Ophthalmology	Robert Hoffman David Dries Scott Larson Don Creel	PCMC	801 581-2352	
Neurosurgery	Marion Walker Doug Brockmeyer	PCMC	801 662-5340	
Plastic surgery	Louis Morales	PCMC	801 743-0700	
Neurooncology	Carol Bruggers	PCMC	801 662-4700	
Medical Oncology/Radiation Oncology	Zeinab Afify	PCMC	801 662-4700	
Endocrinology	Mary Murray	PCMC	801 587-3922	
Audiology/ENT	Michael White	PCMC		
Radiology/ Neuroradiology	Kevin Moore Richard Boyer	PCMC	801 588-2000	
General Surgery/Surgical Oncology	Richard Black Lor Randall	PCMC Huntsman Cancer Inst.	801 662-2950 801 662-5600	
Dermatology	Sheryll Vanderhoof	PCMC	801 662-1603	
Cardiovascular Disease	Lloyd Tami	PCMC	801 662-5400	
Oral and Maxillofacial Surgery	Duane Yamashiro	PCMC	801 588-2000	
Behavioral Issues	Lisa Sampson-Fang	PCMC	801 581-3501	

ADULT CARE

SPECIALTY	DOCTOR	CLINIC ADDRESS	PHONE	EMAIL (if available)
Genetics	See Pediatrics			
Neurology	Jacinda Sampson	U of Utah Health Sciences Center (UUHSC)	801 585-6387	
Orthopedics	Darrel Brodke (spine) Lor Randall	University Orthopaedic Center	801 587-5451	
Developmental pediatrics/LD	See Pediatrics			
Ophthalmology	Bradley Katz Judith Warner	Moran Eye Center	801 581-2352	
Neurosurgery	Meic Schmidt Daniel Fults	UUHSC	801 585-6040	
Plastic surgery	Louis Morales Faizi Siddiqi	PCMC UUHSC	801 743-0700 801 581-5132	
Neurooncology		UUHSC	801 585-6040	
Medical Oncology/Radiation Oncology	Lei Chen Ying Hitchcock	UUHSC UUHSC	801 585-0255 801 581-2396	
Endocrinology		UUHSC	801 581-7761	
Audiology/ENT	Clough Shelton	UUHSC	801 581-8915	
Radiology/Neuroradiology	Ric Harnsberger	UUHSC	801 581-7553	
General Surgery/Surgical Oncology	Courtney Scaife	UUHSC	801 585-6911	
Dermatology	Douglas Grossman	UUHSC	801 581-2955	
Cardiovascular Disease		UUHSC		
Oral and Maxillofacial Surgery	Faizi Siddiqi	UUHSC	801 581-5132	
Behavioral Issues	Josette Dorius	UUHSC	801 585-1212	

4. NUMBER OF NF PATIENTS YOUR CLINIC SEES

- a. How many NF PATIENTS did you see in the past 12 months?
- b. How many of these were NEW patients to your clinic?

Insert numbers below

	NF1	NF2	SCHWANNOMATOSIS	OTHER
NUMBER OF PATIENTS SEEN IN PAST 12 MONTHS	148	2	1	~10
NUMBER OF <u>NEW</u> PATIENTS SEEN IN PAST 12 MONTHS	46	2	1	~2-5

c. Overall what proportion of patients seen in the past year were (give finite numbers if these are available, or estimate percentage):

Under 18 137 18+ 11 (give numbers - if data available)

OR estimate

Under 18 (%) 92% 18+ (%) 8%

We follow about 10 patients with pigmentary findings suggestive of mosaic NF1, who are evaluated on an annual basis.

5. TRANSITIONING PEDIATRIC TO ADULT NF CARE

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

We attempt to see our patients past 21 years of age, and refer them to appropriate subspecialists at the University of Utah, if insurance allows. Our NF Clinic has not been successful in this transition process, and we began a needs assessment as part of a genetic counselor student research project entitled; Health Care Transitioning: Perspectives of Adult Patients with Neurofibromatosis Type 1, Michelle Kempf, Karin Dent, Caren Frost, and David Viskochil. This research project completed requirements for Michelle Kempf to graduate from our Genetic Counseling Training Program. "Our study aimed at eliciting the perceived transition status of adults with NF1, identifying and understanding concerns surrounding transition from the pediatric to the adult health care setting, identifying perceived barriers and identifying elements to improve the transition process." Ms. Kempf will present these findings as an oral presentation at the 2007 National Society of Genetic Counselors Meeting in Kansas City, MO. The anonymous feedback from our patients will enable us to devise strategies to develop a transitioning plan.

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

Continuity of care from our pediatric-based clinic to adult care is not smooth, and many adults have “dropped out” of routine NF-related care. They come only if there is a significant medical complication, and many subspecialists fail to contact physicians in our NF Clinic for NF-related consultation. Thus, our institution is presently not providing adequate NF Clinic care for our adult population. Exceptions to this rule are the involvement of our sarcoma team under the direction of Dr. Lor Randall for the evaluation and management of peripheral nerve sheath tumors in NF1 and the management of vestibular schwannomas by Dr. Clough Shelton, an ENT subspecialist who has a keen interest in the surgical approaches to the management of these tumors.

We have an opportunity to change this pattern, and it is one of our goals in the next 3 years. Dr. Stefan Pulst is a neurologist who is familiar with NF, and he has recently taken the Chair position for Adult Neurology at the University of Utah. In addition, our clinical genetics service helped train a neurogenetics fellow, Jacinda Sampson, who is now a faculty member. She would like to develop a phacomatosis clinic embedded in neurology. Thus, we have an opportunity to expand our NF Clinic to the neurology clinic for oversight of adult care. We propose to use a clinic coordinator to help with this expansion and serve as a key staff member to integrate the scheduling and follow-up of adult patients as well as transition older adolescents to adult care.

6. INTERNAL CONFERENCES

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

We have a combination of conferences to address issues related to patient care in NF. Our primary review is a weekly case management conference devoted to the case-based discussion of management of clinic patients, including NF patients. All clinical geneticists participate and provide discussion regarding issues related to patient care. There is a weekly neuroradiology conference that allows us to add important and interesting NF subjects for review and discussion with neurologists, neuroradiologists, and neurosurgeons (every other week). We take advantage of these opportunities to review our most concerning cases. Oncology-based tumor board meets weekly and will discuss tumor-related issues for NF patients. This often includes discussion on protocol selection and surgical management of peripheral nerve sheath tumors. We meet with colleagues at the Shriners Intermountain Hospital on a weekly basis regarding orthopedic issues that may be clinically related, as part of our weekly research meeting that encompasses all Shriners-funded projects. Dr. Jacques D’Astous is the primary consultant, however other orthopedists at the Shriners hospital periodically join the conference.

7. CLINICAL TRIALS

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

X Yes No

Do you currently refer patients to clinical trials?

X 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.

Oncology trials are carried out through our respective oncology divisions. All patients treated through Primary Childrens Medical Center and the University of Utah Health Sciences Center (Including the Huntsman Cancer Institute) are placed on protocols approved as multi-center trials through Oncology Groups. Other clinical studies include a number of bone health studies and natural history studies. Our Clinical Genetics Research Program is affiliated with enzyme replacement therapy for lysosomal storage disorders conducted through the GCRC and now infusion centers at the respective medical centers.

8. PATIENT REGISTRY

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

X Yes No

If 'yes', please describe.

We have worked with Patricia Birch and Jan Friedman to pattern our NF1 Registry after the CTF-sponsored International NF Clinical Database. Our NF1 examination forms include information that can populate the modular database devised by the UBC database. We can download our information on a CD and send to UBC for their uploading into the database. Our registry is IRB-approved since 1999, and contains approximately 120 NF1 subjects. We ask for written subject consent to submit their confidential, de-identified but linked data to the Clinical Database. To date, all enrollees in our Registry have consented to have their personal data be sent to the International Clinic Database.

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

X Yes, providing patients agree and give written consent

9. PUBLICATIONS and RESEARCH (IF APPLICABLE)

- a. Please list any relevant NF publications from your clinic in the past 5 years. Include Journal, Citation and Title.

Please see Section 2bii

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

We primarily focus on bone-related issues in NF1 and MPNSTs. We have recently joined the DoD-funded NF Clinical Trials Consortium, and will implement protocols designed by the consortium. We are presently involved with the Shriners Hospitals to provide a tissue repository for bone resected from surgical intervention for NF1 complications. Tissue from this repository is available to other NF investigators. We are overseeing a multi-center natural history study on spine abnormalities in NF1 that includes the University of British Columbia, the University of Cincinnati, and the University of Manchester in the UK. This study began at the end of 2006 and will carry forward to 2011.

10. PATIENT SUPPORT

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

If 'yes' provide details.

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

What resources would help you to do this?

The Utah NF Support Group has been in existence for over 20 years, and at one point provided representatives to attend the NF Clinic. Changes in privacy expectations and confidentiality have led to decreased involvement of non-medical personnel in the clinic; however, patients are provided contact information and encouraged to contact the support group if they have not already done so. We have periodic meetings where medical concerns are presented in a didactic format followed by panel discussions. There has been a focus on adult care in the last 2 years.

11. OTHER INFORMATION

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

One of our goals is to link NF Clinics with Sarcoma Centers and Orthopedic Centers around the country. We support the concept of an infrastructure of clinic coordinators from NF Clinics that provide access to clinical trials, and we envision improved clinical care by systematic evaluation of outcomes that can only be evaluated through patient registries and enrollment in multi-center trials.