CURRICULUM VITAE

NAME Cynthia J. Tifft, MD, PhD

POSITION AND ADDRESS Deputy Clinical Director
National Human Genome Research Institute
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CITIZENSHIP USA

EDUCATION

1969-1973	BA, University of California at San Diego, San Diego, CA (Biology)
1974-1975	MS, Rutgers University, New Brunswick, NJ (Genetic Counseling)
1975-1981	PhD, University of Texas Graduate School of Biomedical Science, Houston,TX (Genetics)
1979-1983	MD, University of Texas Medical School at Houston, Houston, TX

POSTDOCTORAL TRAINING

1984-1985	Resident in Pediatrics, University of Texas Affiliated Hospitals, Houston, TX
1986-1987	Resident in Pediatrics, Johns Hopkins Hospital, Baltimore, MD
1987-1989	Medical Genetics Fellow, Inter-Institute Medical Genetics Program, National Institutes of Health, Bethesda, MD
1989-1991	Medical Staff Fellow, Genetics and Biochemistry Branch, NIDDK, NIH, Bethesda, MD

EMPLOYMENT

1991-1997	Division of Medical Genetics, Children's National Medical Center, Washington, DC
1997-2009	Chief, Division of Medical Genetics, Children's National Medical Center, Washington, DC
2009-	Deputy Clinical Director, National Human Genome Research Institute, Director, Pediatric Undiagnosed Diseases Program, National Institutes of Health, Bethesda, MD
2019-2021	Interim Program Director, Medical Genetic and Genomics Training Program, NHGRI

Academic Appointments

1991-1996	Assistant Professor of Pediatrics, George Washington University

	School of Medicine and Health Sciences
1996-2007	Associate Professor of Pediatrics, George Washington University School of Medicine and Health Sciences
2007-2010	Professor of Pediatrics, George Washington University School of Medicine and Health Sciences

Hospital Appointments

1987-1991	Clinical Fellow in Medical Genetics, Warren G. Magnuson Clinical Center, National Institutes of Health, Bethesda, MD
1987-1993	Contract Physician in Neonatology, Holy Cross Hospital, Silver Spring, MD
1991-2011	Attending Physician, Children's National Medical Center
1993-2011	Consulting Physician in Genetics, Holy Cross Hospital, Silver Spring, MD
1993-2009	Consulting Physician in Genetics, Shady Grove Adventist Hospital, Rockville, MD
1994-	Attending Physician, Warren G. Magnuson Clinical Center, National Institutes of Health, Bethesda, MD
2000-2009	Consulting Physician in Genetics, Washington Hospital Center, Washington, DC

LICENSURE AND CERTIFICATION

Licensure

1983	State of Texas (G5691) (inactive)
1986	State of Maryland (D33664)
1991	Commonwealth of Virginia (0101047619) (inactive)
1991	District of Columbia (19378) (inactive)

Certification

1987	American Board of Pediatrics (138148) (re-certified 1996, 2003, 2013)
1990	American Board of Medical Genetics (870733) (not time-limited, participating in MOC)

SOCIETIES and HONORS/AWARDS

1983	Alpha Omega Alpha
1986-	American Society of Human Genetics
1987-	Founding Fellow, American College of Medical Genetics
2001	"Above and Beyond" Award, National Tay-Sachs and Allied Diseases Association
2000-	Society of Inherited Metabolic Diseases
2003	"See the Light" Award, Matthew Forbes Romer Foundation
2004-	Founding Member, Global Organization for Lysosomal Disorders
2017	"Rare Impact" Award, National Organization for Rare Disorders
2019	Honoree, "Imagine and Believe" National Tay-Sachs and Allied Diseases
2019	NHGRI GREAT Award Scientific/Medical Category "GM1 Gene Therapy"
2019	NHGRI GREAT Peer Recognition for Support of colleagues and NHGRI IRP research projects
2019	NHGRI GREAT Award for Faculty Mentoring of NHGRI trainees
2020	NIH Director's Award for GM1 Gangliosidosis Gene Therapy

ADMINISTRATIVE DUTIES

1993-95	Director, Medical Genetics Fellowship Program, Children's National Medical Center
1994-2009	Co-Director, Children's Craniofacial Center, Children's National Medical Center
1995-	Fellowship Executive Committee, Metropolitan Washington Medical Genetics Residency Program, NHGRI
1996-1997	Member, Search Committee, Director, Center for Genetic Medicine, Children's Research Institute
1996-2000	Member, Research Advisory Committee, Children's Research Institute
2009-	Pediatric Care Committee, NIH Clinical Center
2009-	Medical Advisory, 09-HG-0009 Participant Reactions to Disease Risk Information
2011-	Medical Advisory, 11-HG-0238 Weight Management Interactions in a Virtual Clinical Environment
2013-	Common Fund Working Group, Undiagnosed Diseases Network, NIH
2014-2016	Co-Chair, Clinical Protocols Working Group, NIH Undiagnosed Diseases Network
2017-2019	Co-Chair, Steering Committee, NIH Undiagnosed Diseases Network
2017-	NHGRI Safety & Compliance Representative to the NIH Clinical Center
2019-2020	Member, Diversity Working Group, Office of the Director, NHGRI
2019-2020	Reviewer, Bench to Bedside Awards, Office of the Director, NIH
2019-	Advisory Board Member, International Collaboration on Rare Disorders and Orphan Drugs
2023-	President Elect, International Collaboration on Rare Disorders and Orphan Drugs
2020-	Member, Gene Therapy Task Force, Office of the Director, NIH
2023-	Vice Chair, Gordon Research Conference on Lysosomal Diseases (Chair for 2025 meeting)

EDUCATIONAL ACHIEVEMENTS

Medical School and Trainee Teaching

1993-2012	Gene 500 (Introduction to Genetics), Foundation for Advanced Education in the Sciences, NIH, 3 hours of lecture annually
1996-2009	Medical Genetics Elective Course #378, George Washington University School of Medicine, course director
2011-2023	Director, Medical Student Elective in Medical Genetics and Genomics, Office of Intramural Training and Education, NIH

CONSULTANT APPOINTMENTS

2003-2009	International Zavesca Advisory Group, Actelion Pharmaceuticals (paid)
2003-2009	Pompe Disease Advisory Group, Genzyme Pharmaceuticals, (unpaid)

2005-2009 Chair, Data Safety Monitoring Board, Amicus Therapeutics (unpaid)
2006-2009 Speaker's Bureau (Pompe Disease), Genzyme Pharmaceuticals, (paid)
2007-2009 Fabry Disease Advisory Group, Amicus Therapeutics (unpaid)
2007-2009 Hunter Outcome Survey North American Advisory Board, Shire Pharmaceuticals (unpaid)
2010-2021 Chair, Data Safety Monitoring Board, Amicus Therapeutics (011, 012, and 042 studies) (unpaid)
2010-2019 Data Safety Monitoring Board, National Institute of Child Health and Human Development, Rare Diseases Clinical Research Network (unpaid)

GRANTS AWARDED

Interpersonnel Agreement
National Institutes of Diabetes, Digestive & Kidney Diseases, NIH
1994-1997 (50% salary support)

Molecular Pathogenesis of GM2 Gangliosidoses
Interpersonnel Agreement
Genetics of Development & Diseases Branch
National Institutes of Diabetes, Digestive & Kidney Diseases, NIH
1999-2003 (50% salary support)

A Multicenter, Randomized, Dose Frequency Study of the Safety and Efficacy of Cerezyme Infusion Every Four Weeks in the Maintenance Therapy of Patients with Type 1 Gaucher Disease
Genzyme Therapeutics
$11,000 Site Principal Investigator

An Open-Label Maintenance Clinical Study of Iduronate-2-Sulfatase Replacement Therapy in Patients with Mucopolysaccharidosis (MPS) Type II
Shire Human Genetics Therapies
$22,000 Site Principal Investigator

Glycosphingolipid Quantitation and Inflammatory Protein Characterization in Cerebrospinal Fluid of Patients with Glycosphingolipid Storage Disorders: Surrogate Markers of Disease Progression
National Tay-Sachs and Allied Diseases Association
2003-2004
$45,000 Principal Investigator

Molecular Pathogenesis of Glycosphingolipid Storage Disorders
Genzyme Therapeutics
2003-2006
$150,000 Principal Investigator

Pharmacokinetics, Safety, and Tolerability of Zavesca® (Miglustat) in Patients with Infantile Onset GM2 Gangliosidosis: Single and Steady State Oral Doses
Actelion Pharmaceuticals
2005-2007

$236,000 Principal Investigator

Screening Cranial MRI Impacts Clinical Management in Patients with Neurofibromatosis type 1.
Neurofibromatosis, Inc. Mid Atlantic
2007-2008
$16,600 Principal Investigator

Intramural, NHGRI, Neurodegeneration in Glycosphingolipid & Glycoprotein Disorders; 2009-

Cooperative Research and Development Award with Sio Gene Therapies for a Phase I/II AAV9-GLB1 Intravenous Gene Therapy Trial for GM1 Gangliosidosis
2019-2023
$1,000,000
Principal Investigator

Cooperative Research and Development Award with Sanofi Genzyme for
Sanofi EFC15299 – AMETHIST Study for Late Onset GM2 Gangliosidosis
2020-2025
$900,000
Principal Investigator

BIBLIOGRAPHY

Papers in Refereed Journals

Tifft CJ and Ledermann HM. Immunization of hospitalized preschoolers: the need for hospital-based immunization programs. *Amer J Dis Child* 1988; 142:719-720.

Boose JA, Tifft CJ, Proia RL, Myerowitz R. Synthesis of a human lysosomal enzyme hexosaminidase B, using the baculovirus expression system. *Prot Expr and Purif* 1990; 1:111-120.

Tifft CJ, Proia RL, Camerini-Otero RD. The folding and cell surface expression of CD4 requires glycosylation. *J Biol Chem* 1992; 267:3268-3273.

Bulas DJ, Stern HJ, Rosenbaum KN, Fonda-Allen J, Glass R, and Tifft CJ. Variable prenatal appearances of osteogenesis imperfecta. *J Ultras Medicine* 1994; 13:419-27.

Cohen MS, Samango-Sprouse CA, Stern HJ, Custer DA, Vaught DR, Saal HM, Tifft CJ, Rosenbaum KN. The neuron-developmental profile of infants and toddlers with oculo-auriculo-vertebral spectrum and the correlation of prognosis with physical findings. *Amer J Med Genet*1995; 60:535-540.

Goldstein DS, Hahn SH, Holmes C, Tifft CJ, Harvey-White J, Milstein S, Kaufman S. Monoaminergic effects of folinic acid, L-DOPA, and 5-hydroxytryptophan in dihydropteridine reductase deficiency. *J Neurochem* 1995; 64:2810-2813.

Montemarano H, Bulas DI, Chandra R, Tifft CJ. Prenatal diagnosis of glomerulocystic kidney disease in short-rib polydactyly syndrome type II, Majewski type. *Ped Radiol* 1995; 26:669-671.

Pennybacker M, Liessem B, Moczall H, Tifft CJ, Sandhoff K, Proia RL. Identification of domains in human β-hexosaminidase that determine substrate specificity. *J Biol Chem* 1996; 19:17377-17382.

Sango K, McDonald MP, Crawley JN, Mack ML, Tifft CJ, Skop E, Starr CM, Hoffmann A, Sandhoff K, Suzuki K, Proia RL. Mice lacking both subunits of lysosomal β-hexosaminidase exhibit mucopolysaccharidosis and gangliosidoses. *Nat Genet* 1996; 14:348-352.

Tifft CJ, Proia RL. The β-hexosaminidase deficiency disorders: development of a clinical paradigm in the mouse. *Ann Med* 1997; 29:557-561.

Beaty TH, Maestri NE, Hetmanski JB, Wyszynski DF, Vanderkolk CA, Simpson JC, McIntosh I, Smith EA, Zeiger JS, Raymond GV, Panny SR, Tifft CJ, Lewanda AF, Cristion CA, Wulfsberg EA. Testing for interaction between maternal smoking and TGFA genotype among oral cleft cases born in Maryland 1992-1996. *Cleft Palate and Craniofac J*1997; 34:447-454.

Norflus J, Tifft CJ, McDonald MP, Goldstein G, Crawley JN, Hoffman A, Sandhoff K, Suzuki K, Proia RL. Bone marrow transplantation prolongs life span and ameliorates neurologic manifestations in Sandhoff disease mice. *J of Clin Invest* 1998; 101:1881-1888.

Becker JA, Vlach J, Raben N, Nagaraju K, Adams EM, Hermans MM, Reuser AJJ, Brooks SS, Tifft CJ, Hirschhorn R, Huie ML, Nicolino M, Plotz PH. The African origin of the common mutation in African-American patients with glycogen storage disease type II (GSD II). *Amer J Hum Genet* 1998; 62:991-994.

Shotelersuk V, Tifft CJ, Vacha S, Peters KF, Biesecker LG. Discordance of oral-facial-digital syndrome type 1 in monozygotic twin girls. *Amer J Med Genet* 1999; 86:269-273.

Glass RB and Tifft CJ. Radiologic changes in infancy in McKusick cartilage hair hypoplasia. *Amer J Med Genet* 1999 86:312-315.

Rosenberg MJ, Vaska D, Kiloran CE, Ning Y, Wargowski D, Hudgins L, Tifft CJ, Meck J, Blancato JK, Rosenbaum D, Pauli RM, Weber J, Biesecker LG. Detection of chromosomal aberrations by a whole-genome microsatellite screen. *Amer J Hum Genet* 2000; 66:419-427.

Oya Y, Proia RL, Norflus F, Tifft CJ, Langaman C, Suzuki K. Distribution of enzyme-bearing cells in GM2 gangliosidosis mice: regionally specific pattern of cellular infiltration following bone marrow transplantation. *Acta Neuropathol* 2000; 99:161-168.

Wada R, Tifft CJ, Proia RL. Microglial activation during acute neurodegeneration in Sandhoff disease and suppression by bone marrow transplantation. *Proc Natl Acad Sci* 2000; 97: 10954-10959.

Lacbawan F, Tifft CJ, Luban NL, Schmandt SM, Guerrea M, Weinstein S, Pennybacker M, Wong LI. Clinical heterogeneity in mitochondrial DNA deletion disorders: A diagnostic challenge of Pearson syndrome. *AmerJMed Genet* 2000; 95:266-268.

Tifft CJ, Proia RL. Stemming the tide: glycosphingolipid synthesis inhibitors as therapy for storage diseases. *Glycobiol* 2000; 10:1249-1258.

Jeyakumar M, Norflus F, Tifft CJ, Bortina-Borja M, Butters RD, Proia RL, Perry VH, Dwek RA, Platt FM. Enchanced survival in Sandhoff disease mice receiving a combination of substrate deprivation therapy and bone marrow transplantation. *Blood* 2001; 97:327-329.

Rosenberg MF, Killoran C, Dziadzio L, Chang S, Stone DL, Meck J, Aughton D, Bird LM, Bodurtha J, Cassidy SB, Graham JM Jr, Grix A, Guttmacher AE, Hudgins L, Kozma C, Michaelis RC, Pauli R, Peters KF, Rosenbaum KN, Tifft CJ, Wargowski D, Williams MS, Biesecker LG. Scanning for telomeric deletions and duplications and uniparental disomy using genetic markers in 120 children with malformations. *Hum Genet* 2001; 109:311-318.

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Ng D, Hadley Dw, Tifft CJ, Biesecker LG. Genetic heterogeneity of syndromic X-linked recessive microphthalmia-anophthalmia: Is Lenz microphthalmia a single gene disorder? *Amer J Med Genet* 2002; 11:308-314.

Slavotinek AM, Tifft CJ. Fraser syndrome and crypthophthalmos: review of the diagnostic criteria and evidence for phenotypic modules in complex malformation syndromes. *J Med Genet* 2002 39:623-633.

Gropman A, Chen TJ, Krasnewich D, Chernoff E, Tifft CJ, Wong LF. Variable clinical manifestations of homoplasmic G14459A mitochondrial DNA mutation. *Amer J Med Genet* 2004; 124A:377-282.

Ng D, Thakker N, Corcoran CM, Donnai D, Perveen R, Schneider A, Hadley DW, Tifft C, Zhang L, Wilkie AO, van der Smagt JJ, Gorlin RJ, Burgess SM, Bardwell VJ, Black GC, Biesecker LG. Oculofaciocardiodental and Lenz microphthalmia syndromes result from distinct classes of mutations in BCOR. *Nat Genet* 2004; 36:411-416.

Myerowitz R, Mizukami J, Richardson KL, Finn LS, Tifft CJ, Proia RL. Global gene expression in a type 2 Gaucher disease brain. *Mol Genet Metab* 2004; 83:288-296.

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Ries M, Moore DF, Robinson CJ, Tifft CJ, Rosenbaum KN, Brady R, Schiffman R, Krasnewich D. Quantitative dysmorphology assessment in Fabry disease. *Genet Med* 2006; 8:96-101.

Cabral WA, Chang W, Barnes AM, Weis M, Scott MA, Leikin S, Makareeva E, Kuznetsova NV, Rosenbaum KN, Tifft CJ, Bulas DI, Kozma C, Smith PA, Eyre DR, Marini JC. Prolyl 3-hydroxylase 1 deficiency causes a recessive metabolic bone disorder resembling lethal/severe osteogenesis imperfecta. *Nat Genet* 2007; 39:259-65.

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Solomon BD, Mohan P, Tifft CJ. Dysmorphic findings in two cases of abeta/hypobetalipoproteinemia. *Clin Dysmorphol* 2009; 18:90-91.

Lavenstein B, Tifft C, Lichter-Konecki. Dystonia secondary to tetrahydrobiopterin deficiency (BH4), recognition and therapy of the disorder. *Mov Disord* 2009 24(suppl).

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Gahl WA, Tifft CJ. The NIH Undiagnosed Diseases Program: lessons learned. *JAMA* 2011 305: 1904-5.

Bley AE, Giannikopoulos OA, Hayden D, Kubilus K, Tifft CJ, Eichler FS. Natural history of infantile G(M2) gangliosidosis. *Pediatr* 2011; 128:1233-41.

Gahl WA, Markello TC, Toro C, Fajardo KR, Sincan M, Gill F, Carlson-Donohoe H, Gropman A, Pierson TM, Golas G, Wolfe L, Groden C, Godfrey R, Nehrebecky M, Wahl C, Landis DM, Yang S, Madeo A, Mullikin JC, Boerkoel CF, Tifft CJ, Adams D; for the NISC Comparative Sequencing Program. The National Institutes of Health Undiagnosed Diseases Program: Insights into rare diseases. *Genet Med*, 2012; 14:51-9.

Adams DR, Sincan M, Fajardo KF, For the NISC Comparative Sequencing Program, Pierson TM, Toro C, Boerkoel CF, Tifft CJ, Gahl WA, Markello TC. Analysis of DNA sequence variants detected by high throughput sequencing. *Hum Mut* 2012 Apr;33(4):599-608.

Pierson TM, Simeonov DR, Sincan M, Golas G, Adams DA, Markello T, NISC Comparative Sequencing Program, Hansen NF, Cherukuri PF, Cruz P, Mullikin JC, Blackstone C, Tifft C, Boerkoel CF, Gahl WA. Exome sequencing and SNP analysis

detects heterozygosity in fatty acid hydroxylase-associated neurodegeneration. *Eur J Hum Genet* 2012 20(4):476-9.

Pierson TM, Adams DA, Markello T, Golas G, Yang S, Sincan M, Simenov DR, Fuentes Frjardo K, Hansen NF, Cherukuri PF, Cruz P, Teer JK, Mulikin JC; NISC Comparative Sequencing Program, Boerkoel CF, Gahl WA, Tifft CJ. Exome sequencing as a diagnostic tool in a case of undiagnosed juvenile-onset GM1 gangliosidosis, *Neurol,* 2012 79(2):123-6.

Duan X, Markello T, Adams D, Toro C, Tifft C, Gahl WA, Boerkoel CF. Cultural differences define diagnosis and genomic medicine practice: implications for undiagnosed diseases program in China. Front Med. 2013 Sep;7(3):389-94.

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Lim YH, Ovejero D, Sugarman JS, Deklotz CM, Maruri A, Eichenfield LF, Kelley PK, Jüppner H, Gottschalk M, Tifft CJ, Gafni RI, Boyce AM, Cowen EW, Bhattacharyya N, Guthrie LC, Gahl WA, Golas G, Loring EC, Overton JD, Mane SM, Lifton RP, Levy ML, Collins MT, Choate KA. Multilineage somatic activating mutations in HRAS and NRAS cause mosaic cutaneous and skeletal lesions, elevated FGF23 and hypophosphatemia. *Hum Mol Genet*. 2014 Jan 15;23(2):397-407.

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Pierson TM, Yuan H, Marsh ED, Fuentes-Fajardo K, Adams DR, Markello T, Golas G, Simenov DR, Holloman C, Tankovic A, Karamchandani MM, Schreiber JM, Mullikin JC for the NISC Comparative Sequencing Program, Tifft CJ, Toro C, Boerkoel CF, Tranelis SF, Gahl WA. *GRIN2A* mutation and early-onset epileptic encephalopathy: personalized therapy with memantine. *Ann Clin Transl Neurol.* 2014 1(3):190-198.

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Ng BG, Wolfe LA, Ichikawa M, Markello T, He M, Tifft CJ, Gahl WA, Freeze HH. Biallelic mutations in CAD, impair *de novo* pyrimidine biosynthesis and decrease glycosylation precursors. *Hum Mol Genet*. 2015 Jun 1;24(11):3050-7.

Karkashon S, Raghupathy R, Bhatia H, Dutta A, Hess S, Higgs J, Tifft CJ, Little JA. Intermediaries of branched chain amino acid metabolism induce fetal hemoglobin, and repress SOX6 and BCL11A, in definitive erythroid cells. *Blood Cells Mol Dis*. 2015 Aug;55(2):161-7.

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