Harvard Medical School/Harvard School of Dental Medicine
Format for the Curriculum Vitae

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Place of Birth: Cambridge, MA

Education

1988	B.F.A	Painting	Rhode Island School of Design
1997	M.D.	Medicine	University of Vienna

Postdoctoral Training

11/97-11/99	Resident	Pediatrics	Vienna General Hospital
11/99-08/00	Neurogenetics Staff	Neurogenetics	Kennedy Krieger Institute, Johns Hopkins Medical Institutions
09/00-06/01	Postdoctoral Fellow	Neurology	Johns Hopkins School of Medicine
07/01-06/02	Resident	Pediatrics	Geisinger Medical Center
07/02-06/05	Resident	Child Neurology	Massachusetts General Hospital

Faculty Academic Appointments

2005-2006	Instructor	Neurology	Harvard Medical School
2007-	Assistant Professor	Neurology	Harvard Medical School

Appointments at Hospitals/Affiliated Institutions

07/05-	Assistant	Neurology	Massachusetts General Hospital

2006-	Attending Physician	Child Neurology	Massachusetts General Hospital

Other Professional Positions

2005-2006	Visiting Scientist	Lerner Institute, Cleveland Clinic Foundation, Cleveland, OH

Major Administrative Leadership Positions

Local

2004-2005	Chief Resident	Pediatric Neurology, Massachusetts General Hospital

Regional

2005-	Director of the Leukodystrophy Service, the only clinic that specializes in leukodystrophy patients in New England	Massachusetts General Hospital

National and International

2008	Chair and Organizer of the International "Symposium on X-linked Adrenoleukodystrophy and Adrenomyeloneuropathy"	Massachusetts General Hospital
2008	Chair and Organizer of the "Symposium on Hereditary Sensory and Autonomic Neuropathy Type 1"	Massachusetts General Hospital
2009	Chair and Organizer of the "Future Directions in Diagnosis and Treatment of Leukodystrophies" Symposium	Child Neurology Society
2011	Chair and Organizer of the "Mechanisms and Interventions in Childhood Neurodegenerative Diseases" Symposium	National Tay Sachs and Allied Diseases Association
2011	Chair of the Peripheral Neuropathy Platform Session	American Academy of Neurology

Committee Service

Local

2009-	MGH Research Council 2009-	Massachusetts General Hospital Member

National and International

2005-	Scientific Advisory Board 2005-	United Leukodystrophy Foundation Board Member
2007-	Tay Sachs Gene Therapy Consortium 2007-	Tay Sachs Gene Therapy Consortium Clinician and Clinical Trialist for Human

		Studies in GM2 Gangliosidosis
2008-	Scientific Advisory Board	National Tay Sachs and Allied Diseases Association
	2008-	Board Member
2011-	Neurologic & Ophthalmic Gene & Cell Therapy Committee	American Society of Gene and Cell Therapy
	2011-	Committee Member

Professional Societies

2004-	Child Neurology Society	
	2004-	Member
2004-	American Academy of Neurology	
	2004-	Member
2007-	Society for Neuroscience	
	2007-	Member
2007-	International Society for Magnetic Resonance	
	2007-	Member
2008-	American Society for Neurochemistry	
	2008-	Member
2008-	Society for Inborn Errors of Metabolism	
	2008-	Member
2008-	American Neurological Association	
	2008-	Member
2011-	American Society of Gene and Cell Therapy	
	2011-	Member

Grant Review Activities

2006-	ULF Scientific Advisory Board	United Leukodystrophy Foundation
	2006-	Board Member
2006-	European Leukodystrophy Foundation	European Leukodystrophy Foundation
	2006-	External Grant Reviewer
2006	NTSAD Scientific Advisory Board	National Tay Sachs and Allied Diseases Association
	2006-	Board Member

Editorial Activities

NeuroRx, Neurology, New England Journal of Medicine, Annals of Neurology, American Journal of Neuroradiology

Other Editorial Roles

2010-	Editor	Frontiers in Neurodegeneration

Honors and Prizes

1999-2000	Stipendium	Milupa Metabolics	Award for metabolic brain

	Metabolicum		investigations in children
2000-2000	VIII International Congress of Inborn Errors of Metabolism Award	Society for Inborn Errors of Metabolism	Travel award
2002-2002	President's Award	American Neurological Association	
2004-2004	Top Scholar Fellow	Child Neurology Society	Award for excellence as a child neurology fellow
2005-2006	Harvard Center for Neurodegeneration and Repair Pilot Study Grant Award	Harvard Medical School	Award for the study of immune suppression in X-ALD
2005-2005	Marine Biological Laboratory Scholarship	Marine Biological Laboratory	Award to study neuroimmunology in the setting of neurodegenerative disease
2009	Wolfe Research Prize	American Neurological Association	Award for translational work in inherited neuropathies

Report of Funded and Unfunded Projects

Funding Information

Past

1997-1998 Transcranial Doppler Sonography in the Neonate
Austrian National Bank Foundation, ÖNB 7507
Consultant
This project aimed at defining cerebral blood flow in fullterm and preterm human newborns.

1998-1999 Transcranial Doppler Sonography in Pediatric Neurooncology
Scientific Foundation of the Major of Vienna
Consultant
We investigated cerebral blood flow alterations during chemotherapy in children with brain tumors.

2000-2001 Development of MRI Transmission through Internet-2 as a novel tool for remote evaluation of patients with X-linked Adrenoleukodystrophy (X-ALD)
NIH / N01-LM-9–3537
Consultant
The aim was to connect remote sites around the world and enable DICOM transmission of patients with rare neurodegenerative disorders.

2005-2006 Imaging the Animal Model of X-ALD
William Randolph Hearst Foundation, 520-45458-600377-746021-0000-6
PI
The goal of the stupid was to define the phenotype of ALD mice using advanced MR imaging.

2007-2009 A Natural History of Tay-Sachs Disease
Tay-Sachs Gene Therapy Consortium
PI
This project aimed to define and quantify the course of clinical decline in children and adults with GM2 gangliosidosis.

Current

2006- Imaging the Pathophysiology of AMN in Mice and Humans
NIH / K08 NS052550-01A1
PI ($175,635)
The goal of this project is to apply advanced MR techniques in the study of an animal model of adrenoleukodystrophy,

2009- Imaging the Pathophysiology of AMN in Mice and Humans
NIH / K08NS052550-04S1
PI ($105,372)
The goal of this supplement is to study monocyte recruitment and microglial activation in mice with AMN.

2009-2013 AAV-mediated gene therapy for GM2-gangliodoses
NIH / U01 NS064096
Co-Investigator ($16,181)
The goal of this proposal is to translate results obtained with AAV vectors in animal models of GM2-gangliosidoses into a human clinical trial.

2010-2015 The Role of Desoxysphingoid bases in HSAN1
NIH / NINDS / R01 NS072446-01
PI ($431,224)
The goal of this study is to determine whether desoxysphingoid bases are toxic to nerves and assess dietary means to lower these lipids.

2011-2013 Clinical Outcome Measures for a Gene Therapy Trial in Infantile and Juvenile GM2
National Tay Sachs and Allied Diseases Association Research Initiative Award
PI ($250,000)
The major goal of this project is to define clinical outcome measures that accurately quantify disease progression in patients with childhood GM2.

Current Unfunded Projects

2010- **PI** / Phase 2 Trial of L-serine in HSAN1
This is a double-blinded, randomized, placebo controlled clinical trial testing the efficacy of L-serine supplementation as treatment for hereditary sensory and autonomic neuropathy type 1. This proposal to the Orphan Products Grants Program at the FDA received a priority score of 133.

2010- **PI** / The Role of ELOVL1 in X-Linked Adrenoleukodystrophy
The major goal of this study is to evaluate the influence of ELOVL1, the gene responsible for fatty acid elongation, upon fatty acid metabolism and inflammatory demyelination in adrenoleukodystrophy, utilizing transgenic mouse models derived from lentiviral overexpression as well as conditional constructs.

Report of Local Teaching and Training

Teaching of Students in Courses

2010	Gene Therapy Graduate and Medical Students	Harvard Medical School Lecture

Formal Teaching of Residents, Clinical Fellows and Research Fellows (post-docs)

2005-	Peroxisomal Disorders Pediatric Residents	Massachusetts General Hospital Chief Rounds in Pediatrics
2006-	Leukodystrophy Review Partners Neurology Residency Program	Harvard Medical School Noon Conference
2006-	X-linked adrenoleukodystrophy - a genetic disorder with progressive inflammatory demyelination of the brain Neurology Residents	Massachusetts General Hospital Seminar
2007-	Brain Inflammation in Peroxisomal Disorders Pediatric Residents	Massachusetts General Hospital Seminar
2007	Endocrinological Aspects of Adrenoleukodystrophy Residents and Staff in Pediatrics	Massachusetts General Hospital Lecture
2008	Advanced MR Techniques in Leukodystrophies Residents and Staff in Radiology	Massachusetts General Hospital Seminar
2009-	X-linked Adrenoleukodystrophy: A Genetic Disorder with Spreading Brain Inflammation Graduate Students and Faculty in Genetics	Center for Human Genetic Research, MGH Seminar
2009-	Review of Leukodystrophies Pediatric Neurology Residents	Massachusetts General Hospital Seminar
2011-	Inherited Peripheral Neuropathies and HSAN1 Pediatric Neurology Residents	Massachusetts General Hospital Seminar
2011	Inherited Peripheral Neuropathies Pediatric Neurology Residents	Massachusetts General Hospital Seminar
2011	Myelination and Hypomyelination Pediatric Neurology Residents	Massachusetts General Hospital Seminar

Clinical Supervisory and Training Responsibilities

2006-	Supervise medical students on Child Neurology Service	1 month/year
2006-	Supervise residents attending Inpatient Child Neurology Service	1 month/year
2006-	Supervise residents attending Outpatient Leukodystrophy Service	1 day/week

Laboratory and Other Research Supervisory and Training Responsibilities

2006-	Director of research laboratory in the MGH	4-5 days/week

	Charlestown Navy Yard campus	
2006-	Supervise MR imaging group in the Martinos Center for Biomedical Imaging	1 day/week

Formally Supervised Trainees

2006-2007 Sameer Nagpal, MD / Medical Student at Emory
Dr. Nagpal was coauthor on a paper regarding microglial apoptosis in cerebral ALD, and I supervised his contribution to this paper during his time in my laboratory.

2006-2007 Megan Blackwell, PhD / Patent lawyer
I was thesis supervisor to Ms. Blackwell's thesis entitled "Target-specific contrast agents for MR microscropy," which was submitted to the Harvard-MIT Division of Health Sciences and Technology.

2007-2008 Dika Kuljis, BS / Graduate Student in Neuroscience at UCLA
Ms. Kuljis was coauthor on a paper regarding the discovery of atypical sphingolipids in a mouse model of hereditary sensory and autonomic neuropathy type 1, and I supervised her contribution to this paper during her time in my laboratory.

2007-2008 Ali Seyed Fatemi, MD / Assistant Professor at Johns Hopkins
I facilitated Dr. Fatemi research fellowship at Johns Hopkins and subsequent residency at MGH in child neurologies.

2007-2010 Kevin Garofalo, MS / Research Manager in Biotechnology
Mr. Garofalo's contribution to my laboratory led to a first author paper in the Journal of Clinical Investigation, as well as several other coauthorships.

2007-2011 Ourania Giannikopoulos, BS / Dental Student at Tufts Medical School
As research coordinator, Ms. Giannikopoulos contributed to the natural history of infantile GM2 gangliosidosis and created web-based database for this purpose, and subsequently earned coauthorship on a manuscript in Pediatrics.

2009-2011 Brian Schmidt, BS / Graduate Student in Neurobiology at University of Washington (Seattle)
Mr. Schmidt was coauthor on a paper regarding amino acid supplementation in both culture and animal work, and I supervised his contribution to this paper during his time in my laboratory.

2010-2011 Annette Bley, MD / Faculty at University of Hamburg, Germany, Department of Pediatrics
I supervised Dr. Bley during her work on the largest retrospective survey of infantile GM2 gangliosidosis to date, which led to her first authorship in a manuscript in Pediatrics.

2010- Patricia Musolino, MD, PhD / Research Fellow in Neurology
I mentored Dr. Musolino in preparation of several successful proposals for NIH (R25 and K12) and Foundation (Hearst) grants.

Formal Teaching of Peers

No presentations below were sponsored by outside entities.

2005, 2007, 2009, 2011	Leukodystrophies and Hypomyelinating Disorders, Case Presentations Child Neurology, Continuing Medical Education (Harvard Medical School)	2-3 talks/course Boston, MA
2009	MRI Patterns of Inherited White Matter Disorders Neuroradiology, Continuing Medical Education (Harvard Medical School)	1 talk/course Boston, MA

Local Invited Presentations

No presentations below were sponsored by outside entities.

2008, 2009 NEJM Clinicopathological Case / Grand Rounds
Massachusetts General Hospital

2010- A Clinical Approach to Leukodystrophies / Invited Lecture
Division of Cognitive and Behavioral Neurology, Brigham & Women's Hospital

Report of Regional, National and International Invited Teaching and Presentations

Invited Presentations and Courses

Regional

No presentations below were sponsored by outside entities.

2004- Imaging in Translational Research of Neurodegenerative Disorders
Eunice Kennedy Shriver Center, University of Massachusetts Medical School, Waltham, MA

2010- Natural History of GM2 Gangliosidosis
New England Regional Genetics Group, Concord, NH

National

No presentations below were sponsored by outside entities.

2005- Small Animal Imaging
United Leukodystrophy Foundation, Sycamore, IL

2007- Immunology of Adrenoleukodystrophy
United Leukodystrophy Foundation, Sycamore, IL

2007 Is Microglial Apoptosis an Early Pathogenic Change in X-ALD?
Society of Neuroscience, San Diego, CA

2008- Review of Leukodystrophies
American Academy of Neurology, Chicago, IL

2008- The Role of Inflammation in the Leukodystrophies
Hunter's Hope, Buffalo, NY

2009- Therapeutics for X-linked Adrenoleukodystrophy: Past, Present and Future
University of Minnesota, Minneapolis, MN

2010- ABCD1 Deficiency Impairs Mononuclear Phagocytic Cells: Implications for Neurodegeneration
Society for Neuroscience, San Diego, CA

2010- Hereditary Sensory Neuropathy and Mutant Serine Palmitoyltransferase
United Leukodystrophy Foundation, Sycamore, IL

2011 Oral L-serine supplementation provides treatment for hereditary sensory autonomic neuropathy type 1
American Academy of Neurology, Honolulu, HA

International

2007- Highfield Proton MRSI in Adult Patients with X-linked Adrenoleukodystrophy / Plenary Presentation

	International Society for Magnetic Resonance, Berlin, Germany
2007-	Mission Statement on the International MLD Registry / Invited Lecture Neuropediatrics, Hamburg, Germany
2008-	Overexpression Of Serine Palmitoyltransferase Rescues The Phenotype Of Hereditary Sensory And Autonomic Neuropathy / Invited Lecture Gordon Conference, Barga, Italy
2008-	Update on Leukodystrophies / Plenary Presentation Panamerican Society for Neurovirology, Guadalajara, Mexico
2009-	Abnormalities Affecting Postnatal White Matter Development: Leukodystrophies / Invited Lecture International Society for Magnetic Resonance in Medicine, Honolulu, HA
2011-	Lessons from Mice and Humans in HSAN1 / Invited Lecture Gordon Conference on Sphingolipids, La Ventura, CA

Report of Clinical Activities and Innovations

Current Licensure and Certification

2006	Massachusetts Registered Physician
2009	American Board for Psychiatry and Neurology

Practice Activities

2005-	Clinical activities	Massachusetts General Hospital	10 patients/week
2010-	Leukoboard, an interdisciplinary board of white matter experts to discuss referrals both nationally and internationally	Massachusetts General Hospital	Bi-weekly

Clinical Innovations

L-serine supplementation therapy	Based on my finding of atypical sphingolipids arising from mutant serine-palmitoyl transferase activity, I discovered that these lipids could be lowered by substrate supplementation. This was successful in mouse and human pilot studies, and is now leading to a first multicenter clinical trial of L-serine in the United States.

Report of Technological and Other Scientific Innovations

2008	I led a group at the Martinos Center for Biomedical Imaging in performing 7 Tesla MR Spectroscopic Imaging for the first time in adult patients with X-linked adrenoleukodystrophy, revealing novel metabolites in the brain at high resolution (Arch Neurol 2008).
2009	Using animal models that overexpress mutant transgenes involved in inherited peripheral neuropathies, I discovered the accumulation of atypical desoxysphingoid

lipids (J Neuroscience 2009). These have been proven to be neurotoxic lipids and represent a novel mode of neurodegeneration.

Report of Education of Patients and Service to the Community

Activities

No presentations below were sponsored by outside entities.

2006-	United Leukodystrophy Foundation / Expert Clinician Talk to lay group regarding symptomatic management of spasticity and seizures in leukodystrophy patients.
2008-	National Tay Sachs and Allied Diseases Association / Expert Clinician Talk to lay group regarding health issues related to neurodegeneration of childhood.

Educational Material for Patients and the Lay Community

Books, monographs, articles and presentations in other media

2008	Family Afflicted by Adrenoleukodystrophy	Expert Consultant	New England Cable News report, regarding a family with three siblings affected by ALD (May 2008)
2009	Gene therapy makes major stride in 'Lorenzo's Oil' disease	Expert Consultant	Los Angeles Times article (Nov. 6, 2009)
2009	Gene Therapy Halts Brain Disease in Two Boys	Expert Consultant	Science Now article (Nov. 6, 2009)

Report of Scholarship

Publications

Peer reviewed publications in print or other media

1. Ipsiroglu OS, **Eichler F**, Stockler-Ipsiroglu S, Trattnig S. Cerebral blood flow velocities in an infant with moyamoya disease. Pediatric Neurology. 1999;21(4):739-41.
2. Ipsiroglu OS, **Eichler F**, Stoeckler-Ipsiroglu S. Cerebral Doppler sonography of the neonate. A resume after 20 years and future aspects. . Clinics of Perinatology. 1999;26(4):905-46.
3. Ito R, Melhem ER, Mori S, **Eichler FS**, Raymond GV, Moser HW. Diffusion tensor brain MR imaging in X-linked cerebral adrenoleukodystrophy. Neurology. 2001;56(4):544-7.
4. **Eichler F**, Ipsiroglu O, Arif T, Popow C, Heinzl H, Urschitz M, Pollak A. Position dependent changes of cerebral blood flow velocities in premature infants. European Journal of Pediatrics. 2001;160(10):633-9.
5. **Eichler FS**, Barker PB, Cox C, Edwin D, Ulug AM, Moser HW, Raymond GV. Proton MR spectroscopic imaging predicts lesion progression on MRI in X-linked adrenoleukodystrophy. Neurology. 2002;58(6):901-7.
6. **Eichler FS**, Wang P, Wityk RJ, Beauchamp NJ Jr, Barker PB. Diffuse metabolic abnormalities in

reversible posterior leukoencephalopathy syndrome. AJNR American Journal of Neuroradiology. 2002;23(5):833-7.

7. **Eichler FS**, Itoh R, Barker PB, Mori S, Garrett ES, van Zijl PC, Moser HW, Raymond GV, Melhem ER. Proton MR spectroscopic and diffusion tensor brain MR imaging in X-linked adrenoleukodystrophy: initial experience. Radiology. 2002;225(1):245-52.
8. Tan WH, **Eichler FS**, Hoda S, Lee MS, Baris H, Hanley CA, Grant PE, Krishnamoorthy KS, Shih VE. Isolated sulfite oxidase deficiency: a case report with a novel mutation and review of the literature. Pediatrics. 2005;116(3):757-66.
9. Smith EE, **Eichler F**. Cerebral amyloid angiopathy and lobar intracerebral hemorrhage. Archives of Neurology. 2006;63(1):148-51.
10. **Eichler F**, Tan WH, Shih VE, Grant PE, Krishnamoorthy K. Proton Magnetic Resonance Spectroscopy And Diffusion-weighted Imaging In Isolated Sulfite Oxidase Deficiency. Journal of Child Neurology. 2006;21(9):801-805.
11. Liu CH, Kim YR, Ren JQ, **Eichler F**, Rosen BR, Liu PK. Imaging cerebral gene transcripts in live animals. Journal of Neuroscience. 2007;27(3):713-22.
12. **Eichler FS**, Mahmood A, Loes D, Bezman L, Lin D, Moser HW, Raymond GV. Magnetic Resonance Imaging Detection of Lesion Progression in Adult Patients With X-linked Adrenoleukodystrophy. Archives of Neurology. 2007;(64):659-664.
13. Moll NM, Rietsch AM, Ransohoff AJ, Cossoy MB, Huang D, **Eichler FS**, Trapp BD, Ransohoff RM. Cortical demyelination in PML and MS: Similarities and differences. Neurology. 2007;70(5):1-8.
14. **Eichler F**, Krishnamoorthy K, Grant PE. Magnetic resonance imaging evaluation of possible neonatal sinovenous thrombosis. Pediatric Neurology. 2007;37(5):317-23.
15. **Eichler FS**, Ren JQ, Cossoy M, Rietsch AM, Nagpal S, Moser A, Frosch MP, Ransohoff RM. Is microglial apoptosis an early pathogenic change in cerebral X-ALD? Annals of Neurology. 2008;63(6):729-742.
16. Ratai E, Kok T, Wiggins C, Wiggins G, Grant E, Gagoski B, O'Neill G, Adalsteinsson E, **Eichler FS**. 7 Tesla proton magnetic resonance spectroscopic imaging in adult X-linked adrenoleukodystrophy. Archives of Neurology. 2008;65(11):1488-1494.
17. **Eichler F**, Grodd W, Grant E, Sessa M, Biffi A, Bley A, Bley A, Kohlschuetter A, Kraegeloh-Mann I. Metachromatic Leukodystrophy: A Scoring System for Brain MR Observations. AJNR. 2009;30(10):1893-7.
18. Han G, Gupta SD, Gable K, Niranjanakumari S, Moitra P, **Eichler FS**, Brown RH, Harmon J, Dunn TM. Identification of small subunits of serine palmitoyltransferase: multiple SPT isozymes with distinct substrate specificities. PNAS. 2009;106(20):8186-91.
19. Mahmood A, Berry J, Wenger D, Escolar M, Sobeih M, Raymond G, **Eichler F**. Metachromatic Leukodystrophy: a Case of Triplets with the Late Infantile Variant and a Systematic Review of the Literature. Journal of Child Neurology. 2009.
20. **Eichler F,** Hornemann T, McCampbell A, Kuljis D, Penno A, Vardeh D, Tamrazian E, Garofalo K, Lee H, Kini L, Selig M, Frosch M, Gable K, von Eckardstein A, Woolf CJ, Guan G, Harmon JM, Dunn TM, Brown RH. Overexpression of the wildtype SPT1 subunit lowers desoxysphingolipid levels and rescues the phenotype of HSAN1. J Neurosci. 2009;29(46):14646-51.
21. Penno A, Reilly MM, Houlden H, Laura M, Rentsch K, Niederkofler V, Stoeckli ET, Nicholson G, **Eichler F**, Brown RH Jr, von Eckardstein A, Hornemann T. Hereditary sensory neuropathy type 1 is caused by the accumulation of two neurotoxic sphingolipids. J Biol Chem. 2010;285(15):11178-

87.

22. Thibert R, Hyland K, Chiles J, Steinberg S, **Eichler F**. Levodopa response reveals sepiapterin reductase deficiency in a female heterozygote with adrenoleukodystrophy. Journal of Inherited Metabolic Disease. 2011.
23. Bley A, Giannikopoulous O, Hayden D, Kubilus K, Tifft CJ, **Eichler FS**. Natural History of Infantile GM2 Gangliosidosis. Pediatrics in press. 2011.
24. Garofalo K, Penno A, Schmidt BP, Lee H, Frosch MP, von Eckardstein A, Brown RH, Hornemann T, **Eicher FS**. Oral L-serine supplementation reduces production of neurotoxic deoxy-sphingolipids in mice and humans with Hereditary Sensory Autonomic Neuropathy Type 1. Journal of Clinical Investigation in press. 2011.

Non-peer reviewed scientific or medical publications/materials in print or other media

1. **Eichler F**, Van Haren K. Immune response in leukodystrophies. Pediatric Neurology. 2007;37(4):235-44.
2. Schmahmann JD, Smith EE, **Eichler FS**, and Filley CM. Cerebral White Matter - Neuroanatomy, Clinical Neurology,and Neurobehavioral Correlates. Annals of the New York Academy of Sciences. 2008;1142:266-309.
3. Braverman N and **Eichler F**. Peroxisomal Disorders and Neurological Disease. In: Squire LR (ed.) Encyclopedia of Neuroscience, 2009, volume 7: 579-588. Oxford: Academic Press.
4. Costello DJ, Eichler AF, **Eichler FS**. Leukodystrophies: classification, diagnosis, and treatment. The Neurologist. 2009;15(6):319-28.
5. Costello DJ, **Eichler FS**, Grant PE, Auluck PK. A 57-year-old man with progressive neurologic decline. NEJM. 2009;360(2):171-81.
6. Krishnamoorthy KS, **Eichler FS**, Goyal NA, Small JE, Snuderl M. A 5-month-old boy with developmental delay and irritability. N Engl J Med. 2010;362(4):346-56.
7. Raymond G, **Eichler F**, Fatemi S, Naidu S. Leukodystrophies. 1st London: Mac Keith Press ;2011.
8. Kohlschuetter A, **Eichler F**. Childhood Leukodystrophies: A Clinical Perspective. Expert Review of Neurotherapeutics in press, 2011.

Professional educational materials or reports, in print or other media

My laboratory has created a website dedicated to the monogenetic lipid disorders of the nervous system that are being studied (http://www.eichlerlab.com/). The website provides information for graduate students and medical professionals regarding the nature of lipid abnormalities in select neurodegenerative diseases. It also provides an outline of the clinical disease course and useful links for professionals in neuroscience and medicine.

Clinical Guidelines and Reports

I wrote a chapter on X-linked adrenoleukodystrophy in Clinical Decisions Support: Pediatrics, per

invitation from Julia McMillan, MD, Director of the Pediatrics Training Program at Johns Hopkins. This chapter will guide residents in their pediatrics training program and provide a succinct clinical guideline to diagnoses and management of patients with X-ALD.

Abstracts, Poster Presentations and Exhibits Presented at Professional Meetings

Blackwell ML, Hubbard WC, Selig M, Rosen BR, Moser AB, **Eichler FS**. Contrast-Enhanced Ex Vivo MR Reveals Inflammatory Zone in X-Linked Adrenoleukodystrophy. Poster Award at the International Society for Magnetic Resonance in Medicine, 2008.

Garofalo K, Ren JQ, Kuljis D, Ransohoff R, **Eicher FS**. Chronic installation of lysophosphatidylcholine (C24:0) induces microglial activation and demyelination. Abstract presented at American Society for Neurochemistry, 2009.

Kuljis D, Garofalo K, Pillai B, Hamilton J, **Eichler FS**. Microglial Activation by Lysophosphatidylcholine is Alkyl Chain Length Dependent. Abstract presented at American Society for Neurochemistry, 2009.

Musolino PL, Rapalino O, Kunst M, **Eichler FS**. Decreased MR Perfusion Predcedes Lesion Progression in X-linked Adrenoleukodystrophy. Outstanding Junior Member Abstract Award from the Child Neurology Society, 2010.

Eichler FS, Garofalo K, Schmidt B, Elpek N, Mempel T, El Khoury J. ABCD1 Deficiency Impairs Mononuclear Phagocytic Cells: Implications for Neurodegeneration Abstract presented at Society for Neuroscience, 2010.

Narrative Report

I am a clinician investigator who specializes in the care of patients with neurogenetic disorders. After completing my training in child neurology at Massachusetts General Hospital, I joined the faculty at MGH and HMS, where I provide clinical care, supervise trainees, administer a clinic, and conduct basic and clinical research on lipid disorders affecting the nervous system to enhance our knowledge and the quality of patient care.

My clinical interests and research expertise revolve around translational neurogenetics. Since 2005, I have been actively involved in clinical work, providing treatment for patients with leukodystrophies and other inherited neuropathies. I receive referrals from colleagues locally, regionally, and nationally. Over the past two years, I have gained further insight into neurodegeneration in inherited neuropathies and identified novel neurotoxic lipids. With this knowledge, gained from my work in animal models and human patients, I discovered a substrate supplementation therapy to lower these neurotoxic lipids. This work has been awarded with an RO1 from the NIH and received the Wolfe Neuropathy Prize at the American Academy of Neurology. Recently, the FDA has awarded further funds to conduct a clinical trial in human patients. I am senior author on a manuscript in press at the Journal of Clinical Investigation describing this translational work in mice and human. The subsequent program that I have created has grown and now evaluates and treats 200 patients each year, and serves as a core training experience for 20

medical students, residents, and fellows each year.

I have been actively involved in the teaching and supervision of medical students and residents since I joined the MGH/HMS faculty. In addition, I lecture regularly in the Continuing Medical Education Courses offered by Harvard to child neurologists from around the country. I recently organized an international course on the mechanisms of childhood neurodegeneration for the National Tay Sachs and Allied Diseases Foundation. I have recently edited a book for the International Child Neurology Association entitled “Leukodystrophies” (2011, MacKeith Press).

In addition to providing clinical care, teaching, and laboratory oversight, I have been conducting clinical research on Tay Sachs that has defined the natural history of this devastating disorder. I am senior author on a manuscript in press in Pediatrics that describes the largest cohort of infantile GM2 gangliosidosis studied to date. This article presents the first survival estimates and quantifies the gain and loss of specific developmental milestones in these patients.

Since my appointment as Assistant Professor at HMS, I have endeavored to provide excellent clinical care, research oversight, and teaching, while also conducting clinical trials in my area of clinical expertise. Through my lecturing at a local, regional, and national level, my written works (24 peer reviewed original research, 10 topical reviews of the literature, and book chapters, and well-regarded texts), my clinical research and my involvement in professional societies, I have sought to improve e the care of patients with leukodystrophies and inherited neuropathies.