

CURRICULUM VITAE

Franca Cambi, MD, PhD

Address: Department of Neurology
University of Kentucky
KY Clinic L445
Lexington, KY 40536

Education

1985 - 1988 Ph.D. Dottorato Di Ricerca in Neuroscience

1979 M.D. degree, University of Florence

Post-graduate training

7/1/1989 – 6/30/1992 Resident, Department of Neurology, Jefferson Medical College, Thomas Jefferson University, Philadelphia, Pennsylvania

7/1/1988 – 6/30/1989 Intern, Internal Medicine, Medical College of Wisconsin, Milwaukee, Wisconsin

1986 - 1988 Research Associate, Department of Neurology and Neuroscience Laboratories, Tufts University School of Medicine, Boston, Massachusetts

1983 - 1986 Research Associate, Biochemistry Department, E.K. Shriver Center for Mental Retardation, Waltham, Massachusetts

1983 - 1986 Research Fellow in Neurology, Department of Neurology, Massachusetts General Hospital and Harvard, Medical School, Boston, Massachusetts

1980 - 1981 Intern Medicine, Ospedale S.M.N., Florence, Italy

1979 - 1980 Visiting Fellow, Department of Neurochemistry, Institute of Neurology, Queen Square, London, in Multiple Sclerosis Research, Laboratory, Directed by Dr. M.L. Cuzner

Academic Appointments

7/1/2008- Professor, Department of Neurology and SCoBIRC, Associate Professor, Department of Anatomy and Neurobiology, University of Kentucky, Lexington

1/1/2004-6/30/08 Associate Professor, Department of Neurology, Department of Anatomy and Neurobiology and SCoBIRC, University of Kentucky, Lexington, Kentucky

7/1/1998 –12/31/03 Associate Professor, Department of Neurology, Thomas Jefferson University, Philadelphia, Pennsylvania

1/1/1996 -12/31/03 Assistant Professor, Graduate Program in Developmental Biology and Teratology, Thomas Jefferson University, Philadelphia, PA

1/1/1993 – 7/1/1998 Director, Neurology Resident Clinic, Thomas Jefferson University, Philadelphia, PA

8/1/1992 – 7/1/1998 Assistant Professor, Department of Neurology, Thomas Jefferson University, Philadelphia, Pennsylvania

Clinical Activities

2004-present Neurology ward and consult attending, University of Kentucky Medical Center

1992-2003 Neurology ward and consult Attending, Jefferson Medical Center, Philadelphia, PA

Specialty Board Certifications

2003 Recertification in Neurology
1994 Board Certification in Neurology
1989 FLEX
1985 ECFMG Certificate, Foreign Medical Graduate Examination in the Medical Sciences

Honors and Awards

2007-08 Nominated Best Doctors in America
1983 Travel grant from National Multiple Sclerosis Society to attend V International Congress of Immunology.

Grant Activities:

[Current \(Faculty, University of Kentucky\)](#)

ACTIVE

Extramural

Agency/Foundation: NIH/NINDS
Grant type/number: RO1/ NS053905
Role: PI
Title: "PLP alternative splicing and oligodendrocyte differentiation"
Grant period: 1/3/2007-12/31/11
Direct costs: \$1,093,750.

Agency/Foundation: NIH/NINDS
Grant type/number: U10/ NS053368
Role: PI
Title: "Neuroprotection Exploratory Trials in PD (NET-PD)"
Grant period: 5/2006-11/30/10
Direct costs: \$367,500.

Pharmaceutical

Agency/Foundation: Eisai (Pharmaceutical Company)
Grant type/number: E2007-A001-302
Role: co-PI (PI: John Slevin)
Title: A multicenter, randomized, double-blind, placebo-controlled, parallel-group study of the efficacy, safety and tolerability of E2007 in Levodopa-treated Parkinson Disease patients with motor fluctuations.

Grant period: 3/07-8/07
Direct costs: Total direct cost is based on patient recruitment (\$14,000/patient)

PENDING:

Agency/Foundation: ELA (European Leukodystrophy Fondation)
Grant type/number: Research grant
Role: PI
Title: "PLP/DM20 alternative splicing: studies on regulatory sequences, splicing factors and mutations affecting splicing in PMD patients."
Grant period: 12/08-11/30/10
Direct costs: \$180,000

Agency/Foundation: KSEF (Kentucky Science and Engineering Foundation)
Grant type/number: Research grant
Role: PI
Title: "Studies on the mechanisms of neurodegeneration in a novel knockin mouse model"
Grant period: 07/01/08-06/30/10
Direct costs: \$100,000

COMPLETED:

Extramural:

Agency/Foundation: ELA (European Leukodystrophy Fondation)
Grant type/number: International Collaborative grant
Role: PI
Title: "Correcting PLP Driven Myelin Deficiencies: Cellular therapeutic approaches to restore a normal phenotype"
Grant period: 12/05-11/30/07
Direct costs: \$ 97,322 (total 354,000 euros for 4 projects)

Agency/Foundation: ELA (European Leukodystrophy Fondation)
Grant type/number: Research grant
Role: PI
Title: "PLP/DM20 alternative splicing: studies on regulatory sequences, splicing factors and mutations affecting splicing in PMD patients"
Grant period: 12/06-11/30/08 (reduced to one year 12-06-12/07 because of alternative funding)
Direct costs: \$139,824

Agency/Foundation: NIH/NICHD
Grant type/number: RO3/HD043104
Role: PI
Title: "PLP splicing: in vivo analysis in a mouse model"
Grant period: 3/03-2/05 (no cost-extension to 2/07)
Direct costs: \$100,000.

Agency/Foundation: NIH
Grant type/number: COBRE
Role: coPI (sub-project)
Title: "Center for Pediatric Research"

Grant period: 2004-2009 (reduced to 2 years 2004-2006)
Direct costs: \$89,092.

Agency/Foundation: NIH/NINDS
Grant type/number: RO1/NS36711
Role: collaborator (PI: Richard Myers, Boston University)
Title: "Genetic linkage study in Parkinson's disease"
Grant period: 07/01/1998 – 06/30/2007
Direct costs: \$1,250/recruited sib pair

Intramural:

Agency/Foundation: University of Kentucky Research Fund
Grant type/number: Pilot for microarrays
Role: PI
Title: Genes differentially expressed in differentiated oligodendrocytes
Grant period: 3/05-9/05
Direct costs: \$4,800

Completed (Faculty, Thomas Jefferson)

Extramural:

Agency/Foundation: ELA (European Leukodystrophy Fondation)
Grant type/number: Research grant
Role: co-PI (PI: Odile Boespflug-Tanguy)
Title: "PLP gene expression: diagnosis and treatments of inherited disorders of CNS myelin formation"
Grant period: 9/1/03- 10/1/05
Direct costs: \$20,000

Agency/Foundation: NMSS (National Multiple Sclerosis Society)
Grant type/number: Pilot/ PP0860T
Role: PI
Title: PLP splicing: in vivo analysis in a mouse model
Grant period: 04/01/2002-3/31/03 (no cost extension 12/31/2004)
Direct costs: \$33,000.

Agency/Foundation: State of PA
Grant type/number: RFA 01-07-26 (PPG, Director Dr. Feldman)
Role: PI (sub-project)
Title: PLP splicing: in vivo analysis in a mouse model
Grant period: 1/1/03-1/1/06
Direct costs: \$285,273.

Agency/Foundation: PMD Foundation
Grant type/number: Grant-in-Aid
Role: PI
Title: PLP splicing: in vivo analysis in a mouse model
Grant period: August 1, 2002-June 30, 2003
Direct costs: \$8,000.

Agency/Foundation: NIH/NINDS

Grant type/number: U10/ NS053368
Role: co-PI (PI: Jay Schneider)
Title: "Neuroprotection Exploratory Trials in PD (NET-PD)"
Grant period: 05/01/03- 04/30/08
Direct costs: \$375,000.

Agency/Foundation: NIH/NINDS
Grant type/number: NS 38681
Role: co-PI (PI: Jay Schneider)
Title: GM1 Ganglioside (Sygen®) Therapy in Parkinson's Disease
Grant period: 7/1/99 – 6/30/04
Direct costs: \$1,795,000.

Agency/Foundation: MDA
Grant type/number: Research grant (PI: Enrica Arnaudo)
Role: co-PI
Title: "Human Tissue Culture System to Study Motor Neuron Disease"
Grant period: 7/1/99 – 6/30/04
Direct costs: \$100,000.

Agency/Foundation: NIH/NINDS
Grant type/number: KO8
Role: PI
Title: "Transcriptional Regulation of Myelin Proteolipid Protein"
Grant period: 9/30/94 – 8/31/99
Direct costs: \$378,666.

Intramural

Grant type/number: Internal grant Thomas Jefferson University
Role: PI
Title: "A Regulatory Element in Intron 3 of the Proteolipid Protein Gene: Characterization of its Role in Alternative Splicing and in Human Disease"
Grant period: July 1, 2001 – June 30, 2002
Direct costs: \$10,000.

Grant type/number: Internal grant Thomas Jefferson University
Role: PI
Title: "Cell Cycle Control and Oligodendrocyte Differentiation"
Grant period: February 1, 1999 – October 31, 2000
Direct costs: \$10,000.

Professional Organization Memberships

1992 - American Academy of Neurology
1999- Society for Neuroscience
2001- American Neurological Association

Editorial Activities

Ad Hoc reviewer:

Annals of Neurology
Neurology
J Neuroscience Research

Glia
European Journal of Human Genetics
American Journal of Medical Genetics
Experimental Neurology
Human Mutation
Current Medicinal Chemistry

Grant reviewer

1998 - Ad Hoc Reviewer for the National Science Foundation
2004-06 The Myelin Project
2004- European Leukodystrophy Association (ELA Fondation)
2004- Ministere de la Recherche, France
2006- French National Research Agency (ANR)
2006 The Austrian Science Fund
2007- National Multiple Sclerosis Society (NMSS)
2007 Reviewer and Study Section member for Department of Defense (DoD)
Congressionally Directed Medical Research Programs (CDMRP) Intramural
Traumatic Brain Injury (TBI)

Licensure

2004 Kentucky State Medical Board
1992 Commonwealth of Pennsylvania Licensed Physician and Surgeon
1989 State of Wisconsin

University Committee's work

University Services (Faculty, University of Kentucky):

2007 Member of the Review Committee for the 6 year review of the Department of
Microbiology
2005 Served in the Review Committee for the 6 year review of the Family Practice
department.

University Services (Faculty, Thomas Jefferson)

1997 - 2002 Chairman of Graduate Education Committee, Dept of Neurology
1997 - 2002 Member of the Executive Committee, Dept of Neurology
1996 - 2002 Member of Committee for Selection of Neurology Resident Applicants

External Committee work (Faculty, University of Kentucky)

2007 External Member Committee for the "Habilitation a diriger des recherches", INSERM,
University of Clermont-Ferrand, France
2004-2006 Reviewer: Membership Committee of the American Neurological Association
2004 Pelizaeus-Merzbacher disease (PMD) Clinical Task Force, this task force has to
establish uniform protocols for clinical, imaging studies, tissues banking and
databases.
2000- Member of the Scientific Advisory Board of the PMD Foundation

Teaching and Educational Activities: (Faculty University of Kentucky)

- 2008-09 MI816: Cellular Structure and Function/Genetics: Glutamine Repeat Disorders and Clin Corr: Spinocerebellar Ataxia
- 2007 Course ANA 780: 10-17-07: Post-Traumatic Demyelination and Remyelination, University of Kentucky
- 2005 Education agreement between the Doctorate School in Oncology and Genetics, University of Siena and the Department of Neurology, University of Kentucky
- 2005 Course ANA 605.10-12-05: Post-Traumatic Demyelination and Remyelination, University of Kentucky
- 2004-present Teaching Medical Students two lectures/year during Neurology rotation

Teaching and Educational Activities: (Faculty Thomas Jefferson)

- 1997 - 2002 Coordinator, Neurology Resident Journal Club
- 1997 Lecturer on Neurological Exam, Oral Surgery Residency Program
- 1997 - 2002 Teacher, course on "Developmental Biology", Graduate students
- 1997 - 2002 Teacher, course on "The Science of Clinical Neurology", Sophomore students
- 1997 - 2002 Teacher, course "IDEPT 150", Freshman medical students
- 1995 - 1996 Teacher, course "ID150", First year medical students
- 1995 - 2002 Teacher, course on "The Science of Clinical Neurology", Sophomore Course
- 1994- 1995 Teacher, course on "Neurogenetics", Department of Neurology, Pennsylvania Hospital, Philadelphia, PA
- 1994 Teacher, course "ID450"
- 1993 Teacher, course on "The Science of Clinical Neurology", Sophomore students

Clinical Instruction

- 1993 - 1998 Supervisor, Neurology Resident Clinic
- 1993 - 2000 Morning Reports once a week

Graduate Students (Faculty, University of Kentucky)

- 2007- Advisor, thesis for medical school degree, student Giulio Disanto to be defended at the University of Siena (2009).
- 2006- Thesis Committee member in Physiology Program, student I-Fang Ling, Advisor Dr. Steve Estus.

Graduate Students (Faculty, Thomas Jefferson)

- 2002-2005 Member of the Thesis Committee for PhD student, Mariam Zaka, "IGF-1 protects oligodendrocytes from psychosine-induced cell death". Department of Biochemistry and Molecular Pharmacology, Thomas Jefferson U.
- 2001-2003 Member of the Thesis Committee for MD-PhD student, Christopher O'Brien, Department of Biochemistry and Molecular Pharmacology, Thomas Jefferson U.
- 1997 -1999 External Member: Ph.D. Thesis, Wei Shen: The Expression of The Brain Creatine Kinase Gene and The Ubiquitous Mitochondrial Creatine Kinase Gene During Postnatal Rat Brain Development, Biology Department, University of Delaware.
- 1995 - 1996 Member of the Thesis Committee for Gibbon Program Scholar, Ms. Teresa Victoria: "Cloning Of the Canine Galactocerebrosidase cDNA. Determination Of Possible Mutations That Would Cause Globoid Cell Leukodystrophy In This Animal Model."

1993 - 1994 Member of the Thesis Committee for Gibbon Program Scholar, Ms. Solar-Pastor: "Metachromatic Leukodystrophy Among the Navaho Indians. A Study of the Arylsulfatase A Gene Mutations Present in This Population."

Mentoring:

Post-doctoral Fellows and PhD students mentored (Faculty, Thomas Jefferson)

1994-1997	Xue-Ming Tang, MD	<u>Current position:</u> Senior Technical Consultant, CSS Informatics, A division of PPD
1998-1999	Tomaz Mars, PhD student	<u>Current position:</u> Assistant Professor, University of Ljubljana, Slovenia
1999-2003	Zhong Huang, PhD	<u>Current position:</u> Lab manager, Dept of Surgery, Thomas Jefferson University, PA

Undergraduate Students mentored (Faculty, University of Kentucky)

Summer 2005	Gomathie Shelvahoyan	<u>Current position:</u> Medical student OSU
Summer 2006	David Soleimani-Meigooni	<u>Current position:</u> Senior, U of Louisville
Summer 2007	Nick Steinmetz	<u>Current position:</u> PhD student, Neuroscience Program, Stanford University
Summer 2007	Giulio Disanto	<u>Current position:</u> Medical Student University of Siena, Italy

Publications

Doctoral Theses:

Cambi F: Neutral proteases in demyelinating diseases. Thesis for fulfillment of M.D. degree, 1979.

Cambi, F: Regulation of Tissue Specific Expression of Tyrosine Hydroxylase, Thesis for fulfillment of Ph.D. degree, 1989.

PEER-REVIEWED ORIGINAL ARTICLES

1. Williams RM, Lees MB, **Cambi F**, Macklin WG: Chronic EAE induced in rabbits with bovine proteolipid. *Journal of Neuropathology and Experimental Neurol* 1982;41(5):508-521.
2. **Cambi F**, Massacesi L, Amaducci L: La sclerosi multipla: malattia dagli anticorpi senza senso. In: *Aggiornamento del medico pratico*, 1982.
3. **Cambi F**, Lees MB, Williams RM, Macklin WB: Chronic EAE produced by bovine proteolipid apoprotein: immunologic studies. *Annals of Neurology* 1983;303-308.
4. **Cambi F**, Lees MB: Chronic EAE in guinea pigs: immunologic studies on the two major myelin proteins. *Cellular Immunology* 1984;86:567-574
5. Amaducci L, Massacesi L, Guarnieri B, **Cambi F**: Proteases, protease inhibitors and diseases of the nervous system. In *Neuroimmunology* (PO Behan and F Spreafico, eds), Raven Press. 1984;12:127-142.

6. Lees MB, **Cambi F**: Non-contamination of proteolipid protein preparation by MBP. *Annals of Neurology* 1985;18:268.
7. **Cambi F**, Fung B, Chikaraishi DM: 5' flanking DNA sequences direct cell-specific expression of rat tyrosine hydroxylase. *Journal of Neurochemistry* 1989;53:1656-1659.
8. **Cambi F**, Kamholz J: Transcriptional regulation of rat PLP gene in primary oligodendrocytes. *Neurochemical Research* 1994;19:1055-1060.
9. **Cambi F**, Tartaglino L, Lublin F, McCarren D. X-linked pure familial spastic paraparesis: characterization of a large kindred with magnetic resonance imaging. *Archives of Neurology* 1995;52:664-669.
10. Strocchi P, Tang X-M, **Cambi F**. Molecular diagnosis of transthyretin Met 30 mutation in an Italian family with familiar amyloidotic polyneuropathy. *FEBS Letters* 1995;359:203-205.
11. **Cambi F**, Tang X-M, Cordray P, Fain P, Keppen L, Barker D. Refined genetic mapping and proteolipid protein (PLP) mutation analysis in X-linked pure hereditary spastic paraplegia (HSP). *Neurology* 1996; 46:112-117.
12. Fink JK, Heiman-Patterson T, **Cambi F** for the hereditary spastic paraplegia working group (1996): Hereditary spastic paraplegia: Advances in genetic research. *Neurology* 1996; 46: 1507-1519.
13. Berghella V, Danae Steele C, Spector T, **Cambi F**, Johnson A. Successful pregnancy in a neurologically-impaired Wilson's disease patient: a case report. *American Journal of Obstetrics and Gynecology* 1997;176:712-714.
14. Garbern J, **Cambi F**, Tang X-M, et al. PLP protein is necessary in both central and peripheral myelin. *Neuron* 1997; 19: 205-218.
15. Tang X-M, Strocchi, P, **Cambi, F**. Changes in the activity of cdk2 and cdk5 accompany differentiation of rat primary oligodendrocytes. *Journal of Cellular Biochemistry* 1998; 68: 128-137.
16. Garbern J, **Cambi F**, Shy M, Lewis RA, et al. Peripheral neuropathy caused by proteolipid protein gene mutations. *Annals of the NY Academy of Sciences* 1999; 883:351-65.
17. Tang X-M, Beesley J, Grinspan J, Seth P, Kamholz J, **Cambi F**. Cell cycle arrest induced by p27 is not sufficient to induce oligodendrocyte differentiation. *Journal of Cellular Biochemistry* 2000;76:270-279.
18. Awatramani R. Beesley J. Yang H, **Cambi F**, Tang X-M, Grinspan J, Garbern J, and Kamholz J. Gtx, an oligodendrocyte-specific homeodomain protein, is a repressor. *Journal of Neuroscience Research* 2000; 61:376-87.
19. Hentati A, Han-Xiang D, Hong, Z, Wenjie C, **Cambi F**, et al. Novel mutations in spastic and absence of correlation with age at onset of symptoms. *Neurology* 2000; 55:1388-1390.
20. Mars T, Yu JK, Tang XM, Miranda A, Grubic Z, **Cambi F** and King MP. Differentiation of glial cells and motor neurons during development of the neuromuscular junction in co-cultures of rat spinal cord explant and human muscle. *Journal of Comparative Neurology*, 2001;438(2):239-51.

21. Garbern J, Shy M, Krajewski K, Kamholz J, Hobson G, **Cambi F**. Evidence for neuro-axonal injury in patients with proteolipid gene mutations. *Neurology*, 2001; 57(10):1938-39.
22. Huang Z, Tang X-M, **Cambi F**. Differentiation of oligodendrocytes is associated with loss of expression of the retinoblastoma protein. *Molecular and Cellular Neuroscience* 2002;19:250-262.
23. Starling A, Rocco P, **Cambi F**, Hobson G, Passos Bueno MR, Zatz M. Further Evidence for a Fourth Gene Causing X-Linked Pure Spastic Paraplegia. *American Journal of Medical Genetics* 2002; 111:152-156.
24. Hobson G, Huang Z, Sperle K, Stabley D, Marks HG, **Cambi F**. A PLP splicing abnormality is associated with an unusual presentation of PMD. *Annals of Neurology* 2002; 52:477-488

Faculty, University of Kentucky

25. Wang E, Huang Z, Hobson G, Dimova N, Sperle K, A McCullough and **Cambi F**. PLP1 alternative splicing in differentiating oligodendrocytes: characterization of an exonic splicing enhancer. *Journal of Cellular Biochemistry* 2006; 97:999-1016.
26. Hobson G, Huang Z, Sperle K, Sistermans E, Rogan P, Garbern J, Kolodny E, Naidu S and **Cambi F**. Analysis of the contribution of splice site strength in the regulation of proteolipid protein and DM20 alternative splicing in differentiating oligodendrocytes *in vitro*. *Human Mutation* 2006; 27:69-77.
27. Sabet A, Li J, Ghandour K, Pu Q, Wu X, Kamholz J, Shy M and **Cambi F**. Skin biopsies demonstrate MPZ splicing abnormalities in CMT1B. *Neurology* 2006; 67:1141-1146.
28. Wilk JB and the GenePD study group. The effect of GSTP1 polymorphisms on onset age of Parkinson disease is modified by occupational exposure to herbicides in males: The *GenePD* Study. *Neurology* 2006; 67:2206-10.
29. Squillaro T, **Cambi F**, Ciacci G, Rossi S, Olivelli M, Malandrini A, Mari F, Renieri A, Ariani F. Frequency of the LRKK2 G2019S mutation in Italian patients affected by Parkinson Disease. *Journal of Human Genetics* 2007; 52:201-4.
30. Wang E, Dimova N, and **Cambi F**. PLP/DM20 ratio is regulated by hnRNPH and F and a novel G-rich enhancer in oligodendrocytes. *Nucleic Acids Research* 2007; 35: 4164-78.
31. Tobin JE, and the GenePD study group. Haplotypes and gene expression implicate the *MAPT* region for Parkinson disease: The *GenePD* Study. *Neurology*, 2008 (**ePub**).
32. Wang E, Dimova N, Sperle K, Huang Z, Lock L, Hobson G and **Cambi F**. Targeted deletion of a PLP1 intronic splicing enhancer in a novel knockin mouse impairs the developmental increase in PLP1 to DM20 ratio and affects myelin stability and function. **In review, 2008**
33. De Stefano A, and the GenePD study group. Replication for association of *ELAV4* and Parkinson Disease; the *GenePD* study. **In review, 2008**
34. Latourelle JC, and the GenePD study group. The G2019S mutation in LRKK2 is not fully penetrant: the *GenePD* study. **In Review, 2008**
35. Wang E, Dimova N, and **Cambi, F**. hnRNPH is required for optimal binding of U1 snRNP and regulation of the PLP/DM20 ratio through a complex array of G runs. **In review, 2008**

PEER-REVIEWED INVITED REVIEWS

1. **Cambi F**, Breakefield XO: DNA linkage analysis in the study of inherited neurologic diseases. *Rassegna Clinico-Scientifica Fondazione Lorenzini*. March-April, 1986.
2. Breakefield XO, **Cambi F**: Molecular insights into human neuropathology: selected examples. *Annual Reviews in Neuroscience* 1987;10:535-594.
3. Garbern, J., **Cambi, F**, Kamholz, J., The Molecular Pathogenesis of Pelizaeus-Merzbacher Disease, *Archives of Neurology*, 1999;56:1210-1214.
4. Renieri A, Meloni I, Longo I, Ariani F, **Cambi F**. Rett Syndrome: The Complex Nature of a Monogenic Disease. *J Mol Med* 2003; 81:346-54.

Faculty at University of Kentucky

5. **Cambi, F**. Hereditary Myelopathy. (Chapter 2) *Continuum*, Spinal Cord Disorders, June 2005.
6. Mari F, Nielsen C, **Cambi F**, Speciale C, Mencarelli MA, Renieri A. Genetics and mechanisms of disease in Rett syndrome. Invited review. *Drug Discovery Today: Disease Mechanisms, Volume 2, Issue 4, Winter 2005, 419-425*

SELECTED PEER-REVIEWED ABSTRACTS

1. **Cambi F**, Tang X-M, Cordray P, Fain PR, Keppen L, Barker D. Linkage of pure X-linked spastic paraplegia to Xq22 in two pedigrees. *Neurol.* 1995;45, Supplement.
2. Garbern J., **Cambi F**, et al. Proteolipid protein (PLP) expression is critical for PNS as well as CNS myelination: molecular and pathological characterization of a family with a novel PLP mutation. *Neurology, Supplement*, 1997.
3. **Cambi F**. The same mutation in exon 5 of the proteolipid protein gene causes Pelizaeus-Merzbacher disease in one kindred and pure x-linked spastic paraplegia in an apparently unrelated family with a similar haplotype. *American Academy of Neurology Annual Meeting*, May 5-11, 2001.
4. **Cambi F**, Huang Z, Sperle K. et al. Mutations that reduce PLP-specific splicing affect myelin and axonal integrity. Platform presentation at the American Neurological Association 127th Annual Meeting in NY October 13-16, 2002.
5. **Cambi F**, Huang Z, Rogan P, Sperle K and Hobson G. Characterization of PLP and DM20 splicing in differentiating oligodendrocytes. 6th IBRO (International Brain Research Organization) World Congress of Neuroscience meeting, Prague, July 10-15, 2003
6. **Cambi F**, Williams S, Davis-Williams A, Huang Z and Hobson G. A novel PLP mutation is associated with neurobehavioral phenotype. *American Neurological Association 128th Annual Meeting*, October 18-22, 2003

Faculty, University of Kentucky

7. **Cambi F**, Huang Z and Hobson G. Analysis of splicing factors and PLP splicing in differentiating oligodendrocytes. Myelin Gordon Conference, Il Ciocco, Italy, May 23-28, 2004
8. Sabet A, Jun Li, Ghandour K, Pu Q, Wu X, Kamholz J, Shy M, and **Cambi F**. Novel MPZ Mutation at Splice site in a Family with Hereditary Neuropathy. American Neurological Association 130th Annual Meeting, September 25-28, 2005
9. Wang E, Dimova N, Hobson G, Huang Z, Sperle K, McCullough A, **Cambi F**. PLP1 alternative splicing in differentiating oligodendrocytes: characterization of an exonic splicing enhancer. Poster presentation, Cold Spring Harbor meeting on Eukaryotic mRNA processing, Aug 22-26, 2005
10. Wang E, Dimova N, **Cambi F**. Alternative 5' splice site selection of the PLP/DM20 gene involves synergistic effect of hnRNPH/F in part mediated by a G-rich enhancer. Poster presentation, Cold Spring Harbor meeting on Eukaryotic mRNA processing, Aug 22-26, 2007

BOOK CHAPTERS

Cambi F, et al. Disease Staging: Clinical Criteria, 5th Edition.

International Lectures

Genetic lecture series, Department of Genetics, University of Siena, December 21, 2007. Title: "Tight control of myelin PLP alternative splicing is necessary for brain development and function: evidence from human disease and a novel mouse model".

First ELA symposium: The vital function of myelin: Development and Maintenance. Title: Inhibiting PLP mutations". Paris October 5-7, 2006.

University of Siena, Department of Neurology and Centro Nazionale Ricerca. November 11, 2004. Title: "Pelizaeus-Merzbacher Disease and X-linked Hereditary Spastic Parapareses: Clinical, Genetic and Molecular Studies".

University of Siena, Department of Human Genetics, April 15, 2002. Title: Hereditary Spastic Parapareses: Genetic and Molecular Studies.

Istituto Superiore di Sanita', Rome, Italy, November 28-29, 1996. Progress Report Presentation. Title: "Expression of Cyclins And Their Inhibitors During Oligodendrocyte Differentiation",

University of Bologna, Italy, November 7, 1995. Title: "PLP Gene Regulation in Rat Oligodendrocytes and PLP Mutation Analysis in Human Dysmyelinating Diseases".

Local and National Lectures

Department of Physiology, University of Kentucky, March 14, 2007: "G rich enhancers and hnRNPH and F regulate alternative splicing of the proteolipid protein gene"

Speaker at the 15th Annual meeting of The Myelin Project Work Group, Sept 10-12, 2004. Title "Mutations that affect PLP splicing".

Discussant for a workshop on "treatment modalities for PMD", Washington, March 26-28, 2004.

Platform presentation at the ANA 127th Annual Meeting in NY October 2002. Cambi F, Huang Z, Sperle K. et al. Mutations that reduce PLP-specific splicing affect myelin and axonal integrity.

Speaker at the First International Symposium for Hereditary Spastic Paraplegia, University of Michigan, Ann Arbor, May 25-27, 2000. 2 talks: "HSP due to proteolipoprotein gene mutations" and "X-linked HSP: clinical features"

Seminar, Workshop on Pelizaeus-Merzbacher Disease/X-linked Spastic Paraplegia Type 2, National Institutes of Health, October 14-15, 1999. Title: "Experience with SPG2 in the US".

Talk Entitled: "Abnormalities of the oligodendrocyte and myelin formation", A.I. Dupont, November 15, 1997.

Seminar Series, Department of Cell Biology, University of Delaware, November 13, 1996. Title: "PLP Gene Regulation And Cell Cycle Control of Rat Oligodendrocytes",

Seminar Series, Graduate Program in Developmental Biology and Teratology, Thomas Jefferson University, October 31, 1996. Title: "Oligodendrocyte Differentiation: PLP Transcriptional Regulation And Cell Cycle Control".

The Charcot-Marie Tooth (CMT) Conference at A.I. Dupont, organized by the CMT Association, September 21, 1996. Title: "What's New In CMT Research?"

Talk Entitled: "PLP Gene Regulation In Rat Oligodendrocytes and PLP Mutation Analysis In Human Dysmyelinating Diseases", A.I. Dupont, Department of Clinical Science, October 3, 1995.

The Medical Center of Delaware, Neuroscience Conference, Christiana Hospital, August 3, 1995. Title: "Neuro Degenerative Diseases Caused By Triplet Repeats: The Price of DNA Amplification",

Seminar, the Medical Center of Delaware, Neuroscience Conference, Christiana Hospital, October 6, 1994. Title: "X-Linked Pure Spastic Paraparesis: Linkage Analysis in a Large Pedigree".

Seminar, Neuroscience Series, The Nathan-Kline Institute, September 27, 1994. Title: "Transcriptional Regulation of PLP Promoter and Analysis of PLP Mutation in X-Linked Hereditary Spastic Paraparesis",

Seminar, Molecular Biology Seminars Series at Wayne State University, September 12, 1994. Title: "Transcriptional Regulation of PLP Promoter and Analysis of PLP Mutation in X-Linked Hereditary Spastic Paraparesis",

Seminar, the Medical Center of Delaware, Neuroscience Conference, Christiana Hospital, October 7, 1993.: "X-Linked Pure Spastic Paraparesis: Characterization of a Large Pedigree",

Grand Rounds in the Department of Neurology at the University of Kentucky:

January 31, 2007: "Genetics & Therapeutics of Parkinson Disease"

November 8, 2006: "Inherited Disorders of Myelin: Genetic Studies & Therapeutic Approaches"

January 2005: X-linked Spastic Paraplegia: Clinical, Genetic & Molecular Studies

August 4, 2005: "Case Presentation/CPC: A 60 year old man with rapid memory decline & unsteady gait"

June 10, 2004: "Pelizaeus-Merzbacher Disease: The Complexity of a Monogenic Disorder"

September 2, 2004: "Families with Spastic Paraparesis: Phenotypes & Genetic Testing"

Grand Rounds (Faculty at Thomas Jefferson University)

MCP Hahnemann University, Department of Neurology, February 8, 2002. Title: X-linked Spastic Parapareses: Genetic and Molecular Studies.

Thomas Jefferson University, Department of Neurology, March 30, 2001. Title: PLP splicing defects in human disease.

Thomas Jefferson University, Department of Neurology, December 6, 2000. Title: X-linked Spastic Parapareses: Genetic and Molecular Studies.

Thomas Jefferson University, Department of Neurology, "Genetics of Spino Cerebellar Degeneration", September 24, 1999.

Thomas Jefferson University, Department of Neurology, "Myelination: Molecular and Clinical Aspects", September 19, 1997.

Thomas Jefferson University, Department of Neurology. Title: "Spinocerebellar Degeneration: Clinical and Genetic Aspects", May 2, 1997.

Thomas Jefferson University, Department of Neurology, July 7, 1995. Title: "Neurogenetics 101",

Thomas Jefferson University, Department of Neurology, December 16, 1994. Title: "Neurogenetics 101"

Mercy Hospital, Scranton, PA, December 7, 1994. Title: "Genetic Studies In Neurological Disorders: A Review",

Thomas Jefferson University, Department of Neurology, October 7, 1994. Title: "Analysis of PLP Mutations In X-Linked Spastic Paraplegia".