

BIOGRAPHICAL SKETCH

Provide the following information for the key personnel in the order listed for Form Page 2.

Follow the sample format for each person. **DO NOT EXCEED FOUR PAGES.**

NAME	Professor, Departments of Neurology and Molecular & Medical Genetics		
Patricia L. Kramer, Ph.D			
EDUCATION/TRAINING (Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)			
INSTITUTION AND LOCATION	DEGREE (if applicable)	YEAR(s)	FIELD OF STUDY
Portland State University, Portland, Oregon	B.A.	1969-72	Anthropology
University of Oregon, Eugene, Oregon	M.S.	1973-76	Anthropology
University of New Mexico, Albuquerque, New Mexico	Ph.D.	1976-79	Anthropology
University of Pavia, Pavia, Italy	PostdocFellow	1980-81	Population Genetics
Yale University School of Medicine, New Haven, Ct	PostdocFellow	1982-86	Population Genetics

RESEARCH AND PROFESSIONAL EXPERIENCE: Concluding with present position, list, in chronological order, previous employment, experience, and honors. Include present membership on any Federal Government public advisory committee. List, in chronological order, the titles, all authors, and complete references to all publications during the past three years and to representative earlier publications pertinent to this application. If the list of publications in the last three years exceeds two pages, select the most pertinent publications.

Positions and Honors:

1984	Visiting Lecturer, Department of Anthropology, Yale University
1985-86	Assistant Research Scientist, Human Genetics Department, Yale University
1988-1994	Assistant Professor, Department of Neurology, Oregon Health and Science University
1991-1994	Assistant Professor, Department of Molecular & Medical Genetics, OHSU
1994-2001	Associate Professor, Departments of Neurology and Molecular & Medical Genetics
2001-present	Professor, Departments of Neurology and Molecular & Medical Genetics, OHSU
2002-present	Genetics Core Leader, Aging and Alzheimer's Center, OHSU

Selected peer-reviewed publications:

- Kramer PL**, de Leon D, Ozelius L, Risch N, Bressman S, Brin M, Schuback D, Burke R, Kwiatkowski D, Shale H, Gusella J, Breakefield XO, Fahn S. Dystonia gene in Ashkenazi Jewish population located on chromosome 9q32-34. *Ann Neurol* 27:114-120, 1990.
- Kramer P**, Heiman G, Gasser T, Ozelius L, de Leon D, Brin M, Burke R, Hewett J, Hunt A, Moskowitz C, Nygaard T, Wilhelmsen K, Fahn S, Breakefield X, Risch N, Bressman S. The DYT1 gene on 9q34 is responsible for most cases of early-onset Idiopathic Torsion Dystonia (ITD) in non-Jews. *Am J Hum Genet* 55:468-475, 1994.
- Risch N, de Leon D, Ozelius L, **Kramer P**, Almsy L, Singer B, Fahn S, Breakefield XO, Bressman S. Genetic analysis of idiopathic torsion dystonia in Ashkenazi Jews: evidence for the recent descent of Ashkenazim from a small founder population. *Nat Genet* 9:152-159, 1995.
- Kramer PL**, Yount JL, Mitchell TN, Lovrien EW, La Morticella DM, Maumenee IH, Litt M. A second gene for cerulean cataracts maps to the CRYB 2 region on chromosome 22. *Genomics*, 35:539-542, 1996.
- Taylor T, Litt M, **Kramer P**, Pandolfo M, Angelina L, Nardocci N, Davis S, Pineda M, Hattori H, Flett P, Cilio M, Bertini E, Hayflick S. Homozygosity Mapping of Hallervorden-Spatz Syndrome to Chromosome 20p12.3-p13. *Nat Genet*, 14:479-481, 1996.
- Litt M, Carrero-Valenzuela R, LaMorticella DM, Schultz DW, Mitchell TN, **Kramer P**, Maumenee I. Autosomal dominant cerulean cataract is associated with a chain termination mutation in the human beta crystallin gene CRYBB2. *Hum Mol Genet*, 6:665-668, 1996.

- Bressman SB, de Leon D, Raymond MS, Greene PE, Brin MF, Fahn S, Ozelius LJ, Breakefield XO, **Kramer PL**, Risch NJ. The Role of the DYT1 Gene in Secondary Dystonia. *Neurology*, 48:1571-1577, 1997.
- Wirtz MK, Samples JR, **Kramer PL**, Rust K, Topinka JR, Yount J, Koler RD, Acott TS. Mapping a gene for adult-onset primary open-angle glaucoma to chromosome 3q. *Am J Hum Genet*, 60(2):296-304, 1997.
- Ozelius L, Hewett J, Page CE, Bressman S, **Kramer P**, Shalish C, de Leon D, Brin M, Raymond D, Corey DP, Fahn S, Risch N, Buckler A, Gusella J, Breakefield XO. The early-onset torsion dystonia gene (DYT1) encodes an ATP-binding protein. *Nat Genet*, 17:40-47, 1997.
- Almasy L, Bressman SB, **Kramer PL**, Yount J, Greene PE, Heiman GA, Ford B, Raymond D, de Leon D, Jones AC, Hong Shen, Fahn S, Risch NJ, Nygaard TG. Idiopathic torsion dystonia linked to chromosome 8 in two families of German Mennonite origin. *Ann Neurol*, 42:670-673, 1997.
- Litt M, **Kramer P**, LaMorticella DM, Murphey W, Lovrien EW, Weleber RG. Autosomal dominant congenital cataract associated with a missense mutation in the human alpha crystallin gene CRYAA. *Hum Mol Genet*, 7(3):471-474, 1998.
- Klein C, Pramstaller P, Castellan C, Breakefield XO, **Kramer P**, Ozelius LJ. Clinical and genetic evaluation of a family with a mixed dystonia phenotype from South Tyrol. *Ann Neurol*, 44:394-398, 1998.
- Wirtz MK, Samples JR, Rust KR, Lie J, Nordling L, Schilling K, Acott TS, **Kramer PL**, GLC1F, A new primary open-angle glaucoma locus, maps to 7q35-q36. *Arch Ophthalmol*, 117:237-241, 1999.
- Klein C, Brin MF, **Kramer PL**, Sena Esteves M, de Leon D, Doheny D, Bressman S, Fahn S, Breakefield XO, Ozelius LJ. Association of a missense change in the D2 dopamine receptor with myoclonus-dystonia. *PNAS*, 96(9):5173-5176, 1999.
- Kramer PL**, Mineta M, Klein C, Schilling K, de Leon D, Farlow MR, Breakefield XO, Dobyns WB, Ozelius LJ, Brashear A. Rapid-onset Dystonia-Parkinsonism: Linkage to chromosome 19q13. *Ann Neurol*, 46:176-182, 1999.
- Nygaard TG, Raymond D, Chen C, Nishino I, Greene PE, Jennings D, Heiman GA, Klein C, Saunders-Pullman RJ, **Kramer PL**, Ozelius LJ, Bressman SB. Localization of a gene for myoclonus-dystonia to chromosome 7q21-q31. *Ann Neurol* 46:794-798, 1999.
- Kramer PL**, LaMorticella D, Schilling K, Billingslea A, Weleber RG, Litt M. A new locus for autosomal dominant congenital cataracts maps to chromosome 3. *Inv Ophthalmol Vis Sci*, 41:36-39, 2000.
- Jakobs PM, Hess JF, FitzGerald PG, **Kramer PL**, Weleber RG, Litt M. Autosomal-Dominant Congenital Cataract Associated with a Deletion Mutation in the Human Beaded Filament Protein BFSP2. *Am J Hum Genet* 66(4):1432-1436, 2000.
- Bressman SB, Sabatti C, Raymond D, de Leon D, Klein C, **Kramer PL**, Brin MF, Fahn S, Breakefield X, Ozelius LJ, Risch NJ. The *DYT1* phenotype and guidelines for diagnostic testing. *Neurology* 54:1746-1752, 2000.
- Klein C, Pramstaller PP, Kis B, Page CC, Kann M, Leung J, Woodward H, Castellan CC, Scherer M, Vieregge P, Breakefield XO, **Kramer PL**, Ozelius LO. *Parkin* deletions in a family with adult-onset, tremor-dominant parkinsonism: expanding the phenotype. *Ann Neurol*, 48:65-71, 2000.
- Klein C, Schumacher K, Jacobs H, Hagenah J, Kis B, Garrels J, Schwinger E, Ozelius L, Pramstaller P, Vieregge P, **Kramer PL**. Association studies of Parkinson's disease and parkin polymorphisms. *Ann Neurol*, 48:126-127, 2000.
- Klein C, Schilling K, Saunders-Pullman R, Garrels J, Breakefield X, Brin M, deLeon D, Doheny D, Rahn S, Fink J, Forsgren L, Friedman J, Frucht S, harris J, Holmgren G, Kis B, Furlan R, Kyllerman M, Land A, Leung J, Raymond D, Rogishaw J, Sanner G, Schwinger R, Tabame R, Tagliati M, Vieregge P, Wahlstrom J, Wendt K, **Kramer PL**, Bressman SB, Ozelius LJ. A major locus for myoclonus-dystonia maps to chromosome 7q in eight families. *Am J Hum Genet*, 67:1314-1319, 2000.
- Bressman SB, Fahn S, Ozelius LJ, **Kramer PL**, Risch NJ. The DYT1 mutation and nonfamilial primary torsion dystonia. *Arch Neurol*, 58:681-2, 2001.
- Kitsos G, Eiberg H, Economou-Petersen E, Wirtz M, **Kramer PL**, Aspiotis M, Tommerup N, Petersen MB, Psilas K. Genetic linkage of autosomal dominant primary open angle glaucoma to chromosome 3q in a Greek pedigree. *Eur J Hum Genet*, 9(6):452-457, 2001.
- Leung JC, Klein C, Friedman J, Vieregge P, Jacobs H, Doheny D, Kamm C, DeLeon D, Pramstaller PP, Penney JB, Eisengart M, Kankovis J, Gasser T, Bressman SB, Corey DP, **Kramer P**, Brin MF, Ozelius LJ, Breakefield XO. Novel mutation in the *TOR1A (DYT1)* gene in atypical, early onset dystonia and polymorphisms in dystonia and early onset parkinsonism. *Neurogenetics*, 3(3):133-143, 2001.

- Pramstaller P, Kis B, Eskelson C, Hedrich K, Scherer M, Schwinger E, Vieregge P, Breakfield XO, **Kramer PL**, Ozelius L, Klein C. Phenotypic variability in a large kindred with deletions in the parkin gene. *Mov Disord*, 17(2):424-426, 2002.
- Saunders-Pullman R, Shriberg J, Heiman G, Raymond D, Wendt K, **Kramer P**, Schilling K, Kurlan R, Klein C, Ozelius LJ, Risch N, Bressman S. The spectrum of myoclonus-dystonia: possible association with OCD and alcohol dependence. *Neurology*, 58:242-245, 2002.
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- Klein C, Liu L, Doheny D, Kock N, Müller B, de Carvalho Aguiar P, Leung J, de Leon D, Bressman SB, Silverman J, Smith C, Danisi F, Morrison C, Walker RH, Velickovic M, Schwinger E, **Kramer PL**, Breakefield XO, Brin MF, Ozelius LJ. ϵ -sarcoglycan mutations found in combination with other dystonia gene mutations. *Ann Neurol* 2002;52:675-679.
- Kock, N, Culjkovic B, Maniak S, Schilling K, Muller B, Zuhlke C, Ozelius L, Klein C, Pramstaller P, **Kramer PL**. Mode of inheritance and susceptibility locus for Restless Legs Syndrome on chromosome 12q. *Am J Hum Genet*, 71:205-208, 2002.
- Foncke EMJ, Klein C, Koelman JHTM, **Kramer PL**, Schilling K, Muller B, Garrels J, de Carvalho Aguiar P, Liu L, de Froe A, Speelman JD, Ozelius LJ, Tijssen MAJ. Hereditary myoclonus-dystonia associated with epilepsy. *Neurology*, 60:1988-1990, 2003.
- Kramer PL**, Wilmot B, O'Malley JP, Fife DJ, Sheridan B, Murdoch G, Nagalla S, Kaye J. Effect of Phenotypic Definition on Gene Expression in Alzheimer's Disease. *Neurol* 60(Suppl 1):A454, 2003.
- Wilmot B, Sheridan B, O'Malley JP, Fife DJ, Murdoch G, Nagalla S, **Kramer PL**, Kaye J. Differential Expression of Genes Involved in p53 Activation and Stabilization in Aging and Alzheimer's Disease Brain. *Neurol* 60(Suppl 1):A68, 2003.
- Wilmot B, Sheridan B, Fife DJ, O'Malley JP, Murdoch G, Nagalla S, Kaye J, **Kramer PL**. Effect of Apolipoprotein E Genotype on Differential Gene Expression in Control and Alzheimer's Disease Brain. *Neurol* 60(Suppl 1):A455, 2003.

Research Support (last three years):

ONGOING

R01NS26656, P.I. on OHSU subcontract	9/1/88-8/31/04	20%
NIH/NINDS	\$44,000 (OHSU subcontract)	
<i>The Clinical-Genetic Spectrum of Classic Dystonia</i>		
The major goals of this project are to (1) locate non- <i>DYT1</i> genes for dystonia and characterize their clinical features, and (2) investigate explanations for variable phenotypic expression in dystonia.		

R01EY11650, Co-Investigator	8/1/97-7/31/03	
NIH/NEI	\$167,000	20%
<i>Genetics of Adult-Onset Primary Open-Angle Glaucoma</i>		
The goals of this project are to locate and characterize genes for primary open-angle glaucoma		

5-P30-AG08017-13, Genetics Core Leader	07/06/02-03/31/05	40%
NIH/NIA	\$271,057	
<i>Oregon Alzheimer's Disease Center, Genetics Core</i>		
The goals of this project are to collect family history information and biological sample data for subjects in the Alzheimer's Disease Center		

COMPLETED:

N/A, P.I.	3/1/01-2/28/03
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Dystonia Medical Research Foundation \$30,990
Analysis of Modifying Genes in Early Onset Dystonia
The goal is to identify genes that modify penetrance of the GAG deletion in *DYT1*-related dystonia.

N/A, P.I. 3/1/02-2/28/03
Dystonia Medical Research Foundation \$50,000
Development and Maintenance of the Clinical/Genetic Database for Dystonia
The purpose is to maintain a centralized database for clinical and genetic marker data generated by dystonia gene mapping collaborators. It also serves as the foundation for linkage and statistical analyses conducted at OHSU

N/A, P.I. 3/1/00-8/31/01
Alzheimer's Research Alliance of Oregon \$24,000
Identification of genes involved in healthy aging using DNA microarray technology
The major goal was to identify genes involved in Alzheimer's disease using DNA microarray data

R01NS38142, P.I. on OHSU subcontract 4/1/98-3/31/01
NIH/NINDS \$65,000 (OHSU subcontract)
Role of the torsin gene family in penetrance of DYT1 and other dystonias
The aims of this grant were to characterize the *DYT1* gene and determine genetic factors that affect penetrance.