

**CURRICULUM VITAE**

**PARVONEH POORKAJ NAVAS, PhD**

**PERSONAL DATA**

Present Address: University of Washington  
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Citizenship: United States of America

**EDUCATION**

1981-1985 University of California at Los Angeles, Los Angeles, CA  
B.S. in Biology

1985-1992 University of California at Los Angeles, Los Angeles, CA  
Ph.D. in Biology

**POSTGRADUATE TRAINING**

1992-1994 Senior Fellow, Division of Medical Genetics,  
Department of Medicine, University of Washington, Seattle, WA 98195  
Research Advisor: James P. Evans, M.D., Ph.D.

1994-1998 Senior Fellow, Division of Gerontology and Geriatric Medicine  
University of Washington and  
Geriatric Research Education Clinical Center (GRECC)(182B),  
Veterans Affairs Puget Sound Health Care System, Seattle, WA 98195  
Research Advisor: Gerard D. Schellenberg, Ph.D.

**FACULTY POSITIONS**

1998- 2002 Acting Assistant Professor  
Division of Gerontology and Geriatric Medicine  
Department of Medicine, University of Washington, Seattle, WA 98195

2002- 2003 Research Assistant Professor  
Division of Gerontology and Geriatric Medicine  
Department of Medicine, University of Washington, Seattle, WA 98195

2003-2005 Research Assistant Professor  
Department of Psychiatry and Behavioral Sciences  
University of Washington, Seattle, WA 98195

2004-2005 Adjunct Research Assistant Professor

Division of Medical Genetics, Department of Medicine  
University of Washington, Seattle, WA 98195

2005-2008      Assistant Professor  
Department of Psychiatry and Behavioral Sciences  
Adjunct Assistant Professor Division of Medical Genetics,  
Department of Medicine, University of Washington, Seattle, WA 98195

2008-present      Research Associate Professor  
Department of Psychiatry and Behavioral Sciences  
Adjunct Assistant Professor Division of Medical Genetics,  
Department of Medicine, University of Washington, Seattle, WA 98195

### **HONORS AND AWARDS**

1990-1991      Dr. Ursula Mandel Fellowship  
University of California, Los Angeles

1989-1990      Dr. Ursula Mandel Fellowship  
University of California, Los Angeles

### **PROFESSIONAL AFFILIATIONS**

Member, American Association for the Advancement of Science (AAAS)

Member, Society for Neuroscience

Member, American Society of Human Genetics

### **TEACHING / MENTORING RESPONSIBILITIES**

#### **PBSCI 499 - Undergraduate Independent Research Mentorship:**

2004-2006      Nayiri Gazi  
2005              Grace (Jiwon) Lee  
2005-2007      Karen (Yoonjung) Chang  
2007-2008      Sidney Law

#### **Mary Gates Research Scholar's Mentorship:**

2004-2005      Tiffany A. Stager  
2004-2006      Sophia (JiHae) Kim

#### **UW GenOM Project Mentorship:**

2005              Katherine E. Taylor  
2006-2007      Harkirat Sohi

#### **Other Research Teaching:**

2002-2003      Veteran's Affairs Puget Sound Health Care System, Genetics Training Course  
"Translational Research in Alzheimer's disease and Neurodegeneration: From the

Bench to the Clinic”. Lecture (one of ten in series): Molecular Genetics II: transgenic models and tau.

2008 Department of Psychiatry R3 Didactic Neuroscience Series Lecture: The Aging Brain- “Neurological disease and the senescing brain: A shorter path to the same destination?”

## **SERVICE**

### **Ad Hoc reviewer for the following journals:**

Annals of Neurology  
Nucleic Acids Research  
Neuroscience Letters  
Proceedings of the National Academy of Sciences

### **National responsibilities:**

#### Ad Hoc Grant Reviewer

2002, 2007 The Oesterreichische Nationalbank, Austria  
2005, 2007-2009 National Alzheimer’s Association

#### Center for Scientific Review External Grant Reviewer, NIH

2009 Special Emphasis panel ZRG1 BDA-A (52)

### **Local responsibilities:**

#### Committees

2004-2007 Member, Faculty Council on University Relations, University of Washington  
2005-2008 Member, Faculty Senate, University of Washington  
2006-2009 Member, Faculty Council for Women in Academia, University of Washington

#### Ad Hoc Grant Reviewer

2007 Veteran’s Affairs Puget Sound Healthcare System, Research program review  
2001-2008 University of Washington, Alzheimer’s Disease Research Center Pilot Award  
2004 University of Washington, Royalty Research Foundation

#### Departmental Services

2005-2010 UW Psychiatry Residency Training Program Interviews

## **RESEARCH FUNDING**

### Completed

Project Title: Spinocerebellar-Ataxia Type 14: Mouse models of human disease.

University of Washington Royalty Research Fund Pilot Project Grant, Navas PI, 7/16/08-7/15/09.  
Total Direct Costs: \$39,926

UW Provost Bridge Funding 1/1/08-12/31/08  
Total Direct Costs: \$10,000

Project Title: Genetic interactions in Progressive Supranuclear Palsy. 5K01AG024329-02  
NIH/NIA. Navas PI, 6/1/05-5/31/08.  
Total Direct Costs: \$351,509

Project Title: Parkin and Tau mutational effects on tangle formation in PSP. Society for  
Progressive Supranuclear Palsy, Private Foundation Grant. Navas PI, 7/01/06-9/30/07.  
Total Direct Costs: \$50,000

Project Title: Genomic mouse models of Spinocerebellar Ataxia, National Ataxia Foundation,  
Navas PI, 1/1/06-3/31/07.  
Total Direct Costs: \$20,000

Project Title: BAC transgenic mouse models of Spinocerebellar Ataxia type 14, National  
Organization of Rare Disorders (NORD), Navas PI, 10/1/05-1/27/07.  
Total Direct Costs: \$30,000.

Project Title: Alzheimer's disease and Tau gene regulation. Alzheimer's disease Related  
Research Grant, Friends of Alzheimer's Research, Department of Medicine, University of  
Washington, Navas PI, 1/1/04-12/31/05.  
Total Direct Costs: \$100,000

Project Title: Parkin mutations in a mouse model of PSP, Society for Progressive Supranuclear  
Palsy, Navas PI. 3/01/04-2/28/05.  
Total Direct Costs: \$44,000

Project Title: Candidate genes for Parkinson's disease with variably penetrant spasticity.  
University of Washington Royalty Research Fund Pilot Project Grant, Navas PI, 3/1/04-2/28/05.  
Total Direct Costs: \$37,000

Project Title: Molecular and genomic analysis of the parkin gene. PADRECC Pilot Project Grant,  
Veterans Affairs Administration, T.D. Bird and Navas, Co-PI's, 3/1/01- 9/28/03.  
Total Direct Costs: \$100,000

Project Title: Identification of the Progressive Supranuclear Palsy gene, Society for Progressive  
Supranuclear Palsy Private Foundation Grant, Navas PI, 3/1/99-2/28/00.  
Total Direct Costs: \$50,000

Project Title: Analysis of tau gene regulation and expression, Pilot Project Grant University of  
Washington Alzheimer's disease Research Center, Navas PI, 8/1/99-7/31/00.  
Total Direct Costs: \$30,000

**Co-Investigator Funding:**  
Completed

Project Title: Genetic analysis of onset age of Parkinson's disease, 2 R01 NS36960-04, NIH, Payami PI, Sub-contract Schellenberg PI, 4/1/02-3/28/06.

Total Direct Costs: \$87,000

Project Title: Project 1, Genetic studies of familial Alzheimer's disease, 5 P50 AG 05136-17, NIA, Raskind PI, 5/1/00-4/30/05.

Total Direct Costs: \$201,026

Project Title: Project 2, Tauopathies: genotype and phenotype, PO1 AG17586-01, NIA, Lee PI, 3/15/00-2/28/05.

Total Direct Costs: \$160,497

Project Title: Genomic analysis of Alzheimer's disease, 1 RO1 AG 11762-06, NIA, Schellenberg PI, 6/1/94-5/31/03.

Total Direct Costs: \$170,476

## **BIBLIOGRAPHY**

### **Publications in refereed journals**

1. Scherer, S.W., **Poorkaj, P.**, Allen, T., Kim, J., Geshuri, D., Nunes, M., Soder, S., Stephens, K., Pagon, R., Patton, M., Berg, M.A., Donlon, T., Rivera, H., Pfeiffer, R.A., Naritomi, K., Hughes, H., Genuardi, M., Gurrieri, F., Neri, G., Lovrein, E., Magenis, E., Tsui, L-C. and Evans, J.P. (1994) Fine mapping of the autosomal dominant split hand/split foot locus on chromosome 7, band q21.3-q22.1. Am. J. Hum. Genet. (55)1:12-20.
2. Scherer, S.W., **Poorkaj, P.**, Massa, H., Soder, S., Allen, T., Nunes, M., Geshuri, D., Wong, E., Belloni, E., Little, S., Zhou, L., Becker, D., Kere, J., Ignatius, J., Niikawa, N., Fukushima, Y., Hasegawa, T., Weissenbach, J., Boncinelli, E., Trask, B., Tsui, L-C. and Evans, J.P. (1994) Physical mapping of the split hand/split foot locus on chromosome 7 and implication in syndromic ectrodactyly. Human Molecular Genetics (3)8: 1345-1354.
3. Levy-Lahad, E., Wasco, W., **Poorkaj, P.**, Romano, D.M., Oshima, J., Pettingell, W.H., Yu, C-E., Jondro, P.D., Schmidt, S.D., Wang, K., Crowley, A.C., Fu, Y-H., Guenette, S.Y., Galas, D., Nemens, E., Wisjman, E., Bird, T.D. Schellenberg, G.D., and Tanzi, R.E. (1995) Candidate gene for the chromosome-1 familial Alzheimer's disease locus. Science 269: 973-977.
4. Cascarano, J., Chambers, P.A., Schwartz, E., **Poorkaj, P.** and Gondo, R.E. (1995) Organellar clusters formed by mitochondrial-rough endoplasmic reticulum associations: an ordered arrangement of mitochondria in hepatocytes. Hepatology (22)3: 837-846.
5. Levy-Lahad, E., **Poorkaj, P.**, Wang, K., Fu, Y.H., Oshima, J., Mulligan, J. and Schellenberg, G.D. (1996) Genomic structure and expression of STM-2, the chromosome 1 familial Alzheimer disease gene. Genomics (34)2: 198-204.
6. Scheuner, D., Eckman, C., Jensen, M., Song, X., Citron, M., Suzuki, N., Bird, T.D., Hardy, J., Hutton, M., Kukull, W., Larson, E., Levy-Lahad, E., Viitanen, M., Peskind, E., **Poorkaj, P.**, Schellenberg, G., Tanzi, R., Wasco, W., Lannfelt, L., Selkoe, D. and Younkin, S.

- (1996) Secreted amyloid- $\beta$  protein similar to that in the senile plaques of Alzheimer's disease is increased *in vivo* by the presenilin 1 and 2 and APP mutations linked to familial Alzheimer's disease. Nature Medicine (2)8: 864-870.
7. Mann, D.M.A., Iwatsubo, T., Cairns, N.J., Lantos, P.L., Nochlin, D., Sumi, S.M., Bird, T.D., **Poorkaj, P.**, Hardy, J., Hutton, M., Prihar, G., Crook, R., Rossor, M.N. and Haltia, M. (1996) Amyloid  $\beta$  protein (A $\beta$ ) deposition in chromosome 14-linked Alzheimer's disease: Predominance of A $\beta$ <sub>42(43)</sub>. Annals of Neurology (40)2: 149-156.
  8. Crackower, M.A., Scherer, S.W., Rommens, J.M., Hui, C.C., **Poorkaj, P.**, Soder, S., Cobben, J.M., Buys, C., Hudgins, L., Evans, J.P. and Tsui, L.-C. (1996) Characterization of the split hand/split foot malformation locus SHFM1 at 7q21.3-q22.1 and analysis of a candidate gene for its expression during limb development. Human Molecular Genetics (5)5: 571-579.
  9. McMillan, P.J., Leverenz, J.B., **Poorkaj, P.**, Schellenberg, G.D. and Dorsa, D.M. (1996) Neuronal expression of STM2 mRNA in human brain is reduced in Alzheimer's disease. Journal of Histochemistry and Cytochemistry (44)11: 1215-1222.
  10. Murphy, G.M., Forno, L.S., Ellis, W.G., Nochlin, D., Levy-Lahad, E., **Poorkaj, P.**, Bird, T.D., Jiang, Z. and Cordell, B. (1996) Antibodies to presenilin proteins detect neurofibrillary tangles in Alzheimer's disease. American Journal of Pathology (149)6:1839-1846.
  11. Ikeda, M., Sharma, V., Sumi, S.M., Rogaeva, E.A., **Poorkaj, P.**, Sherrington, R., Nee, L., Tsuda, T., Oda, N., Watanabe, M., Aoki, M., Shoji, M., Abe, K., Itoyama, Y., Hirai, S., Scellenberg, G.D., Bird, T.D. and St George-Hyslop, P.H. (1996) The clinical phenotype of two missense mutations in the presenilin 1 gene in Japanese patients. Annals of Neurology (40)6: 912-917.
  12. Bird, T.D., Levy-Lahad, E., **Poorkaj, P.**, Sharma, V., Nemens, E., Lampe, T. and Schellenberg, G. (1996) Wide range in age of onset for chromosome 1-related familial Alzheimer's disease. Annals of Neurology (40)6: 932-936.
  13. Bird, T.D., Wijsman, E.M., Nochlin, D., Leehey, M., Sumi, S.M., Payami, H., **Poorkaj, P.**, Nemens, E., Raskind, M. and Schellenberg, G.D. (1997) Chromosome 17 and hereditary dementia: linkage studies in three non-Alzheimer families and kindreds with late-onset FAD. Neurology (48)4: 949-954.
  14. **Poorkaj, P.**, Sharma, V., Anderson, L., Nemens, E., Alonso, M.E., Orr, H., White, J., Heston, L., Bird, T.D. and Schellenberg, G.D. (1998) Missense mutations in the chromosome 14 familial Alzheimer's Disease presenilin 1 gene. Human Mutation 11:216-221.
  15. Sharma, V., **Poorkaj, P.**, Hisama, F., Bonnycastle, L., Yu, C-E., Massa, H., Trask, B., Clancy, K.P., Patterson, D., Weissman, S. and Schellenberg, G.D. (1998) An expression map from human chromosome 14q24.3. Genomics (47)2: 314-318.
  16. **Poorkaj, P.**, Wijsman E., Nemens, E., Anderson L., Andreadis, A., Bird, T.D. and Schellenberg, G.D. (1998) Tau is a candidate gene for chromosome 17 frontotemporal dementia. Annals of Neurology 43: 815-825.
  17. Clark, L.N., **Poorkaj, P.**, Wszolek, Z., Geschwind, D. H., Nasreddine Z.S., Miller, B., Li, D., Payami, H., Awert, F., Markopoulou, K., Andreadis, A., D'Souza, I., Lee, V. M.-Y.,

- Reed, L., Trojanowski, J.Q., Zhukareva, V., Bird, T., Schellenberg, G.D. and Wilhelmsen, K.C.(1998) Pathogenic implications of mutations in the tau gene in Pallido-Ponto-Nigral degeneration and related neurodegenerative disorders linked to chromosome-17. Proc. Nat. Acad. Sciences (95)22: 13103-13107.
18. Holden, H., Rizzu, P., Stevens, M., de Knijff, P., van Duijn, C.M., van Swieten, J.C., Heutnik, P., Perez-Tur, J., Thomas, V., Baker, M., Morris, H., Rossor, M., Janssen, J.C., Peterson, R.C., Dodd, P., Dark, F., Boeve, B., Dickson, D., Davies, P., Pickering-Brown, S., Mann, D., Adamson, J., Lynch, T., Payami, H., **Poorkaj, P.**, Bird, T.D., Schellenberg, G.D., Chakraverty, S., Norton, J., Morris, J.C., Goate, A., Hutton, M. and Hardy, J. (1999) Apolipoprotein E genotype does not affect the age of onset of dementia in families with defined tau mutations. Neuroscience Letters. 260: 193-195.
  19. Iijima, M., Tabira, T., **Poorkaj, P.**, Schellenberg, G.D., Trojanowski, J.Q., Lee, V. M.-Y., Schmidt, M.L., Takahashi, K., Nabika, T., Matsumoto, T., Yamashita, Y., Yoshioka, S. and Ishino, H. (1999) A distinct familial presenile dementia with a novel missense mutation in the tau gene. Neuroreport. 10: 497-501.
  20. Bird, T.D., Nochlin, D., **Poorkaj, P.**, Cherrier M., Kaye, J., Payami, H., Peskind, E., Lampe, T. H., Nemens, E., Vonsattel, J.P. and Schellenberg G.D. (1999) A clinical pathological comparison of three families with frontotemporal dementia and identical mutations in the tau gene (P301L). Brain. (122)4: 741-756.
  21. D'Souza, I.D., **Poorkaj, P.**, Hong, M., Nochlin, D., Lee, V. M.-Y., Bird, T.D. and Schellenberg, G.D. (1999) Missense and silent tau gene mutations cause frontotemporal dementia with Parkinsonism - chromosome 17 type by affecting multiple alternative RNA splicing regulatory elements. Proc. Natl. Acad. Sci. USA. 96: 5598-5603.
  22. Yasuda, M., Kawamata, T., Kuno, S., Komure, O., Kuno, S., D'Souza, I., **Poorkaj, P.**, Kawai, J., Tanimukai, S., Yamamoto, Y., Hasagawa, H., Sasahara, M., Hazama, F., Schellenberg, G.D. and Tanaka, C. (1999) A mutation in the microtubule associated protein tau in Pallido-Nigro-Luysian degeneration. Neurology 53: 864-868.
  23. Furukawa, K., D'Souza, I, Crudder C.H., Onedera, H., Itoyama, Y., **Poorkaj, P.**, Bird, T.D. and Schellenberg, G.D. (2000) Pro-apoptotic effects of tau mutations in chromosome 17 frontotemporal dementia and parkinsonism. NeuroReport. (11)1: 57-60.
  24. Kertesz, A., Kawarai, T., Rogaeva, E., St. George-Hyslop, P., **Poorkaj, P.**, Bird, T.D. and Munoz, D. G. (2000) Familial frontotemporal dementia with ubiquitin positive, tau negative inclusions. Neurology (54)4: 818-27.
  25. Yasuda, M., Takamatsu, J., D'Souza, I., Crowther, A., Kawamata, T., Hasegawa, M., Hasegawa, H., Spillantini, M.G., Tanimukai, S., **Poorkaj, P.**, Varani, L., Varani, G., Iwatsubo, T., Goedert, M., Schellenberg, G.D. and Tanaka, C. (2000) A novel mutation at position +12 in the intron following exon 10 of the tau gene in familial frontotemporal dementia (FTD-Kumamoto). Annals of Neurology (47)4: 422-429.
  26. **Poorkaj, P.**, Peterson, K.R. and Schellenberg, G.D. (2000) Single-step conversion of P1 and P1 artificial chromosome clones into yeast artificial chromosomes. Genomics. 68: 106-110.

27. **Poorkaj, P.** Grossman, M., Steinbart, E., Payami, H., Sadovnick, D., Nochlin, D., Finch, C., Tabira, T., Trojanowski, J., Borson, S., Galasko, D., Reich, S., Quinn, B., Schellenberg G.D. and Bird, T. (2001) Frequency of tau gene mutations in familial and sporadic cases of non-Alzheimer dementia. Archives of Neurology. (58)3: 383-387.
28. **Poorkaj, P.**, Kas, A., D'Souza, I., Zhou, Y., Pham, Q., Olson, M.V. and Schellenberg, G.D. (2001) A genomic sequence analysis of the mouse and human microtubule-associated protein tau. Mammalian Genome. 12: 700-712.
29. Steinbart, E.J., Smith, C.O., **Poorkaj, P.** and Bird, T.D. (2001) Impact of DNA testing for early-onset familial Alzheimer's disease and frontotemporal dementia. Arch Neurol. (58)11: 1828-1831.
30. **Poorkaj, P.**, Tsuang, D., Wijsman, E., Steinbart, E., Garruto, R.M., Craig, U., Chapman, N.H., Anderson, L., Bird, T.D., Plato, C. C., Perl, D. P., Weiderholt, W., Galasko, D. and Schellenberg, G.D. (2001) TAU as a susceptibility gene for amyotrophic lateral sclerosis-parkinsonism dementia complex of Guam. Archives of Neurology. (58)11: 1871-1878.
31. **Poorkaj, P.**, Muma, N.A., Zhukareva, V., Cochran, E.J., Shannon, K.M., Hurtig, H., Bird, T.D., Koller, W., Bird, T.D., Trojanowski, J.Q., Lee, V. M.-Y. and Schellenberg, G.D. (2002) An <sup>R5L</sup> Tau mutation in a subject with a Progressive Supranuclear Palsy phenotype. Annals of Neurology. (52)4:511-516.
32. Tsuang, D.W., Dalan, A.M., Eugenio, C.J., **Poorkaj, P.**, Limprasert, P., La Spada, A.R., Steinbart, E.J., Bird, T.D. and Leverenz, J.B. (2002) Familial dementia with Lewy bodies: a clinico-neuropathological study of two families. Archives of Neurology. (59)10:1622-30.
33. Furukawa, K., Wang, Y., Yao, P.J., Fu, W., Mattson, M.P., Itoyama, Y., Onodera, H., D'Souza, I., **Poorkaj, P.**, Bird, T.D. and Schellenberg, G.D. (2003) Alteration in calcium channel properties is responsible for the neurotoxic action of a familial Frontotemporal dementia Tau mutation. Journal of Neurochemistry. (87)2:427-436.
34. **Poorkaj, P.**, Nutt, J.G., James, D., Steinbart, E., Gancher, S., Bird, T.D., Schellenberg, G.D. and Payami, H. (2004) *parkin* mutation analysis in clinic patients with early onset idiopathic Parkinsonism. American Journal of Medical Genetics. (129A)1: 44-50.
35. **Poorkaj, P.**, Moses, L., Montimurro, J.S. Nutt, J.G., Schellenberg, G.D. and Payami, H. (2005) *parkin* mutation dosage and the phenomenon of anticipation: a molecular genetic study of familial parkinsonism. BMC Neurol. Feb 22 (5)1:4.
36. Rademakers, R., Melquist, S., Cruts, M., Theuns, J., Del-Favero, J., **Poorkaj, P.**, Baker, M., Sleegers, K., Crook, R., De Pooter, T., Kacem, S.B., Adamson, J., Van den Bossche, D., Van den Broeck, M., Gass, J., Corsmit, E., De Rijk, P., Thomas, N., Engelborghs, S., Heckman, M., Crook, J., De Deyn, P.P., Dickson, D., Schellenberg, G.D., Van Broeckhoven, C. and Hutton, M.L. (2005) High-density SNP haplotyping suggests altered regulation of tau gene expression in progressive supranuclear palsy. Hum. Mol. Genet. (14)21: 3281-3292.
37. Kay, D.M., Moran, D., Moses, L., **Poorkaj, P.**, Zabetian, C.P., Nutt, J., Factor, S.A., Yu, C.E., Montimurro, J.S., Keefe, R.G., Schellenberg, G.D. and Payami, H. (2007) Heterozygous parkin point mutations are as common in control subjects as in

Parkinson's patients. Ann Neurol. Jan; 61(1): 47-54.

38. McMillan, P., Korvatska, O., **Poorkaj, P.**, Evstafjeva, Z., Robinson, L., Greenup, L., Leverenz, J., Schellenberg, G.D. and D'Souza, I. (2008) Tau isoform regulation is region- and cell-specific in mouse brain. J. Comp. Neurology. Dec; 511(6): 788-803.
39. **Poorkaj, P.**, Raskind, W.H., Leverenz, J.B., Matsushita, m., Zabetian, C.P., Samii, A., Kim, S., Gazi, N., Nutt, J.G., Wolff, J., Yearout, D., Greenup, J.L., Steinbart, E.J. and Bird, T.D. (2010) A novel X-linked 4-repeat tauopathy with Parkinsonism and spasticity. Movement Disorders. *In press.*
40. Kay, D.M., Stevens, C.F., Hamza, T.H., Montimurro, J.S., Zabetian, C. P., Factor, S.A., Mohlo, E., Higgins, D.S., Ganchar, S., Moses, L., Zareparsy, S., **Poorkaj, P.**, Bird, T., Nutt, J., Scjellenberg, G.D. and Payami, H. (2010) A comprehensive analysis of deletions, multiplications, and copy number variations in *PARK2*. Neurology. *Submitted.*

## BOOK CHAPTERS

1. **Poorkaj, P.**, D'Souza, I., Bird, T.D. and Schellenberg, G.D. (2000) Tau mutations: genetics and pathogenetic mechanisms. Foundation IPSEN, Paris, France. In "Fatal Attractions: Protein Aggregates in Neurodegenerative Disorders. V.M.Y. Lee, J.Q. Trojanowski, L. Buee and Y. Christen (eds). Springer Verlag, Berlin. pp 53-64.
2. Yasuda, M., Takamatsu, J., Komure, O., Kuno, S., D'Souza, I., Kawamata, T., Hasegawa, M., Hasegawa, H., Iwatsubo, T., **Poorkaj, P.**, Miyoshi, K., Goedert, M., Schellenberg, G.D. and Tanaka, C. (2001) Tau mutations alter splicing of tau exon 10 in Japanese frontotemporal dementia. *Neuroscientific Basis of Dementia C. Tanaka, P.L. McGeer and Y. Ihara (eds), Birkhauser Verlag, Boston. pp 81-84.*
3. Schellenberg, G.D., D'Souza, I., **Poorkaj, P.** and Bird, T.D. (2001) Tau and Neurodegenerative Disease: Genetics and pathogenetic mechanisms. In: *Neuroscientific Basis of Dementia C. Tanaka, P.L. McGeer and Y. Ihara (eds), Birkhauser Verlag, Boston. pp 71-79.*
4. **Poorkaj, P.**, D'Souza, I. and Schellenberg, G.D. (2004) Genetic susceptibility and animal modeling of PSP. In "Animal Models of Movement Disorders". Academic Press. M.S. LeDoux (ed). Elsevier, USA. 824 pp.

## OTHER PUBLICATIONS

### Reviews:

1. Schellenberg, G.D., D'Souza, I. and **Poorkaj, P.** (2000) The genetics of Alzheimer's disease. Curr. Psychiatry Rep. 2(2): 158-164.

## ABSTRACTS AND PRESENTATIONS

1. **Poorkaj, P.**, Navas, P.A., Brunk, C. and Cascarano, J. (1992) Size related molecular heterogeneity of rat hepatocyte mitochondria and their differential responsiveness to

- thyroxine. Federation of American Societies for Experimental Biology (6)4: A1485 Part 1. Meeting abstract.
2. **Poorkaj, P.**, Scherer, S.W., Massa, H., Nunes, M.E., Geshuri, D., Trask, B., Evans, J.P. and Tsui, L-C. (1994) Physical mapping of the split hand/split foot (SHSF) locus on chromosome 7 reveals a relationship between SHSF and the syndromic ectrodactylies. American Society of Human Genetics Meeting abstract 721:A126.
  3. Sharma, V., Bonnycastle, L., Hisama, F., **Poorkaj, P.**, Weissman, S. and Schellenberg, G. (1994) Construction of a yeast artificial chromosome contig encompassing the chromosome 14 Alzheimer's disease locus. American Society of Human Genetics. Meeting abstract 1581:A270.
  4. Evans, J.P., Scherer, S.W., Crackower, M., **Poorkaj, P.**, Massa, H., Nunes, M.E., Geshuri, D., Hudgins, L., Trask, B. and Tsui, L-C. (1995) Physical mapping of the split hand/split foot (SHSF) locus on chromosome 7 and implication in the syndromic ectrodactylies. Cytogenetics and Cell Genetics 71(1): 29. Meeting abstract.
  5. Schellenberg G.D., Sharma V., **Poorkaj P.**, Hisama F., Wisjman E.M., Weissman S.M. and Bird T.D. (1995) The chromosome-14 Alzheimers-disease locus. Journal of Cellular Biochemistry: 86-86 Suppl. 21B. Meeting abstract.
  6. Schellenberg, G., **Poorkaj, P.**, Wijsman E., Bird, T., Hurtig, H. and Koller, W. (1998) Genetic analysis of Alzheimers disease and related dementias. 6<sup>th</sup> International Conference on Alzheimer's disease and Related Disorders. Meeting abstract 1217.
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14. Schellenberg, G.D., **Poorkaj, P.**, D'Souza, I., Lee, V.M. and Trojanowski, J.Q. (2000) Tau mutations in frontotemporal dementia. Japan. Meeting abstract.
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16. **Poorkaj (Navas), P.**, Muma, N., Jin, Y., Shannon, K., Cochran, E., Trojanowski, J.Q., Lee, V.M.-Y. and Schellenberg, G.D. (2001) Pathological and Biochemical Characterization of a Tau R5L Mutation in a single case of PSP. 31<sup>st</sup> Annual Meeting, Society for Neuroscience, San Diego, CA. Meeting abstract.
17. Furukawa, K., D'Souza, I., **Poorkaj, P.**, Bird, T. and Schellenberg, G. (2002) Tau mutations in FTDP-17 potentiate calcium release from intracellular stores and activate apoptotic pathways. 8<sup>th</sup> International Conference on Alzheimer's disease and Related Disorders. Stockholm, Sweden. Meeting abstract.
18. Bird, T.D., Steinbart, E.M., **Poorkaj, P.** and Smith, C. (2002) The impact of genetic testing and counseling in familial dementia. 8<sup>th</sup> International Conference on Alzheimer's disease and Related Disorders. Stockholm, Sweden. Meeting abstract.
19. Leverenz, J.B., Nochlin, D., **Poorkaj, P.**, Steinbart, E.J., Mastrianni, J.A., Schellenberg, G.D. and Bird, T.D. (2002) Gerstmann-Straussler-Scheinker syndrome in a family with a rare mutation in PRNP (Q160X): Clinical and pathological features. 127<sup>th</sup> Annual Meeting, American Neurological Association, New York, N.Y. Meeting abstract.
20. Frautschy, S.A., Harris-White, M.E., Yang, F., Lim, G.P., Kim, P., Hu, W., Cole, G.M., Navas, P.A., **Poorkaj, P.** and Schellenberg, G.D. (2002) Effects of soluble-amyloid (A) on Tau pathology and neurodegeneration in P301L and wild-type (WT) human tau transgenic mice. Program No. 522.4. 2002 Abstract Viewer/Itinerary Planner. Washington, DC: Society for Neuroscience, 2002. Online.
21. Rubenstein L., Cole G., Schellenberg G., Kraemer B., **Poorkaj P.**, Turner R., Griffin S., Frautschy S., Morley J. and Asthana S. (2002) VA GRECC symposium: from molecular biology and animal models to future treatments for Alzheimer's disease. *Gerontologist* 42: 402-402 Sp. Iss. 1.
22. **Poorkaj, P.**, Nutt J., James, D., Steinbart, E., Gancher, S., Bird, T., Schellenberg, G. and Payami, H. (2003) Parkin mutation analysis in clinic patients with early-onset Parkinson's disease. (2172). *American Journal of Human Genetics* 73(5) 2172. American Society for Human Genetics, Annual Meeting. Los Angeles, CA. Meeting abstract.
23. Hu S., Yang F., Lim G.P., Hu W., Cole G.M., Navas P.A., **Poorkaj P.**, Schellenberg G.D., and Frautschy S.A. (2003) Cognitive and synaptic effects of amyloid in P301L transgenics. Program No. 731.16. 2003 Abstract Viewer/Itinerary Planner. Washington, DC: Society for Neuroscience. New Orleans, LA.

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25. **Poorkaj, P.**, Nutt, J.G., James, D., Gancher, S. Bird, T.D., Steinbart, E., Schellenberg, G. and Payami, H. (2004) parkin mutation analysis in clinic patients with early-onset Parkinson's disease. *American Journal of Medical Genetics Part A* 129A(1): 44-50. Meeting abstract.
26. Payami, H., **Poorkaj, P.**, Nutt, J., Schellenberg, G., James, D. and Bird, T. (2004) Parkin mutation analysis in clinic patients with early-onset Parkinson's disease. *Genetics in Medicine* 6(4): 262. Meeting abstract.
27. Payami, H., Kay, D., Moran, D., Moses, L., **Poorkaj, P.**, Zabetian, C., Factor, S., Yu, C-E., Montimurro, J., Keefe, R., Schellenberg, G. and Nutt, J. (2007) Role of *parkin* in PD. American Neurological Association 132nd Annual Meeting. Abstract number: 950345. Meeting abstract.
28. **Poorkaj-Navas, P.**, Raskind, W.H., Leverenz, J.B., Matsushita, M., Zabetian, C.P., Samii, A., Kim, S., Gazi, N., Tisch, L.M., Nutt, J. G., Wolff, J., Yearout, D., Greenup, J.L., Steinbart, E.J., Bird, T.D. (2009) A Novel X-linked 4-Repeat Tauopathy with Parkinsonism and Spasticity. 59<sup>th</sup> Annual Meeting of The American Society for Human Genetics, Date: October 20-24, 2009 Honolulu, HI, USA. Abstract number: 523. Meeting abstract.

## INVITED PRESENTATIONS

- 2000 Invited speaker - UCLA Gene Medicine Seminar Series, Department of Surgery, August 23, 2000; Los Angeles, California. Tauopathies: towards an animal model.
- 2002 Invited speaker - International PSP Research Symposium, November 9, 2002; Miami Beach, Florida. Toward identification of a PSP gene.
- 2002 Invited Speaker- University of Washington, Division of Gerontology and Geriatric Medicine Seminar; December 2, 2002; Seattle, Washington. Parkin mutations in Parkinson's disease and Parkinsonian-like diseases.
- 2003 Invited State of the Art Lecture - Western Regional Meeting, February 1, 2003; Carmel, California. The molecular genetics of Tauopathies: a timeline of tau.
- 2004 Invited speaker - University of Washington, Department of Psychiatry and Behavioral Sciences Seminar, September 14, 2004; Seattle, Washington. Genetic and phenotypic diversity in Parkinsonian diseases.
- 2004 Invited speaker - The Washington State Regional Family Conference on Progressive Supranuclear Palsy, September 26, 2004; Tukwila, Washington. The Biology of PSP: what we have learned and the paths to discovery.
- 2004 Invited speaker- University of Washington, Division of Medical Genetics Markey Molecular Medicine Seminar, November 5, 2004; Seattle, Washington. The tangled tale of tau.

- 2005 Invited speaker - Society for Progressive Supranuclear Palsy; Fourth Research Symposium, Thursday, November 17, 2005; Washington DC. Mutant parkin transgenic mice and PSP.
- 2007 Invited speaker - Society for Progressive Supranuclear Palsy; 2007 International Research Symposium, Thursday, November 8, 2007; San Diego, CA. Parkin and Tau mutational effects on tangle formation in PSP.
- 2008 Invited Speaker - University of Washington, Department of Speech and Hearing Sciences Colloquium Lecture; Thursday, March 6, 2008. Neurological disease and the senescing brain: A shorter path to the same destination?
- 2009 Workshop Presenter - 64th Annual Scientific Convention & Program, Society of Biological Psychiatry, May 14-16, 2009; Vancouver, Canada. Novel molecular genetic methods to identify causative genes in psychiatric disorders.