

4. Annemarie Opprecht Parkinson Award 2008

Curriculum Vitae Andrew B. Singleton

Personal Information:

Marital status: Married with 2 children

Citizenship: British

Address: 10121 Tenbrook Drive, Silver Spring, MD 20901, USA

Profession: Ph. D., Professor of Neuroscience, Chief of the Molecular Genetics Section, Acting Chief of the Laboratory of Neurogenetics, National Institute on Aging, National Institutes of Health, Bethesda, Maryland, USA

Education:

1991 - 1995 University of Sunderland, UK. BSc, 1st Class (Hons), Applied Physiology

1995 - 1998 University of Newcastle upon Tyne, UK. PhD, Neuroscience

Brief Chronology of Employment:

1996 - 1997 Visiting Lecturer, Physiology, University of Sunderland, UK

1998 - 1999 Research Scientist, Medical Research Council Neurochemical Pathology Unit, Newcastle upon Tyne, UK

1999 - 2000 Postdoctoral Fellow, Mayo Clinic Jacksonville, Florida, USA

2000 - 2001 Instructor in Molecular Biology/Biochemistry, Mayo Clinic Jacksonville, Florida, USA

2001 - 2002 Research Scientist, Johns Hopkins University, Baltimore, Maryland, USA

2002 - 2007 Tenure Track Investigator and Chief, Molecular Genetics Unit, Laboratory of Neurogenetics, National Institute on Aging, National Institutes of Health, Bethesda, Maryland, USA

2007 - present Senior Investigator with Tenure, Chief of the Molecular Genetics Section and Acting Chief of the Laboratory of Neurogenetics, National Institute on Aging, National Institutes of Health, Bethesda, Maryland, USA

2007 - present Visiting Professor, Center for Public Health Genomics, University of Virginia, Virginia, USA

Honors and Other Special Scientific Recognition:

2003 Research describing α -synuclein triplication mutations picked as one of NIH top 5 research advances for 2002-2003

2004 Article entitled "Alpha-synuclein locus triplication causes Parkinson's disease" listed by ThomsonISI as one of the most cited papers (top 0.1%) in

- Neuroscience & Behavior and listed as a Hot New Paper on the ISI website (cited >250 times)
- 2005 Boehringer Mannheim Research Award, 16th International congress on Parkinson's disease and related disorders, Berlin, Germany
- 2005 Research describing *LRRK2* mutations picked as one of NIH top 5 research advances for 2004-2005
- 2005 Article entitled "Alpha-synuclein locus triplication causes Parkinson's disease" listed by ThomsonISI as the second most cited paper in Neuroscience & Behavior for 2005
- 2006 Profiled by *Lancet Neurology* in the first of the monthly Lifeline columns
- 2007 Genome-wide SNP association in Parkinson's disease picked as one of NIA top 20 advances for 2006

External and Internal Funding:

- 1999 - 2000 Co-Principal Investigator "Assessing the role of alpha-synuclein in dementia with Lewy bodies, Parkinson's disease and multi-system atrophy" funded by the Alzheimer's disease Research Center \$30,000
- 2000 - 2001 Principal Investigator "Identification and cloning of the gene defect responsible for X-linked dystonia Parkinsonism (Lubag)" funded by the Dystonia Medical Research Foundation. \$35,000
- 2004 - 2007 Genetics mentor, Elizabeth Peckham; Clinical Research Fellow from the Benign Essential Blepharospasm Research Foundation to study "Characterization and collection of families with blepharospasm; Facilitating the identification of genetic defect(s) causing blepharospasm." Funded by NINDS intramural program. \$195,000
- 2005 - 2006 Co-Principal Investigator, Office of the Scientific Director (NIA) set-aside funding, "Atherosclerotic and inflammatory genetic profiles and the risk for brain disease". \$86,000
- 2006 - 2007 Principal Investigator, USAMRMC proposal #06173004, "Molecular Genetic Characterization of Parkinson's Disease; Delineation of the Pathogenic Function of *LRRK2* Mutations". \$435,200
- 2006 - 2007 Principal Investigator, Office of the Scientific Director (NIA) set-aside funding, "Rapid Genetic Mapping in Movement Disorders using High Density SNP Arrays". \$108,984
- 2006 - 2007 Principal Investigator, Office of the Scientific Director (NIA) set-aside funding, "Statistical Genetic Analysis of Genome-Scale Data". \$109,000
- 2006 - 2007 Co-Principal Investigator, Office of the Scientific Director (NIA) set-aside funding, "The Aging Genome Association Study – 'Age-Gain'". \$2,300,000
- 2006 - 2007 Co-Principal Investigator, Office of the Scientific Director (NIA) set-aside funding, "Whole Genome Association Studies in a Cohort of Patients with Fibromuscular Dysplasia with Connective Tissue Abnormalities". \$35,000
- 2006 - 2007 Co-Principal Investigator, Office of the Scientific Director (NIA) set-aside funding, "The HANDLS Genotyping Project". \$515,250

Plenary and Invited Lectures:

- 2001 Dystonia medical research foundation research workshop, Bethesda, Maryland, USA. Genetic update on X-linked dystonia parkinsonism
- 2001 Dystonia Medical Research Foundation/NINDS Dystonia Workshop, Phoenix, Arizona, USA. From Gene to Function in Dystonia
- 2001 St. Luke's Medical Center Grand Rounds, Quezon City, Philippines. X-linked recessive dystonia parkinsonism
- 2001 Institute of Neurology Grand Rounds, London UK. Genetics of dystonia
- 2002 Fourth International Dystonia Symposium, Atlanta, Georgia, USA. Update on the Genetics of X-linked dystonia parkinsonism
- 2003 Arizona State University Neuroscience series, Phoenix, Arizona, USA. Genetic advances in understanding Parkinson's disease
- 2003 Barros Neuroscience Research Facility, Tempe, Arizona, USA. Genetic advances in Parkinson's disease; α -synuclein multiplication
- 2003 Mongolian School of Health Sciences Grand Rounds, Ulaanbaatar, Mongolia. Genetics of Neurological disease, past, present and future
- 2004 Johns Hopkins-NIH joint neuroscience lecture series on neurogenetics, Baltimore, Maryland, USA. The Iowa kindred, new insights into Parkinson's disease
- 2004 New York Academy of Sciences, New York, USA. Neurodegenerative Diseases Discussion Group. Neurodegenerative disease and dose
- 2004 ASENT, Genetic Testing in Parkinson's disease consensus meeting, Bethesda, Maryland, USA. Genetics of Parkinson's disease
- 2004 Seventh International Congress of Parkinson's disease and Movement Disorders, Rome, Italy. Genetics of Neurological disease
- 2004 Parkinson's Action Network Research and Education Forum Scientific Overview Panel, Bethesda, Maryland, USA. PD genetics; alpha synuclein
- 2004 Secretary for Health and Human Services visit to NIH, Bethesda, USA. Genetic Advances in Parkinson's disease
- 2004 Clinico-Pathological Correlations In the Lewy Body Dementias Workshop, Washington DC, USA. Genetics of Lewy body disease
- 2004 United States Army Neurotoxin Exposure Treatment Research Program, University of Rochester Medical Center, Rochester, New York, USA. Genetics of Parkinson's disease
- 2004 National Society of Genetic Counselors 23rd Annual Education Conference, Washington DC, USA. Genetics of Parkinson's Disease
- 2005 International Stroke Conference, SWISS satellite meeting, New Orleans, Louisiana, USA. Genetics of Stroke
- 2005 Parkinson's Action Network Research and Education Forum Scientific Overview Panel, Bethesda, Maryland, USA. Cloning of the gene underlying PARK8 Parkinson's disease
- 2005 Eighth International Congress of Parkinson's Disease and Movement Disorders, New Orleans, Louisiana, USA. α -synuclein and the Lewy body
- 2005 NIH Clinical Center Grand Rounds, Bethesda, Maryland, USA. Synuclein dose mutations and dardarin

- 2005 University College London Grand Rounds, London, UK. Synuclein, dose and dardarin
- 2005 University of Newcastle upon Tyne, Newcastle upon Tyne, UK. The role of *LRRK2* mutations in Parkinson's disease
- 2005 Spasmodic Dysphonia planning session, Bethesda, Maryland USA. Genetic approaches to neurological disease
- 2005 Henry Stewart talks series on current thinking in trends in research on neurodegenerative diseases, recorded in Bethesda, Maryland, USA. Molecular Genetics of Parkinson's disease
- 2005 Institute of Molecular Medicine for the Prevention of Human Diseases, University of Texas at Houston, Houston, Texas, USA. Synuclein triplication and *LRRK2* disease; recent insights
- 2005 Essential tremor research group consensus meeting, Washington DC, USA. Genetics of neurological disorders
- 2005 University of Florida, Gainesville, FL, USA. Past, present and future approaches to the genetics of Parkinson's disease
- 2005 Siblings with Ischemic Stroke investigators meeting, Orlando, FL, USA. Genetic approaches to Stroke.
- 2005 Institute of Medicine, National Academy of Sciences, Washington DC, USA. Genetics in complex disease
- 2006 Centers-Network for Research in Neurological disorders, Nicosia, Cyprus. Genome wide SNP approaches in Neurological disorders
- 2006 Parkinson's disease World Congress, Washington DC, USA. Genes, Genetics and Parkinson's disease
- 2006 DLB/PDD at a crossroads, Washington DC, USA. Genetics advances in DLB and PDD. Chair, Basic Science Session.
- 2006 Teva Neuroscience practicum in Neurology, Chicago, IL, USA. Genetic aspects of Parkinson's disease
- 2006 University of Pennsylvania School of Medicine, Philadelphia, PA, USA. Genetics of Parkinson's disease
- 2006 Operational Medicine Research Program (MOMRP) research review meeting for the Neurotoxin Exposure Treatment Research Program (NETRP). Herndon, Virginia, USA. Genetic progress and future in Parkinson's disease
- 2006 8th Annual Meeting, Udall Centers of Excellence for Parkinson's Disease Research, Boston, MA, USA. Update on the genetics of Parkinson's disease
- 2006 10th International Movement Disorders Society Meeting. Kyoto, Japan. Molecular mechanisms of nigral cell death in *PARK1* and *PARK4*
- 2006 4th International Workshop on DLB and PDD. Yokohama, Japan. Molecular genetic insights into DLB and PDD; past and future. Chair, Basic Science Symposium
- 2006 Veterans Affairs Medical Center Grand Rounds, Washington DC, USA. Genetic-based insights into synucleinopathies
- 2007 Michael J Fox Foundation scientific advisory board meeting, Houston, TX, USA. The future of Parkinson's disease genetics
- 2007 Gaucher Disease and Parkinsonism Workshop, Bethesda, MD, USA. The genetics of synucleinopathies

- 2007 Uniformed Services University of the Health Sciences, MD, USA. The genetics of Parkinson's disease
- 2007 National Institute of Neurological Disorders and Stroke Neurogenetics Repository advisory board meeting, Camden, NJ, USA. Genome wide SNP data in Parkinson's disease and controls
- 2007 Society for epidemiologic research, Boston, MA, USA. Professional rewards of open data sharing
- 2007 Uniformed Services University of the Health Sciences, Bethesda, MD, USA. The genetics of neurological disorders
- 2007 NIH Research Festival, Bethesda, MD, USA. Genome wide SNP genotyping: association and beyond
- 2007 NIH Research Festival, Bethesda, MD, USA. Genetics of Parkinson's disease
- 2007 InCHIANTI study group, Florence, Italy. Genome Wide Association Studies; practical considerations and application
- 2007 National Institute of Environmental Health Sciences, NC, USA. Genome wide SNP genotyping: simple association and beyond

Ad hoc reviewer for:

American Journal of Human Genetics
American Journal of Medical Genetics; Neuropsychiatric Genetics
Annals of Neurology (editorial board member)
Biomarkers in Medicine
BMC Genetics
BMC Neurology
Brain
Clinical Autonomic Research
Clinical Chemistry
Clinical Genetics
European Journal of Neurology
European Journal of Neuroscience
Experimental Cell Research
Expert Review of Neurotherapeutics
Expert Review of Molecular Diagnostics
Future Neurology
Genetic Epidemiology
Genetics in Medicine
Human Genetics
Human Molecular Genetics
Human Mutation
Journal of Medical Genetics
Journal of Neurology
The Lancet
The Lancet Neurology
Movement Disorders
Nature Genetics
Neurobiology of Aging
Neurobiology of Disease
Neurodegenerative Diseases (editorial board member)

Neurogenetics (editorial board member)
Neurology
NeuroMolecular Medicine
NeuroReport
Neuroscience Letters
Pharmacogenetics
Pharmacogenetics and Genomics
PLoS ONE
Regenerative Medicine
Trends in Molecular Medicine

Service on Advisory Boards, Panels and Editorial Boards:

2005 *Ad hoc* member, Scientific Advisory Board, National Parkinson Foundation, Washington DC, USA
2006 - 2007 Scientific Advisory Board, Alzheimer Research Forum (www.alzforum.org)
2006 - present Scientific Advisory Board, Michael J Fox Foundation for Parkinson's disease
2006 - present Editorial Board member for the journal *Neurodegenerative Diseases*
2006 Co-Chair, Genetics Section, Stroke Progress Review Group
2006 - present Steering Committee, US Army Medical Research and Materiel Command Neurotoxin Exposure Treatment Research Program investigation of Parkinson's disease
2007 Data sharing committee member, NIH Multi-IC Symposium on Genome-Wide Association, Rockville, MD, USA
2007 - present Editorial Board member for the journal *Annals of Neurology*
2007 - present Advisory Board member for the National Institute of Neurological Disorders and Stroke Neurogenetics Repository, Camden, NJ, USA
2007 - present Editorial Board member for the journal *Neurogenetics*
2007 - present Advisory Board, Howard Hughes Medical Institution-NIH Research Scholars Program

Grant Review Committees

2001 NINDS Gene Discovery RFA Grant Review Committee, Washington DC, USA
2003 Michael J Fox Foundation Community Fast Track Grant Review Committee, New York, New York, USA
2004 Michael J Fox Foundation Gene Discovery Grant Review Committee, Washington DC, USA
2004 Parkinson's Disease Society UK, Project Grant Review, Bethesda, MD USA
2004 NINDS Morris K Udall Parkinson's Disease Center of Excellence Review, Washington DC, USA
2004 Singapore Biomedical Research Council Grant Review, The Agency for Science, Technology and Research's (A*STAR), Bethesda, Maryland, USA
2004 Michael J Fox Foundation Community Fast Track Grant Review Committee, New York, New York, USA
2004 Parkinson's Disease Society UK Project Grant Review, Bethesda, Maryland, USA
2005 Michael J Fox Foundation **Clinical Discovery Grants for Parkinson's disease Research**, New York, New York, USA
2005 U.S. Army Medical Research and Materiel Command Grant Review, Bethesda, Maryland, USA

2005 National Health and Medical Research Council (Australia) Project Grant Application Review, Bethesda, Maryland, USA

2005 National Parkinson's Foundation Large and Small Grant Review Committee, Washington DC, USA

2005 Italian Telethon Foundation Grant Review, Bethesda, Maryland, USA

2005 NINDS Fellowship Study Section, Bethesda, Maryland, USA

2005 BUPA foundation for medical research UK Grant Review, Bethesda, Maryland, USA

2005 National Institute on Aging Concept Review, Bethesda, Maryland, USA

2005 Michael J Fox Foundation Community Fast Track Grant Review Committee, New York, New York, USA

2006 Westat, independent Review Committee for proposals to assay SMN expression, Bethesda, Maryland, USA

2006 NINDS Fellowship Study Section, Bethesda, Maryland, USA

2006 Michael J Fox Foundation Animal Models Review, New York, New York, USA

2006 Michael J Fox Foundation Global Genetics Consortia Progress Review, New York, New York, USA

2006 Progressive Supranuclear Palsy Association UK grant review, Bethesda Maryland, USA

2007 NINDS reverse site visit, Chicago, Illinois, USA

2007 Michael J Fox Foundation Rapid Response Innovation Awards, Bethesda, Maryland, USA

2007 Technical Advisory Group member; Center for Inherited Disease Research, Bethesda, Maryland, USA

2007 Mariani Foundation, grant review, Bethesda, MD, USA

2007 Ministry of Health, Singapore, grant review, Bethesda, MD, USA

2007 Agency for Science, Technology and Research, Biomedical Research Council, Singapore, grant review, Bethesda, MD, USA

2007 Danish Medical Research Council, grant review, Bethesda, MD, USA

2007 Health Research Board, Ireland, grant review, Bethesda, MD, USA

NIH Related Activities:

2003 NIH FARE award judge

2004 Host, Neuroscience Seminar Series, Dr Peter Heutink, Vrije University, the Netherlands

2004 NIH FARE award judge

2005 Host, NINDS Grand Rounds speaker, Dr Enza-Maria Valente, CSS Mendel Institute, Italy

2005 NIA representative and Scientific Chair, Porter Neuroscience Research Center Users Committee

2005 NIH FARE award judge

2005 Organizer and host, Porter Neuroscience Research Center Principal Investigator seminar series

2006 Host, Neuroscience Seminar Series, Dr Clive Svendsen, the Waisman Institute, USA

2006 Tenure track investigator search committee member, Laboratory of Neurosciences, Intramural, Program, NIA

2006 Staff Clinician search committee member, Intramural Program, NIA

2007 Promotion and tenure review committee member, Intramural Program, NIA

2007 Chair, Genome wide SNP genotyping, NIH research festival

Previous Staff and Visiting Scientists Mentored:

Crystal Evey, BS – Predoctoral IRTA

2000 Bachelor of Sciences, University of California at Santa Cruz
2001-2002 Molecular Genetics Unit
CURRENT LOCATION: Medical student, University of California at San Diego
Medical School, CA, USA

Crystal Wilson, BS – Summer IRTA

2001 Premed. University of North Carolina at Chapel Hill
2002 Molecular Genetics Unit
2003 Molecular Genetics Unit
CURRENT LOCATION: Med. student, Univ. of North Carolina Medical School,
NC, USA

Shannon Chang, BS – Summer IRTA

2003 Premed. Johns Hopkins University
2002 Molecular Genetics Unit
2003 Molecular Genetics Unit
CURRENT LOCATION: Medical student, Johns Hopkins University Medical
School, MD, USA

Jason Freeman, BS – Summer IRTA

2002 Premed. Georgetown University
2002 Molecular Genetics Unit
CURRENT LOCATION: Medical student, Georgetown University Medical School,
Washington DC, USA

Stephen Hague, PhD – Research Fellow

2002 PhD, University College London
2002-2003 Molecular Genetics Unit
CURRENT LOCATION: Research fellow, University of Sheffield, UK

Okan Dogu, MD – Visiting Scientist

1990 MD, University of Istanbul, Turkey
2003 Molecular Genetics Unit
CURRENT LOCATION: Assistant Professor, Department of Neurology, Mersin
University, Turkey

Gabriella Pavlic, MD – Visiting Scientist

2002 MD, State University of Medicine and Pharmacy, Chisinau, Moldova
2004 Molecular Genetics Unit
CURRENT LOCATION: Clinical Fellow, Department of Neurology, State
University of Medicine and Pharmacy, Chisinau, Moldova

Gauri Gaikwad, MD – Visiting Scientist

2000 MD, Rural Medical College, Pune, India
2003 Molecular Genetics Unit
CURRENT LOCATION: Resident in Pediatrics, Louisiana State Univ., LA, USA

Georgia Xiromerisiou, MD – Visiting Scientist

2000 MD, University of Thessaly School of Medicine, Greece
2004 Molecular Genetics Unit
2005 Molecular Genetics Unit
CURRENT LOCATION: Clinical Fellow, Department of Neurology, Neurogenetics
Branch, University of Thessaly, Greece

Sarah Camargos, MD – Visiting Scientist

2003 MD, UFMG, Brazil.

2005 Molecular Genetics Unit

CURRENT LOCATION: Clinical Fellow, Faculdade de Medicina – UFMG, Brazil

Erin W Evans, BA – Post baccalaureate IRTA

2003 BA, Duke University, North Carolina, USA

2003-2005 Molecular Genetics Unit

CURRENT LOCATION: Masters student in Nutritional Sciences, Harvard, Boston, MA. USA

Jordi Clarimon, PhD – Visiting fellow

2003 PhD University of Barcelona

2003-2005 Molecular Genetics Unit

CURRENT LOCATION: Research Fellow, Unidad de Memoria. Hospital de la Santa Creu i Sant Pau, Barcelona 0802, Spain

Shushant Jain – Graduate Student

2002 BSc University of Manchester

2004-2006 Molecular Genetics Unit

CURRENT LOCATION: Postdoctoral Fellow, Department of Medical Genomics, Vrije University, Amsterdam, the Netherlands

Coro Paisan-Ruiz – Visiting Fellow

2003 PhD Student University of San Sebastian, Spain

2004-2007 Molecular Genetics Unit

CURRENT LOCATION: Senior Lecturer, University College London, United Kingdom

Peer Reviewed Articles:

1. Christie J, Lamb H, **Singleton A**, Leake A, Melton L, Perry R, McKeith I, Morris CM (1996) Determination of the alpha-1-antichymotrypsin polymorphism in Alzheimer's disease. *Alzheimer's Research*. 2:201-204
2. **Singleton AB**, Gibson AM, Atkinson AL, Daly A, Morris CM (1997) Presenilin polymorphisms in Alzheimer's disease [letter; comment]. *Lancet* 350:958-959
3. **Singleton AB**, Lamb H, Leake A, McKeith IG, Perry RH, Morris CM. (1997) No association between an intronic polymorphism in the presenilin-1 gene and Alzheimer's disease. *Neurosci Letters* 234:19-22
4. Thompson J, Thomas N, **Singleton A**, Piggott M, Lloyd S, Perry EK, Morris CM, Perry RH, Ferrier IN, Court JA (1997) D2 dopamine receptor gene (DRD2) Taq1 A polymorphism: reduced dopamine D2 receptor binding in the human striatum associated with the A1 allele. *Pharmacogenetics* 7:479-484
5. **Singleton AB**, Lamb H, Leake A, McKeith IG, Ince PG, Perry RH, Morris CM (1997) No association between a polymorphism in the presenilin 1 gene and dementia with Lewy bodies. *Neuroreport* 8:3637-3639
6. Woodward R, **Singleton AB**, Gibson AM, Edwardson JA, Morris CM (1998): LRP gene and late-onset Alzheimer's disease. *Lancet* 352:239-240
7. **Singleton AB**, Thomson JH, Morris CM, Court JA, Lloyd S, Cholerton S (1998) Lack of association between the dopamine D2 receptor gene allele DRD2*A1 and cigarette smoking in a United Kingdom population. *Pharmacogenetics* 8:125-128
8. **Singleton, AB**, Gibson AM, Edwardson JA, McKeith IG, Morris CM (1998) Butyrylcholinesterase K: an association with dementia with Lewy bodies. *Lancet* 351:1818

9. Lamb H, Christie J, **Singleton AB**, Leake A, Perry RH, Ince PG, McKeith IG, Melton LM, Edwardson JA Morris CM (1998) Apolipoprotein E and alpha-1 antichymotrypsin polymorphism genotyping in Alzheimer's disease and in dementia with Lewy bodies. Distinctions between diseases. *Neurology* 50:388-391
10. **Singleton AB**, Smith G, Gibson AM, Woodward R, Perry RH, Ince PG, Edwardson JA Morris CM (1998) No association between the K variant of the butyrylcholinesterase gene and pathologically confirmed Alzheimer's disease. *Human Molecular Genetics* 7:937-939
11. Atkinson A, **Singleton AB**, Steward A, Ince PG, Perry RH, McKeith IG, Fairbairn AF, Edwardson JA, Daly AK, Morris CM (1999) CYP2D6 is associated with Parkinson's disease but not with dementia with Lewy Bodies or Alzheimer's disease. *Pharmacogenetics* 9:31-35
12. **Singleton AB**, Gibson AM, McKeith IG, Ballard CA, Perry RH, Ince PG, Edwardson JA, Morris CM (1999) Alpha2-macroglobulin polymorphisms in Alzheimer's disease and dementia with Lewy bodies. *Neuroreport* 10:1507-1510
13. De Jonghe C, Cruts M, Rogaeva EA, Tysoe C, **Singleton A**, Vanderstichele H, Meschino W, Dermaut B, Vanderhoeven I, Backhovens H, Vanmechelen E, Morris CM, Hardy J, Rubinsztein, D.C., St George-Hyslop PH, Van Broeckhoven C (1999) Aberrant splicing in the presenilin-1 intron 4 mutation causes presenile Alzheimer's disease by increased Abeta42 secretion. *Human Molecular Genetics* 8:1529-1540
14. Gibson AM, **Singleton AB**, McKeith IG, Ballard CA, Perry RH, Ince PG, Edwardson JA, Morris CM (2000) Lack of association of the α -2 macroglobulin locus on chromosome 12 in AD. *Neurology*. 25;54(2):433-438
15. Gwinn-Hardy K, Chen J-Y, Liu HC, Liu TY, Boss M, Adam A, **Singleton A**, Koroshetz W, Waters C, Hardy J and Farrer M (2000) Spinocerebellar ataxia type 2 with Parkinsonism in ethnic Chinese. *Neurology*. 55:800-805
16. **Singleton A**, Hall R, Ballard CG, Perry RH, Xuereb JH, Rubinsztein D, Tysoe C, Matthews P, Cordell B, Samir KS, De Jonghe C, Cruts M, van Broeckhoven C and Morris CM (2000) Pathology of early-onset Alzheimer's disease cases bearing the Thr113-114ins presenilin-1 mutation. *Brain*. 123:2467-2474
17. Gwinn-Hardy K, **Singleton A**, O'Suilleabhain P, Boss M, Nicholl D, Adam A, Hussey J, Thomas GEM, Critchley P, Hardy J and Farrer M (2001) Spinocerebellar ataxia type 3 in blacks presenting as Parkinson's disease. *Archives of Neurology*. Feb;58(2):296-299
18. Farrer M, Destee A, Levescque C, **Singleton A**, Engelender S, Becquet E, Mouroux V, Richard F, Defebvre L, Crook R, Hernandez D, Ross C, Hardy J, Amouyel P and Chartier-Harlin M-C (2001) Genetic analysis of synphilin-1 in familial Parkinson's disease. *Neurobiology of Disease*. 8:317-323
19. **Singleton A**, Gibson A, McKeith I, Ballard C, Edwardson JA and Morris CM (2001) NOS Gene Polymorphisms in Alzheimer's disease and Dementia with Lewy Bodies. *Neuroscience Letters*. 303(1):33-36
20. Farrer M, Maraganore DM, Lockhart P, **Singleton A**, Lesnick TG, de Andrade M, West A, de Silva R, Hardy J and Hernandez D (2001) α -Synuclein Gene Haplotypes are Associated with Parkinson's Disease. *Human Molecular Genetics*. 10(17):1847-1851
21. Maraganore DM, Hernandez D, **Singleton A**, Farrer M, McDonnell S, Hutton M, Hardy J and Rocca W (2001) Case-control study of the extended tau gene haplotype in Parkinson's disease. *Annals of Neurology*. 50: 658-661
22. Farrer M, Chan P, Lincoln S, Chen R, Hussey J, Gwin-Hardy K, Petrucelli L, Hernandez D, **Singleton A**, Forno L, Tanner C, Tan L, Hardy J, Langston JW (2001) Lewy bodies and parkinsonism in families with parkin mutations. *Ann Neurol*. 2001 50:293-300
23. West A, Zimprich A, Lockhart PJ, Farrer M, Holtom B, Lincoln S, **Singleton A**, Hofer A, Hill L, Muller-Myhsok B, Wszolek ZK, Hardy J, Gasser T (2001) Refinement of the Park3 locus on chromosome 2p13 and the analysis of 14 candidate genes. *European Journal of Human Genetics*. 9:659-666
24. O'Farrel C, Murphy D, Petrucelli L, **Singleton A**, Hussey J, Farrer M, Hardy J, Dickson D and Cookson M (2001) Transfected synphilin-1 forms cytoplasmic inclusions in HEK293 cells. *Brain Res Mol Brain Res*.97(1):94-102

25. Amtul, Z., P. A. Lewis, S. Piper, R. Crook, M. Baker, K. Findlay, **A. Singleton**, M. Hogg, L. Younkin, S. G. Younkin, J. Hardy, M. Hutton, B. F. Boeve, D. Tang-Wai and T. E. Golde (2002) A Presenilin 1 Mutation Associated with Familial Frontotemporal Dementia Inhibits gamma-Secretase Cleavage of APP and Notch. *Neurobiol Dis* 9(2): 269-73
26. Evidente VGH, Gwinn-Hardy K, Hardy J, Hernandez D and **Singleton A** (2002) X-linked dystonia ('Lubag') presenting predominantly with parkinsonism: A more benign phenotype? *Movement Disorders*. 17(1) 200-202
27. O'Farrell C, Hernandez DG, Evey C, **Singleton AB** and Cookson MR (2002) Normal localization of Δ F323-Y328 mutant TorsinA in transfected cells. *Neuroscience Letters*. 327(2):75-78
28. Subramony SH, Hernandez D, Adam A, Smith-Jefferson S, Hussey J, Gwinn-Hardy K, Hardy K, Farrer M and **Singleton A** (2002) Ethnic Differences in the Expression of Neurodegenerative Disease: Machado-Joseph Disease in Africans and Caucasians. *Movement Disorders* 17(5):1068-1071
29. **Singleton AB**, Wharton A, O'Brien KK, Walker MP, McKeith IG, Ballard CG, O'Brien J, Perry RH, Ince PG, Edwardson JA and Morris CM (2002) Clinical and Neuropathological Correlates of Apolipoprotein E Genotype in Dementia with Lewy Bodies. *Dementia and Geriatric Cognitive Disorders* 14 167-175
30. Goudreau J, Maraganore D, Farrer M, Lesnick T, **Singleton A**, Bower J, Hardy J and Rocca W (2002) Case control study of Dopamine transporter-1, Monoamine Oxidase-B, and catechol-O-methyl transferase polymorphisms in Parkinson's disease. *Movement Disorders*. 17 1305-1311
31. Evidente VGH, Advincula J, Esteban R, Pasco P, Alfon JA, Natividad F, Cuanang J, Gwinn-Hardy K, Hardy J, Hernandez D and **Singleton A** (2002) The phenomenology of 'Lubag' or X-linked dystonia-parkinsonism. *Movement Disorders*. 17 1271-1277
32. Payami H, Nutt J, Gancher S, Bird T, Gonzales McNeal M, Seltzer WK, Hussey J, Lockhart P, Gwinn-Hardy K, Singleton A, **Singleton AB**, Hardy J, Farrer M (2003) SCA2 may present as levodopa-responsive parkinsonism *Movement Disorders*. 18 425-429
33. Kaufmann H, Saadia D, Polin C, Hague S, Singleton A and **Singleton A** (2003) Primary hyperhidrosis: Evidence for autosomal dominant inheritance. *Clinical Autonomic Research*. Apr;13(2):96-98
34. Hardy J, **Singleton A** and Gwinn Hardy K (2003) Ethnic differences in Neurodegenerative disease phenotypes. *Science*. 300;739-740
35. Hernandez D, Hanson M, Singleton A, Gwinn-Hardy K, Freeman J, Ravina B, Doheny D, Gallardo M, Weiser R, Hardy J and **Singleton A** (2003) Mutation at the SCA17 locus is not a common cause of parkinsonism. *Parkinsonism and Related Disorders*. 9: 317-320
36. Hague S, Rogaeva E, Hernandez D, Gulick C, Singleton A, Hanson M, Johnson J, Weiser R, Gallardo M, Ravina B, Gwinn-Hardy K, Crawley A, St George-Hyslop PH, Lang AE, Heutink P, Bonifati V, Hardy J and **Singleton A** (2003) Identification of a compound heterozygous mutation in *DJ-1* causing early-onset Parkinson's disease. *Annals of Neurology*. 54:271-274
37. Eerola J, Hernandez D, Launes J, Hellström O, Hague S, Gulick C, Johnson J, Peuralinna T, Hardy J, Tienari PJ and **Singleton AB** (2003) Assessment of a *DJ-1* (*PARK7*) polymorphism in Finnish PD. *Neurology*. 61:1000-1002
38. **Singleton AB**, Farrer M, Johnson J, Singleton A, Hague S, Kachergus J, Hulihan M, Peuralinna T, Dutra A, Nussbaum R, Lincoln S, Crawley A, Hanson M, Maraganore D, Adler C, Cookson MR, Muenter M, Baptista M, Miller D, Blancato J, Hardy J and Gwinn-Hardy K (2003) α -Synuclein Locus Triplication Causes Parkinson Disease. *Science*. 302:841
39. Morris CM, O'Brien K, Gibson AM, Hardy JA, **Singleton AB** (2003) Polymorphism in the Human DJ-1 Gene is not Associated with Sporadic Dementia with Lewy Bodies or Parkinson's disease. *Neuroscience Letters*. 352:151-153
40. Evans W, Hague S, **Singleton A** (2004) DJ-1 and Parkinson's disease. *Annals of Neurology*. 55:145-146

41. Hague S, Peuralinna T, Eerola J, Hellström O, Tienari PJ, **Singleton A** (2004) Confirmation of the protective effect of iNOS in an independent cohort of Parkinson's disease. *Neurology*. 62:635-636
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