

Curriculum Vitae and Bibliography Rosa Rademakers, PhD

PERSONAL INFORMATION

Work Address: 4500 San Pablo Road South
Jacksonville, FL 32224

PRESENT ACADEMIC RANK AND POSITION

Senior Associate Consultant - Neuroscience, Mayo Clinic, Jacksonville, Florida 09/02/2009 - Present
Full Faculty Privileges in Neuroscience - Mayo Graduate School, College of Medicine, Mayo Clinic 02/03/2010 - Present
Associate Professor of Neuroscience - College of Medicine, Mayo Clinic 03/01/2010 - Present

EDUCATION

University of Antwerp 1997
BA
University of Antwerp 1999
MA, Biochemistry
University of Antwerp 2004
Ph.D.
University of Antwerp 2004 - 2007
Post Doctoral Fellowship
Additional Education
Genetic Linkage 05/2000
Netherlands Institute for Health Science (NIHES), Erasmus Medical Center.
Rotterdam, Netherlands

HONORS/AWARDS

Ph.D. Student – Dehousse Fellowship – BOF/UIA - University of Antwerp 01/1999 - 01/2000
FWO-Aspirant - University of Antwerp 01/2000 - 01/2004
Travel Award - Alzheimer's Association 01/2002
Travel fellowship from the Fund for Scientific Research - Flanders (F.W.O.) 01/2002
Travel Fellowship Travel fellowship from the Fund for Scientific Research - Flanders (F.W.O.) 01/2004
Young Investigators APOPIS Award - Boehringer-Ingelheim Inc. 01/2005

Stipend for the participation in the 35th Annual Meeting of the Society for Neuroscience - Federation of European Neuroscience Societies	01/2005
Postdoctoral fellowship from the Fund for Scientific Research - Flanders (F.W.O.)	08/2005 - 08/2007
Travel fellowship from the Fund for Scientific Research - Flanders (F.W.O.)	01/2006
One-year 'Mobility fellowship' from the Fund for Scientific Research - Flanders (F.W.O.)	01/2006
"Prijs van de Onderzoeksraad VANDENDRIESSCHE" for outstanding research as a young scientist - University of Antwerp	01/2007
Junior Faculty Award at the 9th International Conference AD/PD 2009 - AD/PD 2009, Prague, Czech Republic	03/2009
2011 Jacksonville Business Journal Health Care Hero Award – Jacksonville Business Journal, Jacksonville, Florida.	10/2011

PREVIOUS PROFESSIONAL POSITIONS AND MAJOR APPOINTMENTS

Postdoctoral Fellow - University of Antwerp	2004 - 2005
Visiting Postdoctoral Scientist - College of Medicine, Mayo Clinic, Jacksonville, Florida Department of Neuroscience	2005 - 2006
Associate Consultant II - Neuroscience, Mayo Clinic, Jacksonville, Florida	08/16/2007 - 09/01/2009
Assistant Professor of Neuroscience - College of Medicine, Mayo Clinic	11/01/2007 - 02/28/2010

PROFESSIONAL & COMMUNITY MEMBERSHIPS, SOCIETIES AND SERVICES

Professional Memberships & Services

Association for Frontotemporal Dementia Medical Advisory Committee Member	01/2008 - Present
Society for Neuroscience Member	01/2007 – Present
International Society for Frontotemporal Dementia Founding Member	12/2011 - Present

EDUCATIONAL ACTIVITIES

Teaching

The Role of Tau in Chromosome 17-linked Frontotemporal Dementia and Related Tauopathies Videoconference seminar series includes both Jacksonville and Rochester sites. Mayo Clinic Alzheimer's and Parkinson's Disease Seminar Series Jacksonville, Florida	10/29/2004
Molecular Neurology and Neuropathology Advanced Course Cold Springs Harbor Laboratories Cold Spring Harbor, New York	06/06/2007 - 06/12/2007
Frontotemporal Dementia Genetics Departments of Neurology and Neuroscience 2007 - 2008 Behavioral Neurology Course Mayo Clinic College of Medicine Jacksonville, Florida	12/2007
PGRN and TDP-43: Mutation and Regulation in Neurodegenerative diseases UCSF Memory and Aging Center Grand Rounds San Francisco, California	04/25/2008
GRN and TDP-43: Mutation and Regulation in Neurodegenerative disease Videoconference seminar series includes both Jacksonville and Rochester sites. Mayo Clinic Neuroscience Works-in-Progress Seminar Series Jacksonville, Florida	05/02/2008
Molecular Neurology and Neuropathology Advanced Course Cold Springs Harbor Laboratories Cambridge, United Kingdom	06/20/2008 - 06/27/2008
Molecular Neurology and Neuropathology Advanced Course Cold Spring Harbor Laboratories Cold Spring Harbor, New York	06/03/2009 - 06/08/2009
Genetics of the frontotemporal lobar degeneration Third International Research Workshop on Frontotemporal Dementia in ALS London, Ontario, Canada	06/24/2009
Update on the genetics of Frontotemporal dementia and Amyotrophic lateral sclerosis Neuroscience Works-in-Progress Seminar Series. Videoconference seminar series includes both Jacksonville and Rochester sites. Mayo Graduate School Jacksonville, Florida	10/09/2009
The FTD and ALS disease spectrum University of Massachusetts Medical Center, MA. Neurology Grand Rounds Worcester, Massachusetts	02/23/2010
Integrated Neuroscience Session: Proteins, Genes, Neurodegeneration, and the Clinical Presentations 62nd American Academy of Neurology Annual Meeting Toronto, Canada	04/13/2010

Frontotemporal Dementia Genetics Departments of Neurology and Neuroscience, 2009 - 2010 Behavioral Neurology Course Jacksonville, Florida	04/21/2010
Progranulin regulation and its role in neurodegeneration Neuroscience Works-in-Progress Seminar Series - Mayo Clinic Jacksonville, Florida	04/22/2011
Molecular Neurology and Neuropathology Advanced Course Cold Springs Harbor Laboratories Cold Spring Harbor, New York	06/27/2011 - 07/02/2011
Frontotemporal Dementia Genetics Departments of Neurology and Neuroscience 2011 - 2012 Behavioral Neurology Course Mayo Clinic College of Medicine Jacksonville, Florida	01/25/2012
Novel insights into the genetics of frontotemporal dementia and amyotrophic lateral sclerosis Distinguished investigator Seminar, University of Florida, Gainesville, Florida	02/07/2012

Mentorship

Pellis, Mireille (Masters Student) Description: Supervisor Master Thesis Current Status: Completed PhD at the University of Brussels, Belgium Outcomes: Thesis: "Moleculaire genetische analyse van kandidaatgenen voor de ziekte van Alzheimer" (Translation: "Molecular genetic analyses of candidate genes for Alzheimer's disease")	2001 - 2002
Lesseliers, Sylvia (Masters Student) Description: Supervisor Master Thesis Current Status: Unknown Outcomes: "Identificatie en analyse van kandidaatgenen voor frontale kwab dementia gekoppeld aan chromosoom 17q21" (Translation: Co-supervisor Master Thesis: "Identification and analysis of candidate genes for frontotemporal dementia linked to chromosome 17q21")	2002 - 2003
Lyubetska, Tanya (Masters Student) Description: Supervisor Master Thesis Current Status: Unknown Outcomes: Thesis: "Genetische analyse van positionele en functionele kandidaatgenen voor de Ziekte van Alzheimer" (Translation: "Genetic analyses of positional and functional candidate genes for Alzheimer's disease")	2003 - 2004
Nuytemans, Karen (Masters Student) Description: Supervisor Master Thesis Current Status: Currently PhD student in laboratory of Prof. C. Van Broeckhoven at University of Antwerp, Belgium Outcomes: Thesis: "Opsporen van nieuwe genen voor dementie"(Translation: "Identification of novel dementia genes")	2004 - 2005

Rosa Rademakers, PhD

- Carlson, Aaron (Summer Undergraduate Research Fellow) 2007
Description: Mayo Foundation Summer Undergraduate Research Fellow program
Current Status: Current student of Mayo Medical School
Outcomes: Project: "Characterization of missense mutations in Progranulin"
- Rutherford, Nicola (Undergraduate Student) 2007 - 2008
Description: Mayo Clinic Biomedical Science Intern Program
Current Status: Completed undergraduate degree at University of Manchester, UK. Currently Special Project Associate in laboratory Dr. R. Rademakers at Mayo Clinic Jacksonville
Outcomes: Project: "Identification of mutations in TDP-43 in frontotemporal dementia and amyotrophic lateral sclerosis"
- Hamada, Masakuza (Ph.D. Student) 2007 - 2008
Description: Mayo Graduate School
Current Status: Current Mayo Graduate Student at Mayo Clinic Rochester
Outcomes: Project: "Mutation analysis of progranulin in Mayo Clinic Jacksonville dementia population"
- Swanson, Katie (Summer Undergraduate Research Fellow) 2008
Description: Mayo Foundation Summer Undergraduate Research Fellow program
Current Status: Completed undergraduate degree at University of Nebraska
Outcomes: Project: "Mutations analyses of the tau gene in Mayo Clinic Jacksonville dementia population"
- Surtees, Rebecca (Undergraduate Student) 2008 - 2009
Description: Mayo Clinic Biomedical Science Intern Program. Undergraduate student University of Leeds
Current Status: Completed degree at University of Leeds completing undergraduate degree
Outcomes: Project: "Identification of the gene responsible for frontotemporal dementia and amyotrophic lateral sclerosis linked to chromosome 9p"
- Kouri, Naomi (Graduate Student) 2009 - Present
Description: Mayo Graduate School Rotation
Current Status: Mayo Graduate School Student
Outcomes: Project: "MicroRNA regulation of MAPT in CBD and related tauopathies"
- Wojtas, Aleksandra (Summer Undergraduate Student) 2009
Current Status: Completed undergraduate degree in Biology at University of Silesia, Katowice, Poland. Currently special project associate in laboratory of Dr. Rademakers at Mayo Clinic Jacksonville.
Outcomes: Project: "Study of the chromosome 9p candidate region for FTD and ALS"

PRESENTATIONS

International

Variable Expression of Presenilin 1 is Not a Major Determinant of Risk for Late-onset Alzheimer's Disease 10th International Congress of Human Genetics Vienna, Austria	05/15/2001 - 05/19/2001
Chromosome 17 Linked Dementia in the Absence of Tau Mutations 51st Annual Meeting of the American Society of Human Genetics San Diego, California	10/12/2001 - 10/16/2001
Tau Negative Frontal Lobe Dementia at 17q21: Significant Finemapping of the Candidate Region to a 4.8cM Interval European Human Genetics Conference 2002 Strasbourg, France	05/25/2002 - 05/29/2002
Significant Finemapping of the Candidate Region to a 4.8cM Interval The 8th International Conference on Alzheimer's Disease and Related Disorders Stockholm, Sweden	07/20/2002 - 07/25/2002
Familial Dementia Caused by MAPT R406W Clinically Resembles Alzheimer's Disease The 8th International Conference on Alzheimer's Disease and Related Disorders Stockholm, Sweden	07/20/2002 - 07/25/2002
Mutation Analysis of Candidate Genes for Chromosome 17-linked Tau Negative FTD 4th International Conference on Frontotemporal Dementias Lund, Sweden	04/24/2003 - 04/26/2003
MAPT R406W Mutations Arose Independently in 3 Families from Western Europe. The 6th International Conference on AD/PD - Alzheimer's and Parkinson's Disease: New Perspectives Seville, Spain	05/08/2003 - 05/12/2003
Genomic Sequencing of MAPT Provides an Extended SNP Map and Identifies >30 H1 Subhaplotypes 4th International Conference on Frontotemporal Dementias Philadelphia, Pennsylvania	07/15/2004 - 07/16/2004
Genomic Sequencing of MAPT Provides an Extended SNP Map and Identifies >30 H1 Subhaplotypes The 9th International Conference on Alzheimer's Disease and Related Disorders Philadelphia, Pennsylvania	07/17/2004 - 07/22/2004
IPSEN Meeting Genotype - Proteotype - Phenotype Relationships in Neurodegenerative Diseases Paris, France	09/13/2004
Identification of a Novel Locus for Alzheimer's Disease at 7q36 Suggests a Role for Impaired DNA Damage Response in Dementia 34th Annual Meeting of the Society for Neuroscience San Diego, California	10/23/2004 - 10/27/2004

Rosa Rademakers, PhD

Significant Association of Specific MAPT H1 Subhaplotypes in a Belgian Epidemiological Study of Parkinson's Disease 7th International Conference AD/PD Sorrento, Italy	03/09/2005 – 03/13/2005
Genomic Architecture of Human 17q21 Uncovers Inverted Low Copy Repeats Explaining Extended Linkage Disequilibrium and Suppressed Recombination in the Tau Region Apopis Satellite Symposium Naples, Italy	03/09/2005
High Density MAPT Haplotyping Reveals an Important Role for Tau in the Pathogenesis of Parkinson's Disease 35th Annual Meeting of the American Society for Neuroscience Washington, Washington	11/13/2005
MAPT H1 Subhaplotyping in European PD Association Samples Implicates Tau Splicing in Early-Onset Parkinson's Disease Susceptibility 9th International Conference on Alzheimer's Disease and Related Disorders Madrid, Spain	07/18/2006
Mutations in Progranulin Cause Tau-Negative Frontotemporal Dementia Linked to Chromosome 17q21 5th International Conference on Frontotemporal Dementias San Francisco, California	09/06/2006
Genetic Analyses of Progranulin and TAR DNA Binding Protein in Frontotemporal Dementia and Related Neurodegenerative Diseases 8th International Conference AD/PD Salzburg, Austria	03/14/2007 - 03/18/2007
The Role of Progranulin in Dementia Alzheimer's disease: From Molecular Mechanisms to Drug Discovery meeting Punta Cana, Dominican Republic	04/20/2007
A Global Initiative to Study the Common R493X Mutation in Progranulin: Genetic and Clinicopathologic Analyses 37th Annual Meeting of the Society for Neuroscience San Diego, California	11/03/2007 - 11/07/2007
Plasma Progranulin Levels Predict Progranulin Mutation Status in Frontotemporal Dementia Patients and At-Risk Individuals 6th International Conference on Frontotemporal Dementias Rotterdam, Netherlands	09/03/2008
GRN Mutations 6th International Conference on Frontotemporal Dementias Rotterdam, Netherlands	09/03/2008
The Genetic Basis of Frontotemporal Lobar Degeneration (FTLD) 133th Annual Meeting of the American Neurological Association - Neurobiology of Disease Symposium Salt Lake City, Utah	09/21/2008
Common Variation in the miR-659 Binding-Site of Progranulin is a Major Risk Factor for TDP43-Positive Frontotemporal Dementia 38th Annual Meeting of the American Society for Neuroscience Washington, District of Columbia	11/18/2008

Common Variation in the miR-659 Binding-Site of Progranulin is a Major Risk Factor for TDP43-Positive Frontotemporal Dementia 38th Annual Meeting of the American Society for Neuroscience Washington, District of Columbia	11/18/2008
Use of a Progranulin ELISA to Identify Progranulin Mutations in Patients with Clinical Probable Alzheimer's Disease The 9th International Conference on AD/PD Prague, Czech Republic	03/12/2009
Progranulin, miRNAs and Neurodegeneration 61st Annual Meeting of the American Academy of Neurology - Hot Topics Symposium Seattle, Washington	04/28/2009
MicroRNA Dysregulation in Frontotemporal Lobar Degeneration 2009 International Conference on Alzheimer's disease(ICAD) Vienna, Austria	07/16/2009
The molecular genetics of FTD 14th International Congress of Parkinson's disease and Movement Disorders Buenos Aires, Argentina	06/16/2010
Mutation and regulation of Progranulin 7th International Conference on Frontotemporal dementias Indianapolis, Indiana	10/07/2010
Genetics of ALS – what's new and what's next? 21st International Symposium on ALS/MND - Ask The Experts Session	12/09/2010
Expanded GGGGCC hexanucleotide repeat in non-coding region of C9ORF72 causes chromosome 9p-linked FTD and ALS 6th Brain Research Conference: RNA binding proteins in neurological diseases Washington, DC	11/10/2011
National	
Tau negative frontal lobe dementia at 17q21. Significant fine mapping of the candidate region to a 4.8 cM interval. 5th VIB Seminar Blankenberge, Belgium	03/08/2002 - 03/09/2002
Update on the molecular genetic analysis of ALS and FTL D 2010 Florida Researchers Workshop ALS Association, University of South Florida Tampa, Florida	02/08/2010
Common variation in a microRNA binding site of progranulin is an important risk factor for FTL D NIH Workshop on the biology of PGRN/TDP-43 Washington, District of Columbia	02/05/2008

Update on the Genetics of Progranulin
 Director's meeting of the Alzheimer's Disease Research Centers (ADRC)
 Seattle, Washington 04/25/2009

RESEARCH GRANTS AWARDED

Active Grants

Federal

Program Director / Principal Investigator	Project 2, Dr R Petersen AG16574 Grant at MCR in: ADRC Jacksonville Consortiums years 11-15. Funded by National Institute on Aging. (P50 AG 16574)	05/2009 - 04/2014
Program Director / Principal Investigator	Progranulin: Mutation and Regulation in Neurodegenerative disease. Funded by National Institute of Neurological Disorders and Stroke. (R01 NS065782)	06/2009 - 05/2013
Co-Investigator	Progranulin, TDP-43 and Cell Death (Competitive Renewal AG026251). Funded by National Institute on Aging. (R01 R01 AG026251-03A2)	04/2010 - 03/2015

Foundation

Program Director / Principal Investigator	Identification of Genes Regulating Progranulin Expression and Processing. Funded by Frontotemporal Dementia Research.	01/2010 - 12/2011
Program Director / Principal Investigator	Understanding the ALS and FTLN disease risk associated with chromosome 9p. Funded by ALS Therapy Alliance.	08/2011 - 08/2013

Mayo Clinic

Program Director / Principal Investigator	NIH Relief Grant entitled "Molecular genetic studies of progranulin regulators in FTLN and ALS". Funded by Cost Sharing – Funded by Mayo FOundation	12/2011 - 11/2013
--	---	-------------------

Completed Grants

Federal

Program Director / Principal Investigator	Project 2, Dr R Petersen AG16574 Grant at MCR in: ADRC Jacksonville Consortiums years 6-10. Funded by National Institute on Aging. (P50 AG 16574)	06/2004 - 05/2009
Co-Investigator	Arizona Alzheimer's Research Consortium Year 10. Funded by Arizona Alzheimer's Research Center. (AGR200737)	07/2007 - 06/2008
Co-Investigator	Arizona Alzheimer's Research Consortium Year 10. Funded by Arizona Alzheimer's Research Center. (AGR2007-37 SCHEDULE 2A)	07/2007 - 06/2008

Rosa Rademakers, PhD

Co-Investigator Progranulin, TAR DNA binding protein-43 and Cell Death (Bridge Funding). Funded by National Institute on Aging. (R56 R56 AG026251-03A1 BRIDGE FUNDING) 07/2009 - 03/2010

Foundation

Principal Investigator Analyse van de rol van genetische variaties in het tau gen in progressieve supranucleaire palsy en parkinsonisme' (Translation: Study of the role of genetic variation in the tau gene in progressive supranuclear palsy and parkinsonism). Funded by Fund for Scientific Research - Flanders (F.W.O.). 01/2006 - 01/2007

Principal Investigator Rademakers Neurogenetics Core Subcontract with UBC/Stoessl for Pacific Alz Res Fnd in: Overlap Syndromes Resulting in Dementia Consortium with Dr. Stoessl at the University of British Columbia. Funded by Pacific Alzheimer Research Foundation. 01/2007 - 12/2009

Program Director / Principal Investigator MicroRNA dysregulation in frontotemporal lobar degeneration. Funded by The Association for Frontotemporal Dementias. 09/2008 - 12/2009

Program Director / Principal Investigator Molecular genetic analyses of novel and known genes implicated in ALS and ALS-FTD. Funded by ALS Association. 06/2009 - 07/2011

Program Director / Principal Investigator MicroRNA regulation of MAPT in CBD and related tauopathies. Funded by Society for Progressive Supranuclear Palsy, Inc. 06/2009 – 05/2011

PATENTS

Title	Patent number	Date filed	Date issued
Diagnostics based on a dementia-causing gene Inventors: C. Van Broeckhoven, M. Cruts and R. Radmakers	EP 41020660		12/05/2004
Detecting and Treating Dementia Inventors: M. Hutton, M. Baker, J. Gass, J. Eriksen, S. Kumar-Singh, M. Cruts, C. Van Broeckhoven, I.R. Maackenzie, H. Feldman, S. Pickering-Brown, R. Rademakers	US 12302691		11/26/2008
Methods and Materials for Detecting and Testing Dementia Inventors: R. Rademakers, J. Eriksen, M. Baker, R. Robinson, D. Dickson	US 12413869		03/30/2009
Detecting Frontotemporal dementia and Amyotrophic Lateral Sclerosis Inventors: R. Rademakers, M. DeJesus-Hernandez	Pending		Pending

BIBLIOGRAPHY

Peer-reviewed Articles

1. Dermaut B, Roks G, Theuns J, **Rademakers** R, Houwing-Duistermaat JJ, Serneels S, Hofman A, Breteler MM, Cruts M, Van Broeckhoven C, van Duijn CM. Variable expression of presenilin 1 is not a major determinant of risk for late-onset Alzheimer's disease. *J Neurol.* 2001 Nov; 248(11):935-9. PMID:11757955.
2. Cruts M, Dermaut B, **Rademakers** R, Roks G, Van den Broeck M, Munteanu G, van Duijn CM, Van Broeckhoven C. Amyloid beta secretase gene (BACE) is neither mutated in nor associated with early-onset Alzheimer's disease. *Neurosci Lett.* 2001 Nov 2; 313(1-2):105-7. PMID:11684351.
3. **Rademakers** R, Cruts M, Dermaut B, Sleegers K, Rosso SM, Van den Broeck M, Backhovens H, van Swieten J, van Duijn CM, Van Broeckhoven C. Tau negative frontal lobe dementia at 17q21: significant finemapping of the candidate region to a 4.8 cM interval. *Mol Psychiatry.* 2002; 7(10):1064-74. PMID:12476321. DOI:10.1038/sj.mp.4001198.
4. Dermaut B, Croes EA, **Rademakers** R, Van den Broeck M, Cruts M, Hofman A, van Duijn CM, Van Broeckhoven C. PRNP Val129 homozygosity increases risk for early-onset Alzheimer's disease. *Ann Neurol.* 2003 Mar; 53(3):409-12. PMID:12601712. DOI:10.1002/ana.10507.
5. **Rademakers** R, Cruts M, Van Broeckhoven C. Genetics of early-onset Alzheimer dementia. *ScientificWorldJournal.* 2003 Jun 16; 3:497-519. Epub 2003 Jun 16. PMID:12847300. DOI:10.1100/tsw.2003.39.
6. Cruts M, Dermaut B, **Rademakers** R, Van den Broeck M, Stogbauer F, Van Broeckhoven C. Novel APP mutation V715A associated with presenile Alzheimer's disease in a German family. *J Neurol.* 2003 Nov; 250(11):1374-5. PMID:14648157. DOI:10.1007/s00415-003-0182-5.
7. **Rademakers** R, Dermaut B, Peeters K, Cruts M, Heutink P, Goate A, Van Broeckhoven C. Tau (MAPT) mutation Arg406Trp presenting clinically with Alzheimer disease does not share a common founder in Western Europe. *Hum Mutat.* 2003 Nov; 22(5):409-11. PMID:14517953. DOI:10.1002/humu.10269.
8. **Rademakers** R, Van den Broeck M, Sleegers K, van Duijn C, Van Broeckhoven C, Cruts M. Absence of pathogenic mutations in presenilin homologue 2 in a conclusively 17-linked tau-negative dementia family. *Neurogenetics.* 2004 Feb; 5(1):79-80. Epub 2003 Oct 08. PMID:14534840. DOI:10.1007/s10048-003-0162-z.
9. **Rademakers** R, Cruts M, Van Broeckhoven C. The role of Tau in frontotemporal dementia and related tauopathies. *Hum Mutat.* 2004; 24:277-95.
10. Dermaut B, Kumar-Singh S, Engelborghs S, Theuns J, **Rademakers** R, Saerens J, Pickut BA, Peeters K, van den Broeck M, Vennekens K, Claes S, Cruts M, Cras P, Martin JJ, Van Broeckhoven C, De Deyn PP. A novel presenilin 1 mutation associated with Pick's disease but not beta-amyloid plaques. *Ann Neurol.* 2004 May; 55(5):617-26. PMID:15122701. DOI:10.1002/ana.20083.
11. Sleegers K, Roks G, Theuns J, Aulchenko YS, **Rademakers** R, Cruts M, van Gool WA, Van Broeckhoven C, Heutink P, Oostra BA, van Swieten JC, van Duijn CM. Familial clustering and genetic risk for dementia in a genetically isolated Dutch population. *Brain.* 2004 Jul; 127(Pt 7):1641-9. Epub 2004 May 06. PMID:15130954. DOI:10.1093/brain/awh179.
12. Weckx S, Del-Favero J, **Rademakers** R, Claes L, Cruts M, De Jonghe P, Van Broeckhoven C, De Rijk P. novoSNP, A novel computational tool for sequence variations discovery. *Genome Res.* 2005; 15(3):436-42.

13. Cruts M, **Rademakers** R, Gijselinck I, van der Zee J, Dermaut B, de Pooter T, de Rijk P, Del-Favero J, van Broeckhoven C. Genomic architecture of human 17q21 linked to frontotemporal dementia uncovers a highly homologous family of low-copy repeats in the tau region. *Hum Mol Genet.* 2005 Jul 1; 14(13):1753-62. Epub 2005 May 11. PMID:15888485.
14. **Rademakers** R, Sleegers K, Theuns J, Van den Broeck M, Bel Kacem S, Nilsson LG, Adolfsson R, van Duijn CM, Van Broeckhoven C, Cruts M. Association of cyclin-dependent kinase 5 and neuronal activators p35 and p39 complex in early-onset Alzheimer's disease. *Neurobiol Aging.* 2005 Aug-Sep; 26(8):1145-51. Epub 2004 Dec 22. PMID:15917097. DOI:10.1016/j.neurobiolaging.2004.10.003.
15. **Rademakers** R, Cruts M, Sleegers K, Dermaut B, Theuns J, Aulchenko Y, Weckx S, De Pooter T, Van den Broeck M, Corsmit E, De Rijk P, Del-Favero J, van Swieten J, van Duijn CM, Van Broeckhoven C. Linkage and association studies identify a novel locus for Alzheimer disease at 7q36 in a Dutch population-based sample. *Am J Hum Genet.* 2005 Oct; 77(4):643-52. Epub 2005 Aug 30. PMID:16175510. PMCID:1275613. DOI:10.1086/491749.
16. **Rademakers** R, Melquist S, Cruts M, Theuns J, Del-Favero J, Poorkaj P, Baker M, Sleegers K, Crook R, De Pooter T, Bel Kacem S, Adamson J, Van den Bossche D, Van den Broeck M, Gass J, Corsmit E, De Rijk P, Thomas N, Engelborghs S, Heckman M, Litvan I, Crook J, De Deyn PP, Dickson D, Schellenberg GD, Van Broeckhoven C, Hutton ML. High-density SNP haplotyping suggests altered regulation of tau gene expression in progressive supranuclear palsy. *Hum Mol Genet.* 2005 Nov 1; 14(21):3281-92. Epub 2005 Sep 29. PMID:16195395. DOI:10.1093/hmg/ddi361.
17. Dermaut B, Kumar-Singh S, **Rademakers** R, Theuns J, Cruts M, Van Broeckhoven C. Tau is central in the genetic Alzheimer-frontotemporal dementia spectrum. *Trends Genet.* 2005 Dec; 21(12):664-72. Epub 2005 Oct 10. PMID:16221505. DOI:10.1016/j.tig.2005.09.005.
18. van der Zee J, **Rademakers** R, Engelborghs S, Gijselinck I, Bogaerts V, Vandenberghe R, Santens P, Caekebeke J, De Pooter T, Peeters K, Lübke U, Van den Broeck M, Martin J-J, Cruts M, De Deyn PP, Van Broeckhoven C, Dermaut B. An Ancestral Haplotype Harbors a Highly Prevalent Mutation for 17q21-Linked Tau-negative FTLD in Belgium. *Brain.* 2006; 129(4):841-52.
19. Engelborghs S, Dermaut B, Marien P, Symons A, Vloeberghs E, Maertens K, Somers N, Goeman J, **Rademakers** R, Van den Broeck M, Pickut B, Cruts M, Van Broeckhoven C, De Deyn PP. Dose dependent effect of APOE epsilon4 on behavioral symptoms in frontal lobe dementia. *Neurobiol Aging.* 2006 Feb; 27(2):285-92. PMID:16399213. DOI:10.1016/j.neurobiolaging.2005.02.005.
20. Pirici D, Vandenberghe R, **Rademakers** R, Dermaut B, Cruts M, Vennekens K, Cuijt I, Lubke U, Ceuterick C, Martin JJ, Van Broeckhoven C, Kumar-Singh S. Characterization of ubiquitinated intraneuronal inclusions in a novel Belgian frontotemporal lobar degeneration family. *J Neuropathol Exp Neurol.* 2006 Mar; 65(3):289-301. PMID:16651890. DOI:10.1097/01.jnen.0000205147.39210.c7.
21. Baker M, Mackenzie IR, Pickering-Brown SM, Gass J, **Rademakers** R, Lindholm C, Snowden J, Adamson J, Sadovnick AD, Rollinson S, Cannon A, Dwosh E, Neary D, Melquist S, Richardson A, Dickson D, Berger Z, Eriksen J, Robinson T, Zehr C, Dickey CA, Crook R, McGowan E, Mann D, Boeve B, Feldman H, Hutton M. Mutations in progranulin cause tau-negative frontotemporal dementia linked to chromosome 17. *Nature.* 2006 Aug 24; 442(7105):916-9. Epub 2006 Jul 16. PMID:16862116. DOI:10.1038/nature05016.

22. Cruts M, Gijselinck I, van der Zee J, Engelborghs S, Wils H, Pirici D, **Rademakers R**, Vandenberghe R, Dermaut B, Martin JJ, van Duijn C, Peeters K, Sciot R, Santens P, De Pooter T, Mattheijssens M, Van den Broeck M, Cuijt I, Vennekens K, De Deyn PP, Kumar-Singh S, Van Broeckhoven C. Null mutations in progranulin cause ubiquitin-positive frontotemporal dementia linked to chromosome 17q21. *Nature*. 2006 Aug 24; 442(7105):920-4. Epub 2006 Jul 16. PMID:16862115. DOI:10.1038/nature05017.
23. Gass J, Cannon A, Mackenzie IR, Boeve B, Baker M, Adamson J, Crook R, Melquist S, Kuntz K, Petersen R, Josephs K, Pickering-Brown SM, Graff-Radford N, Uitti R, Dickson D, Wszolek Z, Gonzalez J, Beach TG, Bigio E, Johnson N, Weintraub S, Mesulam M, White CL 3rd, Woodruff B, Caselli R, Hsiung GY, Feldman H, Knopman D, Hutton M, **Rademakers R**. Mutations in progranulin are a major cause of ubiquitin-positive frontotemporal lobar degeneration. *Hum Mol Genet*. 2006 Oct 15; 15(20):2988-3001. Epub 2006 Sep 01. PMID:16950801. DOI:10.1093/hmg/ddl241.
24. Boeve BF, Baker M, Dickson DW, Parisi JE, Giannini C, Josephs KA, Hutton M, Pickering-Brown SM, **Rademakers R**, Tang-Wai D, Jack CR Jr, Kantarci K, Shiung MM, Golde T, Smith GE, Geda YE, Knopman DS, Petersen RC. Frontotemporal dementia and parkinsonism associated with the IVS1+1G->A mutation in progranulin: a clinicopathologic study. *Brain*. 2006 Nov; 129(Pt 11):3103-14. Epub 2006 Oct 09. PMID:17030535. DOI:10.1093/brain/awl268.
25. Gijselinck I, Bogaerts V, **Rademakers R**, van der Zee J, Van Broeckhoven C, Cruts M. Visualization of MAPT inversion on stretched chromosomes of tau-negative frontotemporal dementia patients. *Hum Mutat*. 2006 Oct; 27(10):1057-9. PMID:16906510. DOI:10.1002/humu.20391.
26. Mackenzie IR, Baker M, Pickering-Brown S, Hsiung GY, Lindholm C, Dwosh E, Gass J, Cannon A, **Rademakers R**, Hutton M, Feldman HH. The neuropathology of frontotemporal lobar degeneration caused by mutations in the progranulin gene. *Brain*. 2006 Nov; 129(Pt 11):3081-90. PMID:17071926. DOI:10.1093/brain/awl271.
27. Dickson DW, **Rademakers R**, Hutton ML. Progressive supranuclear palsy: pathology and genetics. *Brain Pathol*. 2007 Jan; 17(1):74-82. PMID:17493041. DOI:10.1111/j.1750-3639.2007.00054.x.
28. Josephs KA, Ahmed Z, Katsuse O, Parisi JF, Boeve BF, Knopman DS, Petersen RC, Davies P, Duara R, Graff-Radford NR, Uitti RJ, **Rademakers R**, Adamson J, Baker M, Hutton ML, Dickson DW. Neuropathologic features of frontotemporal lobar degeneration with ubiquitin-positive inclusions with progranulin gene (PGRN) mutations. *J Neuropathol Exp Neurol*. 2007 Feb; 66(2):142-51. PMID:17278999. DOI:10.1097/nen.0b013e31803020cf.
29. Whitwell JL, Jack CR Jr, Baker M, **Rademakers R**, Adamson J, Boeve BF, Knopman DS, Parisi JF, Petersen RC, Dickson DW, Hutton ML, Josephs KA. Voxel-based morphometry in frontotemporal lobar degeneration with ubiquitin-positive inclusions with and without progranulin mutations. *Arch Neurol*. 2007 Mar; 64(3):371-6. PMID:17353379. PMCID:2752412. NIHMS:137138. DOI:10.1001/archneur.64.3.371.
30. Schymick JC, Yang Y, Andersen PM, Vonsattel JP, Greenway M, Momeni P, Elder J, Chio A, Restagno G, Robberecht W, Dahlberg C, Mukherjee O, Goate A, Graff-Radford N, Caselli RJ, Hutton M, Gass J, Cannon A, **Rademakers R**, Singleton AB, Hardiman O, Rothstein J, Hardy J, Traynor BJ. Progranulin mutations and amyotrophic lateral sclerosis or amyotrophic lateral sclerosis-frontotemporal dementia phenotypes. *J Neurol Neurosurg Psychiatry*. 2007 Jul; 78(7):754-6 Epub 2007 Mar 19. PMID:17371905. PMCID:2117704. DOI:10.1136/jnnp.2006.109553.
31. **Rademakers R**, Hutton M. The genetics of frontotemporal lobar degeneration. *Curr Neurol Neurosci Rep*. 2007 Sep; 7(5):434-42. PMID:17764635.

32. Davion S, Johnson N, Weintraub S, Mesulam MM, Engberg A, Mishra M, Baker M, Adamson J, Hutton M, **Rademakers R**, Bigio EH. Clinicopathologic correlation in PGRN mutations. *Neurology*. 2007 Sep 11; 69(11):1113-21. Epub 2007 May 23. PMID:17522386. DOI:10.1212/01.wnl.0000267701.58488.69.
33. **Rademakers R**, Baker M, Gass J, Adamson J, Huey ED, Momeni P, Spina S, Coppola G, Karydas AM, Stewart H, Johnson N, Hsiung GY, Kelley B, Kuntz K, Steinbart E, Wood EM, Yu CE, Josephs K, Sorenson E, Womack KB, Weintraub S, Pickering-Brown SM, Schofield PR, Brooks WS, Van Deerlin VM, Snowden J, Clark CM, Kertesz A, Boylan K, Ghetti B, Neary D, Schellenberg GD, Beach TG, Mesulam M, Mann D, Grafman J, Mackenzie IR, Feldman H, Bird T, Petersen R, Knopman D, Boeve B, Geschwind DH, Miller B, Wszolek Z, Lippa C, Phenotypic variability associated with progranulin haploinsufficiency in patients with the common 1477C-->T (Arg493X) mutation: an international initiative. *Lancet Neurol*. 2007 Oct; 6(10):857-68. PMID:17826340. DOI:10.1016/S1474-4422(07)70221-1.
34. Mackenzie IR, **Rademakers R**. The molecular genetics and neuropathology of frontotemporal lobar degeneration: recent developments. *Neurogenetics*. 2007 Nov; 8(4):237-48. Epub 2007 Sep 06. PMID:17805587. DOI:10.1007/s10048-007-0102-4.
35. Rutherford NJ, Zhang YJ, Baker M, Gass JM, Finch NA, Xu YF, Stewart H, Kelley BJ, Kuntz K, Crook RJ, Sreedharan J, Vance C, Sorenson E, Lippa C, Bigio EH, Geschwind DH, Knopman DS, Mitsumoto H, Petersen RC, Cashman NR, Hutton M, Shaw CE, Boylan KB, Boeve B, Graff-Radford NR, Wszolek ZK, Caselli RJ, Dickson DW, Mackenzie IR, Petrucelli L, **Rademakers R**. Novel mutations in TARDBP (TDP-43) in patients with familial amyotrophic lateral sclerosis. *PLoS Genet*. 2008; 4(9):e1000193. Epub 2008 Sep 19. PMID:18802454. PMCID:2527686. DOI:10.1371/journal.pgen.1000193.
36. Gitcho MA, Baloh RH, Chakraverty S, Mayo K, Norton JB, Levitch D, Hatanpaa KJ, White CL 3rd, Bigio EH, Caselli R, Baker M, Al-Lozi MT, Morris JC, Pestronk A, **Rademakers R**, Goate AM, Cairns NJ. TDP-43 A315T mutation in familial motor neuron disease. *Ann Neurol*. 2008 Apr; 63(4):535-8. PMID:18288693. PMCID:2747362. NIHMS:128105. DOI:10.1002/ana.21344.
37. Nuytemans K, **Rademakers R**, Theuns J, Pals P, Engelborghs S, Pickut B, de Pooter T, Peeters K, Mattheijssens M, Van den Broeck M, Cras P, De Deyn PP, van Broeckhoven C. Founder mutation p.R1441C in the leucine-rich repeat kinase 2 gene in Belgian Parkinson's disease patients. *Eur J Hum Genet*. 2008 Apr; 16(4):471-9. Epub 2008 Jan 16. PMID:18197194. DOI:10.1038/sj.ejhg.5201986.
38. Haugarvoll K, **Rademakers R**, Kachergus JM, Nuytemans K, Ross OA, Gibson JM, Tan EK, Gaig C, Tolosa E, Goldwurm S, Guidi M, Riboldazzi G, Brown L, Walter U, Benecke R, Berg D, Gasser T, Theuns J, Pals P, Cras P, De Deyn PP, Engelborghs S, Pickut B, Uitti RJ, Foroud T, Nichols WC, Hagenah J, Klein C, Samii A, Zabetian CP, Bonifati V, Van Broeckhoven C, Farrer MJ, Wszolek ZK. Lrrk2 R1441C parkinsonism is clinically similar to sporadic Parkinson disease. *Neurology*. 2008 Apr 15; 70(16 Pt 2):1456-60. Epub 2008 Mar 12. PMID:18337586. DOI:10.1212/01.wnl.0000304044.22253.03.
39. Josephs KA, Whitwell JL, Knopman DS, Hu WT, Stroh DA, Baker M, **Rademakers R**, Boeve BF, Parisi JE, Smith GE, Ivnik RJ, Petersen RC, Jack CR Jr, Dickson DW. Abnormal TDP-43 immunoreactivity in AD modifies clinicopathologic and radiologic phenotype. *Neurology*. 2008 May 6; 70(19 Pt 2):1850-7. Epub 2008 Apr 09. PMID:18401022. PMCID:2779031. NIHMS:127928. DOI:10.1212/01.wnl.0000304041.09418.b1.
40. Wider C, Uitti RJ, Wszolek ZK, Fang JY, Josephs KA, Baker MC, **Rademakers R**, Hutton ML, Dickson DW. Progranulin gene mutation with an unusual clinical and neuropathologic presentation. *Mov Disord*. 2008 Jun 15; 23(8):1168-73. PMID:18442119. DOI:10.1002/mds.22065.

41. Winton MJ, Van Deerlin VM, Kwong LK, Yuan W, Wood EM, Yu CE, Schellenberg GD, **Rademakers** R, Caselli R, Karydas A, Trojanowski JQ, Miller BL, Lee VM. A90V TDP-43 variant results in the aberrant localization of TDP-43 in vitro. *FEBS Lett.* 2008 Jun 25; 582(15):2252-6. Epub 2008 May 27. PMID:18505686. PMCID:2478749. NIHMS:56338. DOI:10.1016/j.febslet.2008.05.024.
42. Coppola G, Karydas A, **Rademakers** R, Wang Q, Baker M, Hutton M, Miller BL, Geschwind DH. Gene expression study on peripheral blood identifies progranulin mutations. *Ann Neurol.* 2008 Jul; 64(1):92-6. PMID:18551524. PMCID:2773201. NIHMS:135325. DOI:10.1002/ana.21397.
43. Mackenzie IR, **Rademakers** R. The role of transactive response DNA-binding protein-43 in amyotrophic lateral sclerosis and frontotemporal dementia. *Curr Opin Neurol.* 2008 Dec; 21(6):693-700. PMID:18989115. PMCID:2869081. DOI:10.1097/WCO.0b013e3283168d1d.
44. **Rademakers** R, Eriksen JL, Baker M, Robinson T, Ahmed Z, Lincoln SJ, Finch N, Rutherford NJ, Crook RJ, Josephs KA, Boeve BF, Knopman DS, Petersen RC, Parisi JE, Caselli RJ, Wszolek ZK, Uitti RJ, Feldman H, Hutton ML, Mackenzie IR, Graff-Radford NR, Dickson DW. Common variation in the miR-659 binding-site of GRN is a major risk factor for TDP43-positive frontotemporal dementia. *Hum Mol Genet.* 2008 Dec 1; 17(23):3631-42. Epub 2008 Aug 21. PMID:18723524. PMCID:2581433. DOI:10.1093/hmg/ddn257.
45. Jawaid A, **Rademakers** R, Kass JS, Kalkonde Y, Schulz PE. Traumatic brain injury may increase the risk for frontotemporal dementia through reduced progranulin. *Neurodegener Dis.* 2009; 6(5-6):219-20. Epub 2010 Feb 10. PMID:20145419. PMCID:2837889. DOI:10.1159/000258704.
46. Finch N, Baker M, Crook R, Swanson K, Kuntz K, Surtees R, Bisceglia G, Rovelet-Lecrux A, Boeve B, Petersen RC, Dickson DW, Younkin SG, Deramecourt V, Crook J, Graff-Radford NR, **Rademakers** R. Plasma progranulin levels predict progranulin mutation status in frontotemporal dementia patients and asymptomatic family members. *Brain.* 2009 Mar; 132(Pt 3):583-91. Epub 2009 Jan 21. PMID:19158106. PMCID:2664450. DOI:10.1093/brain/awn352.
47. Whitwell JL, Jack CR Jr, Boeve BF, Senjem ML, Baker M, **Rademakers** R, Ivnik RJ, Knopman DS, Wszolek ZK, Petersen RC, Josephs KA. Voxel-based morphometry patterns of atrophy in FTL D with mutations in MAPT or PGRN. *Neurology.* 2009 Mar 3; 72(9):813-20. PMID:19255408. PMCID:2677544. DOI:10.1212/01.wnl.0000343851.46573.67.
48. Rollinson S, Rizzu P, Sikkink S, Baker M, Halliwell N, Snowden J, Traynor BJ, Ruano D, Cairns N, Rohrer JD, Mead S, Collinge J, Rossor M, Akay E, Guerreiro R, **Rademakers** R, Morrison KE, Pastor P, Alonso E, Martinez-Lage P, Graff-Radford N, Neary D, Heutink P, Mann DM, Van Swieten J, Pickering-Brown SM. Ubiquitin associated protein 1 is a risk factor for frontotemporal lobar degeneration. *Neurobiol Aging.* 2009 Apr; 30(4):656-65. Epub 2009 Feb 12. PMID:19217189. PMCID:2753870. NIHMS:104787. DOI:10.1016/j.neurobiolaging.2009.01.009.
49. Kelley BJ, Haidar W, Boeve BF, Baker M, Graff-Radford NR, Krefft T, Frank AR, Jack CR Jr, Shiung M, Knopman DS, Josephs KA, Parashos SA, **Rademakers** R, Hutton M, Pickering-Brown S, Adamson J, Kuntz KM, Dickson DW, Parisi JE, Smith GE, Ivnik RJ, Petersen RC. Prominent phenotypic variability associated with mutations in Progranulin. *Neurobiol Aging.* 2009 May; 30(5):739-51. Epub 2007 Oct 18. PMID:17949857. PMCID:3164546. NIHMS:318035. DOI:10.1016/j.neurobiolaging.2007.08.022.
50. Wider C, Dickson DW, Stoessl AJ, Tsuboi Y, Chapon F, Gutmann L, Lechevalier B, Calne DB, Personett DA, Hulihan M, Kachergus J, **Rademakers** R, Baker MC, Grantier LL, Sujith OK, Brown L, Calne S, Farrer MJ, Wszolek ZK. Pallidonigral TDP-43 pathology in Perry syndrome. *Parkinsonism Relat Disord.* 2009 May; 15(4):281-6. Epub 2008 Aug 23. PMID:18723384. PMCID:2693935. NIHMS:115126. DOI:10.1016/j.parkreldis.2008.07.005.

51. Vilarino-Guell C, Wider C, Soto-Ortolaza AI, Cobb SA, Kachergus JM, Keeling BH, Dachsel JC, Hulihan MM, Dickson DW, Wszolek ZK, Uitti RJ, Graff-Radford NR, Boeve BF, Josephs KA, Miller B, Boylan KB, Gwinn K, Adler CH, Aasly JO, Hentati F, Destee A, Krygowska-Wajs A, Chartier-Harlin MC, Ross OA, **Rademakers R**, Farrer MJ. Characterization of DCTN1 genetic variability in neurodegeneration. *Neurology*. 2009 Jun 9; 72(23):2024-8. PMID:19506225. PMCID:2692178. DOI:10.1212/WNL.0b013e3181a92c4c.
52. Caselli RJ, Dueck AC, Osborne D, Sabbagh MN, Connor DJ, Ahern GL, Baxter LC, Rapcsak SZ, Shi J, Woodruff BK, Locke DE, Snyder CH, Alexander GE, **Rademakers R**, Reiman EM. Longitudinal modeling of age-related memory decline and the APOE epsilon4 effect. *N Engl J Med*. 2009 Jul 16; 361(3):255-63. PMID:19605830. PMCID:2928998. NIHMS:182704. DOI:10.1056/NEJMoa0809437.
53. Jasinska-Myga B, Wider C, Opala G, Krygowska-Wajs A, Barcikowska M, Czyzewski K, Baker M, **Rademakers R**, Uitti RJ, Farrer MJ, Ross OA, Wszolek ZK. GRN 3'UTR+78 C>T is not associated with risk for Parkinson's disease. *Eur J Neurol*. 2009 Aug; 16(8):909-11. Epub 2009 Mar 31. PMID:19473366. DOI:10.1111/j.1468-1331.2009.02621.x.
54. **Rademakers R**, Rovelet-Lecrux A. Recent insights into the molecular genetics of dementia. *Trends Neurosci*. 2009 Aug; 32(8):451-61. Epub 2009 Jul 27. PMID:19640594. PMCID:2864783. NIHMS:199048. DOI:10.1016/j.tins.2009.05.005.
55. Sitek EJ, Narozanska E, Slawek J, Wieczorek D, Brockhuis B, Lass P, Dubaniewicz M, Jasinska-Myga B, Baker M, **Rademakers R**, Wszolek ZK. Unilateral neglect in a patient diagnosed with frontotemporal dementia and parkinsonism linked to chromosome 17. *Acta Neuropsychiatr*. 2009 Aug; 21(4):209-10.
56. Whitwell JL, Jack CR Jr, Boeve BF, Senjem ML, Baker M, Ivnik RJ, Knopman DS, Wszolek ZK, Petersen RC, **Rademakers R**, Josephs KA. Atrophy patterns in IVS10+16, IVS10+3, N279K, S305N, P301L, and V337M MAPT mutations. *Neurology*. 2009 Sep 29; 73(13):1058-65. PMID:19786698. PMCID:2754325. DOI:10.1212/WNL.0b013e3181b9c8b9.
57. Gitcho MA, Bigio EH, Mishra M, Johnson N, Weintraub S, Mesulam M, **Rademakers R**, Chakraverty S, Cruchaga C, Morris JC, Goate AM, Cairns NJ. TARDBP 3'-UTR variant in autopsy-confirmed frontotemporal lobar degeneration with TDP-43 proteinopathy. *Acta Neuropathol*. 2009 Nov; 118(5):633-45. Epub 2009 Jul 18. PMID:19618195. PMCID:2783457. NIHMS:133832. DOI:10.1007/s00401-009-0571-7.
58. Neumann M, **Rademakers R**, Roeber S, Baker M, Kretschmar HA, Mackenzie IR. A new subtype of frontotemporal lobar degeneration with FUS pathology. *Brain*. 2009 Nov; 132(Pt 11):2922-31. Epub 2009 Aug 11. PMID:19674978. PMCID:2768659. DOI:10.1093/brain/awp214.
59. Neumann M, Roeber S, Kretschmar HA, **Rademakers R**, Baker M, Mackenzie IR. Abundant FUS-immunoreactive pathology in neuronal intermediate filament inclusion disease. *Acta Neuropathol*. 2009 Nov; 118(5):605-16. Epub 2009 Aug 09. PMID:19669651. PMCID:2864784. DOI:10.1007/s00401-009-0581-5.
60. Dickson DW, Baker M, **Rademakers R**. Common variant in GRN is a genetic risk factor for hippocampal sclerosis in the elderly. *Neurodegener Dis*. 2010; 7(1-3):170-4. Epub 2010 Mar 03. PMID:20197700. PMCID:2859236. DOI:10.1159/000289231.
61. Kelley BJ, Haidar W, Boeve BF, Baker M, Shiung M, Knopman DS, **Rademakers R**, Hutton M, Adamson J, Kuntz KM, Dickson DW, Parisi JE, Smith GE, Petersen RC. Alzheimer disease-like phenotype associated with the c.154delA mutation in progranulin. *Arch Neurol*. 2010 Feb; 67(2):171-7. PMID:20142525. PMCID:2902004. NIHMS:198343. DOI:10.1001/archneurol.2010.113.

62. Tartaglia MC, Sidhu M, Laluz V, Racine C, Rabinovici GD, Creighton K, Karydas A, **Rademakers R**, Huang EJ, Miller BL, Dearmond SJ, Seeley WW. Sporadic corticobasal syndrome due to FTLN-TDP. *Acta Neuropathol.* 2010 Mar; 119(3):365-374. Epub 2009 Oct 30. PMID:19876635. PMCID:2832091. NIHMS:169120. DOI:10.1007/s00401-009-0605-1.
63. Van Deerlin VM, Sleiman PM, Martinez-Lage M, Chen-Plotkin A, Wang LS, Graff-Radford NR, Dickson DW, **Rademakers R**, Boeve BF, Grossman M, Arnold SE, Mann DM, Pickering-Brown SM, Seelaar H, Heutink P, van Swieten JC, Murrell JR, Ghetti B, Spina S, Grafman J, Hodges J, Spillantini MG, Gilman S, Lieberman AP, Kaye JA, Woltjer RL, Bigio EH, Mesulam M, Al-Sarraj S, Troakes C, Rosenberg RN, White CL 3rd, Ferrer I, Llado A, Neumann M, Kretschmar HA, Hulette CM, Welsh-Bohmer KA, Miller BL, Alzualde A, de Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. *Nat Genet.* 2010 Mar; 42(3):234-9. Epub 2010 Feb 14. PMID:20154673. PMCID:2828525. NIHMS:172758. DOI:10.1038/ng.536.
64. Sitek EJ, Narozanska E, Slawek J, Wojcik J, Wieczorek D, Robowski P, Schinwelski M, Jasinska-Myga B, Baker M, **Rademakers R**, Wszolek ZK. Psychometric evaluation of personality in a patient with FTDP-17. *Psychiatry Clin Neurosci.* 2010 Apr; 64(2):211-2. PMID:20447016. DOI:10.1111/j.1440-1819.2009.02056.x.
65. DeJesus-Hernandez M, Kocerha J, Finch N, Crook R, Baker M, Desaro P, Johnston A, Rutherford N, Wojtas A, Kennelly K, Wszolek ZK, Graff-Radford N, Boylan K, **Rademakers R**. De novo truncating FUS gene mutation as a cause of sporadic amyotrophic lateral sclerosis. *Hum Mutat.* 2010 May; 31(5):E1377-89. PMID:20232451. PMCID:2922682. NIHMS:220421. DOI:10.1002/humu.21241.
66. Urwin H, Josephs KA, Rohrer JD, Mackenzie IR, Neumann M, Authier A, Seelaar H, Van Swieten JC, Brown JM, Johannsen P, Nielsen JE, Holm IE, FReJA Consortium, Dickson DW, **Rademakers R**, Graff-Radford NR, Parisi JE, Petersen RC, Hatanpaa KJ, White CL 3rd, Weiner MF, Geser F, Van Deerlin VM, Trojanowski JQ, Miller BL, Seeley WW, van der Zee J, Kumar-Singh S, Engelborghs S, De Deyn PP, Van Broeckhoven C, Bigio EH, Deng HX, Halliday GM, Kril JJ, Munoz DG, Mann DM, Pickering-Brown SM, Doodeman V, Adamson G, FUS pathology defines the majority of tau- and TDP-43-negative frontotemporal lobar degeneration. *Acta Neuropathol.* 2010 Jul; 120(1):33-41. Epub 2010 May 20. PMID:20490813. PMCID:2887939. NIHMS:214921. DOI:10.1007/s00401-010-0698-6.
67. **Rademakers R**, Stewart H, DeJesus-Hernandez M, Krieger C, Graff-Radford N, Fabros M, Briemberg H, Cashman N, Eisen A, Mackenzie IR. Fus gene mutations in familial and sporadic amyotrophic lateral sclerosis. *Muscle Nerve.* 2010 Aug; 42(2):170-6. PMID:20544928. PMCID:2969843. NIHMS:237353. DOI:10.1002/mus.21665.
68. Xu YF, Gendron TF, Zhang YJ, Lin WL, D'Alton S, Sheng H, Casey MC, Tong J, Knight J, Yu X, **Rademakers R**, Boylan K, Hutton M, McGowan E, Dickson DW, Lewis J, Petrucelli L. Wild-type human TDP-43 expression causes TDP-43 phosphorylation, mitochondrial aggregation, motor deficits, and early mortality in transgenic mice. *J Neurosci.* 2010 Aug 11; 30(32):10851-9. PMID:20702714. PMCID:3056148. NIHMS:227749. DOI:10.1523/JNEUROSCI.1630-10.2010.
69. Kantarci K, Boeve BF, Wszolek ZK, **Rademakers R**, Whitwell JL, Baker MC, Senjem ML, Samikoglu AR, Knopman DS, Petersen RC, Jack CR Jr. MRS in presymptomatic MAPT mutation carriers: a potential biomarker for tau-mediated pathology. *Neurology.* 2010 Aug 31; 75(9):771-8. PMID:20805522. PMCID:2938968. DOI:10.1212/WNL.0b013e3181f073c7.
70. Mackenzie IR, **Rademakers R**, Neumann M. TDP-43 and FUS in amyotrophic lateral sclerosis and frontotemporal dementia. *Lancet Neurol.* 2010 Oct; 9(10):995-1007. PMID:20864052. DOI:10.1016/S1474-4422(10)70195-2.

71. Carrasquillo MM, Nicholson AM, Finch N, Gibbs JR, Baker M, Rutherford NJ, Hunter TA, DeJesus-Hernandez M, Bisceglia GD, Mackenzie IR, Singleton A, Cookson MR, Crook JE, Dillman A, Hernandez D, Petersen RC, Graff-Radford NR, Younkin SG, **Rademakers R**. Genome-wide screen identifies rs646776 near sortilin as a regulator of progranulin levels in human plasma. *Am J Hum Genet*. 2010 Dec 10; 87(6):890-7. Epub 2010 Nov 18. PMID:21087763. PMCID:2997361. DOI:10.1016/j.ajhg.2010.11.002.
72. Whitwell JL, Jack CR Jr, Parisi JE, Senjem ML, Knopman DS, Boeve BF, **Rademakers R**, Baker M, Petersen RC, Dickson DW, Josephs KA. Does TDP-43 type confer a distinct pattern of atrophy in frontotemporal lobar degeneration? *Neurology*. 2010 Dec 14; 75(24):2212-20. PMID:21172844. PMCID:3013590. DOI:10.1212/WNL.0b013e31820203c2.
73. Doppert EGP, Seelaar H, Chiu WZ, De Koning I, Van Minkelen R, Baker MC, Rozemuller AJM, **Rademakers R**, Van Swieten JC. Symmetrical corticobasal syndrome caused by a novel c.314dup progranulin mutation. *J Mol Neurosci*. 2011; 45(3):354-8.
74. Kocerha J, Kouri N, Baker M, Finch N, DeJesus-Hernandez M, Gonzalez J, Chidamparam K, Josephs KA, Boeve BF, Graff-Radford NR, Crook J, Dickson DW, **Rademakers R**. Altered microRNA expression in frontotemporal lobar degeneration with TDP-43 pathology caused by progranulin mutations. *BMC Genomics*. 2011; 12:527. Epub 2011 Oct 27. PMID:22032330. PMCID:3229715. DOI:10.1186/1471-2164-12-527.
75. **Rademakers R**, Baker M, Nicholson AM, Rutherford NJ, Finch N, Soto-Ortolaza A, Lash J, Wider C, Wojtas A, DeJesus-Hernandez M, Adamson J, Kouri N, Sundal C, Shuster EA, Aasly J, Mackenzie J, Roeber S, Kretzschmar HA, Boeve BF, Knopman DS, Petersen RC, Cairns NJ, Ghetti B, Spina S, Garbern J, Tselis AC, Uitti R, Das P, Van Gerpen JA, Meschia JF, Levy S, Broderick DF, Graff-Radford N, Ross OA, Miller BB, Swerdlow RH, Dickson DW, Wszolek ZK. Mutations in the colony stimulating factor 1 receptor (CSF1R) gene cause hereditary diffuse leukoencephalopathy with spheroids. *Nat Genet*. 2011; 44(2):200-5. Epub 2011 Dec 25. PMID:22197934. PMCID:3267847. NIHMS:336734. DOI:10.1038/ng.1027.
76. Hsiung GY, Fok A, Feldman HH, **Rademakers R**, Mackenzie IR. rs5848 polymorphism and serum progranulin level. *J Neurol Sci*. 2011 Jan 15; 300(1-2):28-32. Epub 2010 Nov 02. PMID:21047645. PMCID:3085023. NIHMS:287104. DOI:10.1016/j.jns.2010.10.009.
77. Boxer AL, Mackenzie IR, Boeve BF, Baker M, Seeley WW, Crook R, Feldman H, Hsiung GY, Rutherford N, Laluz V, Whitwell J, Foti D, McDade E, Molano J, Karydas A, Wojtas A, Goldman J, Mirsky J, Sengdy P, Dearmond S, Miller BL, **Rademakers R**. Clinical, neuroimaging and neuropathological features of a new chromosome 9p-linked FTD-ALS family. *J Neurol Neurosurg Psychiatry*. 2011 Feb; 82(2):196-203. Epub 2010 Jun 20. PMID:20562461. PMCID:3017222. NIHMS:257687. DOI:10.1136/jnnp.2009.204081.
78. Finch N, Carrasquillo MM, Baker M, Rutherford NJ, Coppola G, DeJesus-Hernandez M, Crook R, Hunter T, Ghidoni R, Benussi L, Crook J, Finger E, Hantanpaa KJ, Karydas AM, Sengdy P, Gonzalez J, Seeley WW, Johnson N, Beach TG, Mesulam M, Forloni G, Kertesz A, Knopman DS, Uitti R, White CL 3rd, Caselli R, Lippa C, Bigio EH, Wszolek ZK, Binetti G, Mackenzie IR, Miller BL, Boeve BF, Younkin SG, Dickson DW, Petersen RC, Graff-Radford NR, Geschwind DH, **Rademakers R**. TMEM106B regulates progranulin levels and the penetrance of FTL in GRN mutation carriers. *Neurology*. 2011 Feb 1; 76(5):467-74. Epub 2010 Dec 22. PMID:21178100. PMCID:3034409. DOI:10.1212/WNL.0b013e31820a0e3b.
79. Goldman JS, **Rademakers R**, Huey ED, Boxer AL, Mayeux R, Miller BL, Boeve BF. An algorithm for genetic testing of frontotemporal lobar degeneration. *Neurology*. 2011 Feb 1; 76(5):475-83. PMID:21282594. PMCID:3034412. DOI:10.1212/WNL.0b013e31820a0d13.

80. Narozanska E, Jasinska-Myga B, Sitek EJ, Robowski P, Brockhuis B, Lass P, Dubaniewicz M, Wieczorek D, Baker M, **Rademakers R**, Wszolek ZK, Slawek J. Frontotemporal dementia and parkinsonism linked to chromosome 17--the first Polish family. *Eur J Neurol*. 2011 Mar; 18(3):535-7. PMID:20561037. PMCID:2944014. NIHMS:204252. DOI:10.1111/j.1468-1331.2010.03107.x.
81. Caselli RJ, Dueck AC, Locke DE, Sabbagh MN, Ahern GL, Rapcsak SZ, Baxter LC, Yaari R, Woodruff BK, Hoffman-Snyder C, **Rademakers R**, Findley S, Reiman EM. Cerebrovascular risk factors and preclinical memory decline in healthy APOE epsilon4 homozygotes. *Neurology*. 2011 Mar 22; 76(12):1078-84. Epub 2011 Feb 16. PMID:21325652. PMCID:3068011. DOI:10.1212/WNL.0b013e318211c3ae.
82. Chen-Plotkin AS, Martinez-Lage M, Sleiman PM, Hu W, Greene R, Wood EM, Bing S, Grossman M, Schellenberg GD, Hatanpaa KJ, Weiner MF, White CL 3rd, Brooks WS, Halliday GM, Kril JJ, Gearing M, Beach TG, Graff-Radford NR, Dickson DW, **Rademakers R**, Boeve BF, Pickering-Brown SM, Snowden J, van Swieten JC, Heutink P, Seelaar H, Murrell JR, Ghetti B, Spina S, Grafman J, Kaye JA, Woltjer RL, Mesulam M, Bigio E, Llado A, Miller BL, Alzualde A, Moreno F, Rohrer JD, Mackenzie IR, Feldman HH, Hamilton RL, Cruts M, Genetic and clinical features of progranulin-associated frontotemporal lobar degeneration. *Arch Neurol*. 2011 Apr; 68(4):488-97. PMID:21482928. PMCID:3160280. NIHMS:318289. DOI:10.1001/archneurol.2011.53.
83. Kouri N, Whitwell JL, Josephs KA, **Rademakers R**, Dickson DW. Corticobasal degeneration: a pathologically distinct 4R tauopathy. *Nat Rev Neurol*. 2011 May; 7(5):263-72. Epub 2011 Apr 12. PMID:21487420. DOI:10.1038/nrneurol.2011.43.
84. Hoglinger GU, Melhem NM, Dickson DW, Sleiman PM, Wang LS, Klei L, **Rademakers R**, de Silva R, Litvan I, Riley DE, van Swieten JC, Heutink P, Wszolek ZK, Uitti RJ, Vandrovicova J, Hurtig HI, Gross RG, Maetzler W, Goldwurm S, Tolosa E, Borroni B, Pastor P, PSP Genetics Study Group, Cantwell LB, Han MR, Dillman A, van der Brug MP, Gibbs JR, Cookson MR, Hernandez DG, Singleton AB, Farrer MJ, Yu CE, Golbe LI, Revesz T, Hardy J, Lees AJ, Devlin B, Hakonarson H, Muller U, Schellenberg GD. Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. *Nat Genet*. 2011 Jul; 43(7):699-705. Epub 2011 Jun 19. PMID:21685912. PMCID:3125476. NIHMS:297077. DOI:10.1038/ng.859.
85. Mackenzie IR, Ansorge O, Strong M, Bilbao J, Zinman L, Ang LC, Baker M, Stewart H, Eisen A, **Rademakers R**, Neumann M. Pathological heterogeneity in amyotrophic lateral sclerosis with FUS mutations: two distinct patterns correlating with disease severity and mutation. *Acta Neuropathol*. 2011 Jul; 122(1):87-98. Epub 2011 May 21. PMID:21604077. DOI:10.1007/s00401-011-0838-7.
86. Whitwell JL, Weigand SD, Gunter JL, Boeve BF, **Rademakers R**, Baker M, Knopman DS, Wszolek ZK, Petersen RC, Jack CR Jr, Josephs KA. Trajectories of brain and hippocampal atrophy in FTD with mutations in MAPT or GRN. *Neurology*. 2011 Jul 26; 77(4):393-8. Epub 2011 Jul 13. PMID:21753165. PMCID:3140800. DOI:10.1212/WNL.0b013e318227047f.
87. Ross OA, Rutherford NJ, Baker M, Soto-Ortolaza AI, Carrasquillo MM, DeJesus-Hernandez M, Adamson J, Li M, Volkening K, Finger E, Seeley WW, Hatanpaa KJ, Lomen-Hoerth C, Kertesz A, Bigio EH, Lippa C, Woodruff BK, Knopman DS, White CL 3rd, Van Gerpen JA, Meschia JF, Mackenzie IR, Boylan K, Boeve BF, Miller BL, Strong MJ, Uitti RJ, Younkin SG, Graff-Radford NR, Petersen RC, Wszolek ZK, Dickson DW, **Rademakers R**. Ataxin-2 repeat-length variation and neurodegeneration. *Hum Mol Genet*. 2011 Aug 15; 20(16):3207-12. Epub 2011 May 24. PMID:21610160. PMCID:3140823. DOI:10.1093/hmg/ddr227.
88. Whitwell JL, Josephs KA, Avula R, Tosakulwong N, Weigand SD, Senjem ML, Vemuri P, Jones DT, Gunter JL, Baker M, Wszolek ZK, Knopman DS, **Rademakers R**, Petersen RC, Boeve BF, Jack CR Jr. Altered functional connectivity in asymptomatic MAPT subjects: a comparison to bvFTD. *Neurology*. 2011 Aug 30; 77(9):866-74. Epub 2011 Aug 17. PMID:21849646. PMCID:3162637. DOI:10.1212/WNL.0b013e31822c61f2.

89. Kumar N, Boeve BF, Boot BP, Orr CF, Duffy J, Woodruff BK, Nair AK, Ellison J, Kuntz K, Kantarci K, Jack CR Jr, Westmoreland BF, Fields JA, Baker M, **Rademakers** R, Parisi JE, Dickson DW. Clinical characterization of a kindred with a novel 12-octapeptide repeat insertion in the prion protein gene. *Arch Neurol*. 2011 Sep; 68(9):1165-70. PMID:21911696. DOI:10.1001/archneurol.2011.187.
90. Neumann M, Bentmann E, Dormann D, Jawaid A, DeJesus-Hernandez M, Ansorge O, Roeber S, Kretzschmar HA, Munoz DG, Kusaka H, Yokota O, Ang LC, Bilbao J, **Rademakers** R, Haass C, Mackenzie IR. FET proteins TAF15 and EWS are selective markers that distinguish FTLD with FUS pathology from amyotrophic lateral sclerosis with FUS mutations. *Brain*. 2011 Sep; 134(Pt 9):2595-609. Epub 2011 Aug 19. PMID:21856723. PMCID:3170539. DOI:10.1093/brain/awr201.
91. DeJesus-Hernandez M, Desaro P, Johnston A, Ross OA, Wszolek ZK, Ertekin-Taner N, Graff-Radford NR, **Rademakers** R, Boylan K. Novel p.Ile151Val mutation in VCP in a patient of African American descent with sporadic ALS. *Neurology*. 2011 Sep 13; 77(11):1102-3. Epub 2011 Aug 31. PMID:21880997. PMCID:3174069. DOI:10.1212/WNL.0b013e31822e563c.
92. Pao WC, Dickson DW, Crook JE, Finch NA, **Rademakers** R, Graff-Radford NR. Hippocampal sclerosis in the elderly: genetic and pathologic findings, some mimicking Alzheimer disease clinically. *Alzheimer Dis Assoc Disord*. 2011 Oct; 25(4):364-8. PMID:21346515. PMCID:3107353. NIHMS:272500. DOI:10.1097/WAD.0b013e31820f8f50.
93. DeJesus-Hernandez M, Mackenzie IR, Boeve BF, Boxer AL, Baker M, Rutherford NJ, Nicholson AM, Finch NA, Flynn H, Adamson J, Kouri N, Wojtas A, Sengdy P, Hsiung GY, Karydas A, Seeley WW, Josephs KA, Coppola G, Geschwind DH, Wszolek ZK, Feldman H, Knopman DS, Petersen RC, Miller BL, Dickson DW, Boylan KB, Graff-Radford NR, **Rademakers** R. Expanded GGGGCC hexanucleotide repeat in noncoding region of C9ORF72 causes chromosome 9p-linked FTD and ALS. *Neuron*. 2011 Oct 20; 72(2):245-56. Epub 2011 Sep 21. PMID:21944778. PMCID:3202986. NIHMS:327774. DOI:10.1016/j.neuron.2011.09.011.
94. Kouri N, Murray ME, Hassan A, **Rademakers** R, Uitti RJ, Boeve BF, Graff-Radford NR, Wszolek ZK, Litvan I, Josephs KA, Dickson DW. Neuropathological features of corticobasal degeneration presenting as corticobasal syndrome or Richardson syndrome. *Brain*. 2011 Nov; 134(Pt 11):3264-75. Epub 2011 Sep 20. PMID:21933807. PMCID:3212714. DOI:10.1093/brain/awr234.
95. Nicholson AM, Finch NA, **Rademakers** R. Human genetics as a tool to identify progranulin regulators. *J Mol Neurosci*. 2011 Nov; 45(3):532-7. Epub 2011 May 28. PMID:21626010. DOI:10.1007/s12031-011-9554-y.
96. Murray ME, DeJesus-Hernandez M, Rutherford NJ, Baker M, Duara R, Graff-Radford NR, Wszolek ZK, Ferman TJ, Josephs KA, Boylan KB, **Rademakers** R, Dickson DW. Clinical and neuropathologic heterogeneity of c9FTD/ALS associated with hexanucleotide repeat expansion in C9ORF72. *Acta Neuropathol*. 2011 Dec; 122(6):673-90. Epub 2011 Nov 15. PMID:22083254. DOI:10.1007/s00401-011-0907-y.
97. Couthouis J, Hart MP, Shorter J, DeJesus-Hernandez M, Erion R, Oristano R, Liu AX, Ramos D, Jethava N, Hosangadi D, Epstein J, Chiang A, Diaz Z, Nakaya T, Ibrahim F, Kim HJ, Solski JA, Williams KL, Mojsilovic-Petrovic J, Ingre C, Boylan K, Graff-Radford NR, Dickson DW, Clay-Falcone D, Elman L, McCluskey L, Greene R, Kalb RG, Lee VMY, Trojanowski JQ, Ludolph A, Robberecht W, Andersen PM, Nicholson GA, Blair IP, King OD, Bonini NM, Van Deerlin V, **Rademakers** R, Mourelatos Z, Gitler AD. A yeast functional screen predicts new candidate ALS disease genes. *Proceedings Of The National Academy Of Sciences Of The United States Of*. 2011 Dec 27; 108(52):20881-90.

98. Mok K, Traynor BJ, Schymick J, Tienari PJ, Laaksovirta H, Peuralinna T, Myllykangas L, Chio A, Shatunov A, Boeve BF, Boxer AL, DeJesus-Hernandez M, Mackenzie IR, Waite A, Williams N, Morris HR, Simon-Sanchez J, van Swieten JC, Heutink P, Restagno G, Mora G, Morrison KE, Shaw PJ, Rollinson PS, Al-Chalabi A, **Rademakers R**, Pickering-Brown S, Orrell RW, Nalls MA, Hardy J. The chromosome 9 ALS and FTD locus is probably derived from a single founder. *Neurobiol Aging*. 2012; 33(1):209.e3-8.
99. Nicholson AM, Gass J, Petrucelli L, **Rademakers R**. Progranulin axis and recent developments in frontotemporal lobar degeneration. *Alzheimers Res Ther*. 2012 Jan 23; 4(1):4. [Epub ahead of print] PMID:22277331. DOI:10.1186/alzrt102.
100. **Rademakers R**, Baker M, Nicholson AM, Rutherford NJ, Finch N, Soto-Ortolaza A, Lash J, Wider C, Wojtas A, DeJesus-Hernandez M, Adamson J, Kouri N, Sundal C, Shuster EA, Aasly J, MacKenzie J, Roeber S, Kretzschmar HA, Boeve BF, Knopman DS, Petersen RC, Cairns NJ, Ghetti B, Spina S, Garbern J, Tselis AC, Uitti R, Das P, Van Gerpen JA, Meschia JF, Levy S, Broderick DF, Graff-Radford N, Ross OA, Miller BB, Swerdlow RH, Dickson DW, Wszolek ZK. Mutations in the colony stimulating factor 1 receptor (csf1r) gene cause hereditary diffuse leukoencephalopathy with spheroids. *Nat Genet*. 2012 Feb; 44(2):200-5.
101. Rutherford NJ, Finch NA, DeJesus-Hernandez M, Crook RJ, Lomen-Hoerth C, Wszolek ZK, Uitti RJ, Graff-Radford NR, **Rademakers R**. Pathogenicity of exonic indels in fused in sarcoma in amyotrophic lateral sclerosis. *Neurobiol Aging*. 2012 Feb; 33(2):424.e23-4. Epub 2010 Nov 12. PMID:21074900. PMCID:3130814. NIHMS:254648. DOI:10.1016/j.neurobiolaging.2010.09.029.
102. Boeve BF, Boylan KB, Graff-Radford NR, DeJesus-Hernandez M, Knopman DS, Pedraza O, Vemuri P, Jones D, Lowe V, Murray ME, Dickson DW, Josephs KA, Rush BK, Machulda MM, Fields JA, Ferman TJ, Baker M, Rutherford NJ, Adamson J, Wszolek ZK, Adeli A, Savica R, Boot B, Kuntz KM, Gavrilova R, Reeves A, Whitwell J, Kantarci K, Jack CR Jr, Parisi JE, Lucas JA, Petersen RC, **Rademakers R**. Characterization of frontotemporal dementia and/or amyotrophic lateral sclerosis associated with the GGGGCC repeat expansion in C9ORF72. *Brain*. 2012 Mar; 135(Pt 3):765-83. PMID:22366793. PMCID:3286335. DOI:10.1093/brain/aws004.
103. Whitwell JL, Weigand SD, Boeve BF, Senjem ML, Gunter JL, DeJesus-Hernandez M, Rutherford NJ, Baker M, Knopman DS, Wszolek ZK, Parisi JE, Dickson DW, Petersen RC, **Rademakers R**, Jack CR Jr, Josephs KA. Neuroimaging signatures of frontotemporal dementia genetics: C9ORF72, tau, progranulin and sporadics. *Brain*. 2012 Mar; 135(Pt 3):794-806. PMID:22366795. PMCID:3286334. DOI:10.1093/brain/aws001.
104. Khan BK, Yokoyama JS, Takada LT, Sha SJ, Rutherford NJ, Fong JC, Karydas AM, Wu T, Kettle RS, Baker MC, Hernandez MD, Coppola G, Geschwind DH, **Rademakers R**, Lee SE, Rosen HJ, Rabinovici GD, Seeley WW, Rankin KP, Boxer AL, Miller BL. Atypical, slowly progressive behavioural variant frontotemporal dementia associated with C9ORF72 hexanucleotide expansion. *J Neurol Neurosurg Psychiatry*. 2012 Apr; 83(4):358-64. PMID:22399793
105. Hsiung GY, DeJesus-Hernandez M, Feldman HH, Sengdy P, Bouchard-Kerr P, Dwosh E, Butler R, Leung B, Fok A, Rutherford NJ, Baker M, **Rademakers R**, Mackenzie IR. Clinical and pathological features of familial frontotemporal dementia caused by C9ORF72 mutation on chromosome 9p. *Brain*. 2012 Mar; 135(Pt 3):709-22. Epub 2012 Feb 17. PMID: 22344582. PMCID: PMC3286328
106. Sundal C, Lash J, Aasly J, Oygarden S, Roeber S, Kretzschman H, Garbern JY, Tselis A, **Rademakers R**, Dickson DW, Broderick D, Wszolek ZK. Hereditary diffuse leukoencephalopathy with axonal spheroids (HDLS): A misdiagnosed disease entity. *J Neurol Sci*. 2012 Mar 15; 314(1-2):130-7. Epub 2011 Nov 01. PMID:22050953. PMCID:3275663. NIHMS:331541. DOI:10.1016/j.jns.2011.10.006.

107. Wider C, Ross OA, Nishioka K, Heckman MG, Vilarino-Guell C, Jasinska-Myga B, Erketin-Taner N, **Rademakers** R, Graff-Radford NR, Mash DC, Papapetropoulos S, Duara R, Uchikado H, Wszolek ZK, Farrer MJ, Dickson DW. An evaluation of the impact of MAPT, SNCA and APOE on the burden of Alzheimer's and Lewy body pathology. *J Neurol Neurosurg Psychiatry*. 2012 Apr; 83(4):424-9. Epub 2012 Jan 30. PMID:22291217. DOI:10.1136/jnnp-2011-301413.
108. **Rademakers** R. C9orf72 repeat expansions in patients with ALS and FTD. *Lancet Neurol*. 2012 Mar 09. [Epub ahead of print] PMID:22406229. DOI:10.1016/S1474-4422(12)70046-7.