

Carlos Cruchaga PhD

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I am a tenured Professor with affiliations in the Departments of Psychiatry and Neurology at Washington University School of Medicine. I have significant administrative experience both inside and outside of Washington University and a track record of successful leadership in these roles. In addition, I have an exceptional track record of extramural funding, high-quality scholarship, and graduate, postdoc and junior faculty mentoring. My long-term goal is to maintain an active high quality research program while effectively leading and mentoring postdocs and faculties.

Current Position

Professor	2019-Present	Dept. of Psychiatry
Professor	2019-Present	Dept. of Neurology
Scientific Director	2019-Present	McDonnell Genome Institute
Director	2018-Present	NeuroGenomics and Informatics
Director	2015-Present	Hope Center DNA and RNA extraction core
Core Leader	2015-Present	Knight-ADRC and DIAN Genetics Core

Administrative Experience

2019-Present **Scientific Director McDonnell Genome Institute.**

Identify and implement the research priorities for the long-term success of the Institute.
Creation of a new university-wide biospecimen bank. Identifying promising young faculties and manage budget allocations.

2018-Present **Director NeuroGenomics and Informatics.**

Manage budget allocations, annual stewardship interviews, outline the strategic research plan for the center, manage personnel hiring and needs for the different research units and projects within the center, design and implement carrier development plans for young faculties, government approvals for human studies, multidisciplinary research and clinical program, tissue and data collection, analysis, and publications.

2018 **Special Committee for the McDonnell Executive director search.**

Spent more than 200 hours working with a three-member committee and Washington University to identify the next McDonnell executive director.

2017-Present **Hope Center Hope Center Steering Committee member.**

Managed center review document preparations, develop research priorities that fit the goals of the center, and budget allocation decision making.

2015-Present **DNA and RNA extraction Core Director.**

Promotion of the core within the institution and outside of the institution, set up and management budgets, acquisition of instruments, annual stewardship interviews, hiring and managing staff for the optimal operation of the core.

2016-Present **Organizing Committee Member. Hope Center Annual retreat**

Worked with committee to select venue and session topics, invited speakers, managed selection of abstract submissions, hired and managed staff to oversee conference logistics.

2014-2017 **Committee Member**, Alzheimer's Association Intl. Conference Scientific Program
Select session topics, invite and manage dozens of reviewers for hundreds of abstracts, make final decisions on presenters, chair conference sessions.

Research Positions

1998 - 2000 **Student Assistant**, University of Navarra School of Medicine, Department of Biochemistry and Molecular Biology, Pamplona, Spain.
2000 - 2005 **Teaching Assistant**, University of Navarra School of Medicine, Department of Biochemistry and Molecular Biology, Pamplona, Spain.
2005 - 2007 **Postdoctoral Fellow**, University of Navarra, Center for Applied Medical Research (CIMA), Division of Neurosciences, Pamplona, Spain.
2007 - 2010 **Postdoctoral Fellow**, Psychiatry Department, Washington University
2010 - 2015 **Assistant Professor**, Psychiatry Department, Washington University
2010-Present **Division of Biology and Biomedical Sciences Faculty**. Programs of Neurosciences and "Human and Statistical Genetics". Washington University
2010-Present **Hope Center Member**, Washington University School of Medicine
2010-Present **Hope Center on Protein Aggregation and Neurodegeneration Investigator**, Washington University School of Medicine
2014-Present **Neuroscience Advisory Committee**. DBBS Washington University
2015-2019 **Associate Professor**, Psychiatry Department, Washington University
2015-Present Knight-ADRC Genetics **Core leader**. Washington University
2015-Present DIAN (The Dominantly Inherited Alzheimer Network) **Genetics Core leader**
2015-Present **Associate Professor**, Neurology Department, Washington University
2015-Present Hope Center DNA and RNA **Core Director**
2018-Present **Director** NeuroGenomics and Informatics (NGI)
2019-Present **Scientific Director** McDonnell Genome Institute (MGI) - Washington University
2019-Present **Professor**, Psychiatry Department, Washington University

Education:

1996-2000 Degree in Biochemistry, University of Navarra, Pamplona, Spain
2000-2002 MA. University of Navarra, Pamplona, Spain
2000-2005 PhD. in Biochemistry and Molecular Biology.
University of Navarra, Pamplona, Spain
Grade: Summa Cum Laude.
(Thesis Supervisor: Professor Juan J. Martinez-Irujo)
2005-2007 Postdoctoral training in Statistical Human Genetics. Center for Applied Medical Research, Spain
2007-2011 Postdoctoral training in Quantitative Human Genomics. Washington University in St. Louis

Honors and Awards:

2005 Selected to take part in the "Excellence Campus 2005". The best 18 Spanish PhD student show their works in presence of 4 Nobel prize winners and other important members of the scientific community. Selection made by the "Department of Education and Science, Spanish Government".
2007 2007 CurePSP+ Annual International Research Symposium Travel Fellowship
2008 International Conference on Alzheimer's Disease Travel Fellowship
2009 Finalist in "The James L. O'Leary Prize". Neuroscience research. Washington University in Saint Louis. Work presented: "SNPs in the regulatory subunit of

calcineurin are associated with CSF phospho-tau protein levels, and risk for Alzheimer's Disease".

- 2010 International Conference on Alzheimer's Disease Travel Fellowship.
- 2012 Selected for NIH Early Career Reviewer Program
- 2013 Alzheimer's Association International Conference Fellowship
- 2014-2017 Member of the Alzheimer's Association International Conference (AAIC) Scientific Program Committee (SPC).
- 2014-Present Hope Center Annual Retreat Organizing committee.
- 2016 Member of the Alzheimer's Association International Conference (AAIC) Scientific Program Committee (SPC). Head of the Basic and Translational Science program committee.
- 2017 Hope Center Hope Center Steering Committee member.

Fellowships:

- 2000 - 2004 Predoctoral Fellowship of the Education Department Navarre Government. (Departamento de Educación del Gobierno de Navarra).
- 2005 - 2005 Predoctoral Fellowship of "Asociación de amigos de la Universidad de Navarra". Department of Biochemistry and Molecular Biology, Universidad de Navarra (Pamplona, Spain).
- 2005 - 2006 Postdoctoral Fellowship of Fundation for Applied Medical Research. Center for Applied Medical Research (CIMA) (Pamplona, Spain).
- 2006 - 2007 Postdoctoral Fellowship of the Education Department Navarre Government "Departamento de Educación del Gobierno de Navarra".
- 2008 - 2010 Postdoctoral Fellowship of "Fundacion Alfonso Martin Escudero" (<http://www.fundame.org/>).

Consulting Relationships and Board Memberships:

- 2014-2107 Member of the Alzheimer's Association International Conference (AAIC) Scientific Program Committee (SPC).
- 2018-Present ADx Healthcare. Member of the Advisory Board
- 2018-Present Vivid Genomics. Member of the Advisory Board

Editorial Responsibilities:

Ad Hoc Journal Review: Acta Neuropathologica, American Journal Human Genetics, Alzheimer and Dementia, Alzheimer Research and therapy, American Journal of Medical Genetics Part B, Annals of Neurology, Annals of Clinical and Translational Neurology, Archives of Neurology, Biological Psychiatry, BMC Neurology, Dementia and Geriatric Cognitive Disorders, Expert Review, Genes Brain and Behavior, Future Neurology, Genes, Brain and Behavior, Human Molecular Genetics, Human Genome Variation, Human Mutation, JAMA, JAMA Neurology, Journal of Alzheimer Disease, Journal of Geriatric Psychiatry and Neurology, Journal of Neurochemistry, Journal of Neurological Sciences, Journal of Neuroscience, Metabolic Brain Diseases, Molecular Genetics and Genomics, Molecular Psychiatry, Molecular Neurodegeneration, Molecular Neurology, Movement Disorder, Nature, Nature Biotechnology, Nature Communications, Nature Medicine, Nature Neuroscience, Neurobiology of Aging, Neurology, NeuroMolecular Medicine, Neuron, Neuroscience Letters, Ophthalmic Research, PLoS Genetics, PLoS One, Rejuvenation Research, Scientific Reports, The International Journal of Neuroscience, The world Journal of Biological Psychiatry, Translation Psychiatry.

Ad Hoc Grant Review: Alzheimer Association UK, Alzheimer Association USA, Alzheimer Association Nederland, ANECA (Spanish Science Institute), BRACE - Rosetrees Phd Studentship

(UK), BrightFocus Foundation, Foundation for Alzheimer Research, Research Parkinsons UK, Geoffrey Beene Foundation, NIH, National Research Council Romanian Government, National Science Center, Poland, Medical Research Council (MRC) UK, Knight-ADRC, SOA-FRA (Stichting Alzheimer Onderzoek-Fondation Recherche Alzheimer). DOD (department of defences) 2016: AD and TBI. 2017: PD and tau pathology

Editorial board: Associated Editor BMC Neurology, Associated Board Editor Acta Neuropathologica. Associate Editor Frontiers in Genetics / Neurogenomics

NIH-Grant reviewer: Study section: GHD (Genetics of Health and Disease Study Section): June 2013, October 2014. ZRG1 MDCN-Q(03) Special Emphasis Panel: March 2015. GHD Permanent Member: June 2015

Professional Societies and Organizations:

Memberships:

2007- Present: American Society of Human Genetics

2007-Present Alzheimer's Association International Society to Advance Alzheimer Research and Treatment (ISTAART)

2013-Present The Gerontological Society of America

2015-Present American Heart Association

Major Invited Professorships and Lectureships:

Invited Seminars

1. **Psychiatry Department Seminar.** Washington University School of Medicine, St. Louis, MO 2007. Use of cerebrospinal fluid tau levels as endophenotype for late-onset Alzheimer's disease.
2. **Neurodegeneration interest group.** Washington University, St. Louis, MO 2009. SNPs in the regulatory subunit of calcineurin are associated with CSF phospho-tau protein levels, and risk for Alzheimer's Disease.
3. **O'Leary Award for Research in Neuroscience Finalists Seminar.** Washington University, St. Louis, MO 2009. SNPs in the regulatory subunit of calcineurin are associated with CSF phospho-tau protein levels, and risk for Alzheimer's Disease.
4. **Neuroscience lectures.** Center for Applied Medical Research, Pamplona, Spain 2009. SNPs associated with CSF tau levels modify rate of progression in Alzheimer's Disease.
5. **Neurology Department Seminars.** University of the Basque Country, San Sebastian, Spain 2010. Identification of rare variants implicated in Alzheimer's Disease by Next-generation sequencing technology.
6. **AD Risk & Protective Factors and Early Interventions.** Southern Illinois University School of Medicine. Springfield, IL 2010. Advances in the genetic studies of Alzheimer's disease.
7. **Alzheimer's Disease Research Center seminar.** Washington University, St. Louis, MO 2010. Advances in the genetic studies of Alzheimer's disease.
8. **The HOPE Center for Neurological Disorders at Washington University.** Washington University, St. Louis, MO **2010.** Pathogenic Mutations in APP, PSEN1, PSEN2, GRN and MAPT in Families with Late Onset Alzheimer Disease.
9. **Alzheimer's Disease Research Center seminar.** Washington University, St. Louis, MO. March 2012. CSF biomarkers as quantitative endophenotypes for genetics studies in AD.

10. **Alzheimer's Disease Research Center seminar.** Washington University, St. Louis, MO. August 2012. 2012 Alzheimer's Association International Conference Update.
11. **Neuroscience seminar.** King's College London. Alternative approaches to identify novel genes and variants implicated on AD. April 2013
12. **Invited neurogenetics seminar.** UCL London. Identifying rare coding variants associated with AD. April 2013
13. **Neuroscience lectures.** Center for Applied Medical Research, Pamplona. Alternative approaches to identify novel genes and variants implicated on AD. April 2013
14. **Alzheimer's Disease Research Center seminar.** Washington University, St. Louis, MO. July 2013. 2013 Alzheimer's Association International Conference Update.
15. **ADC (Alzheimer's Disease Center) Directors meeting.** New Orleans. October 2013. Identifying functional genetics variants associated with Alzheimer's disease.
16. **NIAC Seminar Series.** Washington University. St. Louis. January 2014. Next-generation sequencing technology to identify novel genes implicated on Alzheimer's Disease
17. **Invited Neuroscience Seminar at Mount Sinai.** April 2014: Identification of low-frequency and rare coding variants implicated on Alzheimer's and Parkinson Disease
18. **Alzforum Webinar Speaker. July 2014:** Mutations Impair TREM2 Maturation, Processing, and Microglial Phagocytosis. <http://www.alzforum.org/webinars/mutations-impair-trem2-maturation-processing-and-microglial-phagocytosis>
19. **Invited Neuroscience Seminar at UC Irvine.** Octubre 2014: Genetic Risk for Alzheimer's disease: An emerging role for rare variants
20. **Invited Neuroscience Seminar at Mayo Clinic (Jacksonville).** Octubre 2014: Genetic Risk for Alzheimer's disease: An emerging role for rare variants.
21. **Psychiatry Wednesday Research Seminars.** March 2017. Genomic Approaches to Understand the Genetic Architecture of Neurodegenerative Processes
22. **Biogen. Neuroscience (Boston).** February 2018. Genomics approaches to understand the biology of Alzheimer disease and create better prediction models.
23. **Alzheimer's Disease Research Center seminar.** February 2018. Polygenic Risk score and prediction models for Alzheimer Disease.
24. **McDonnell Genome Institute Symposium February 2018.** Genomics approaches to understand the biology of Alzheimer disease and create better prediction models
25. **National Health Insurance Service Ilsan Hospital. Korea.** April 2018. Genetics of Alzheimer's Disease: An emerging role for rare variants
26. **EISAI. Neuroscience (Boston).** May 2018. Genomics approaches to understand the biology of Alzheimer disease and create better prediction models.
27. **Hope Center Neurogenetics and transcriptomics research group. Washinton University** Oct 2018. The role of MS4A4A in TREM2 biology
28. **Inspired by Ramón y Cajal. MIT (Boston).** Oct 2018. Alzheimer's Disease Genetics: from genetic variants to biological pathways
29. **Biogen. Neurogenetics (Boston). Oct 2018.** The role of MS4A4A in TREM2 biology

Meeting Talks

1. **Alzheimer's Association International Conference on Alzheimer's Disease (ICAD) 2010.** Impact of APP, PSEN1, PSEN2, GRN and MAPT mutations in a well-characterized series of late-onset AD families.
2. **Alzheimer's Association International Conference on Alzheimer's Disease (ICAD) 2011.** Association and Expression analyses with SNPs in TOMM40 in Alzheimer's Disease
3. **Alzheimer's Association International Conference on Alzheimer's Disease (AAIC) 2012.** Genome-wide association studies using cerebrospinal fluid biomarkers as an endophenotype for Alzheimer's disease
4. **Alzheimer's Association International Conference on Alzheimer's Disease (AAIC) 2013.** Rare coding variants in Phospholipase D3 (PLD3) confer risk for Alzheimer's disease
5. **Organizer for the Featured Research Session (FRS) at AAIC 2013:** Rare variants in Alzheimer's Disease.
6. **New Investigators in Alzheimer's Disease Grantee Meeting. June 2014. CA.** Identification of additional risk variants and functional characterization of TREM2 and PLD3
7. **Alzheimer's Association International Conference on Alzheimer's Disease (AAIC) 2014:** Exome-sequencing in late-onset families identified additional candidate genes for Alzheimer's disease
8. **Genetics: Next Generation Sequencing in Dementia (O1-04),** Alzheimer's Association International Conference on Alzheimer's Disease (AAIC) 2014: Session Chair
9. **AGBT meeting (February 2017).** Novel rare variants in known genes, a look into the Familiar Alzheimer Sequencing (FASe) project
10. **AGBT meeting (February 2018).** The Omnigenic model of Alzheimer disease
11. **The International Stroke Genetics Consortium (ISGC).** Genetic Architecture Overlap of Stroke Risk, Early Outcomes and Cardiovascular Risk Factors. Japan April 2018
12. **The Genetics of Early Neurological Instability after Ischemic Stroke (GENISIS) meeting.** New advances in transcriptome studies: from tissue to cell. Sevilla. July 2018
13. **Alzheimer's Association International Conference on Alzheimer's Disease (AAIC) July 2018.** Review of GWAS Genes and Polygenic Risk Score

Research Support:

Ongoing Research Support

a. Governmental:

5 R01 AG044546-05 (Cruchaga)

8/15/13 – 5/31/18

NIA

\$295,655

Identifying Rare Variants that Increase Risk for Alzheimer's Disease

The aim of this research is to identify rare functional variants with large effect size on risk for Alzheimer's disease

1RF1 AG053303-01 (Cruchaga)

9/15/16 – 6/30/21

NIA

\$2,102,945

Using Quantitative Traits to Identify Novel Genes for Alzheimer's Disease and Other Complex Traits

This project focuses on understudied aspects of disease etiology namely the role of common and rare genetic variation on quantitative diagnostic and prognostic endophenotypes of Alzheimer's disease (AD)

5 R01 NS085419-03 (Lee; Cruchaga; Montaner)	4/1/14 – 3/31/19
NINDS	\$398,113
Genetic Architecture of Acute Human Brain Ischemia	
This study will identify genetic variants associated with early neurological outcome in acute ischemic stroke (AIS) patients (untreated or treated with IV tPA)	
1U01 AG052411-01 (Goate; Cruchaga)	7/15/16 – 5/31/21
NIA	\$122,966
Identification and Characterization of AD Risk Pathways using Multi-Dimensional “OMICS” Data	
Beyond refining AD genetic architecture the goal of this study is to identify and validate therapeutic targets for AD identifying genes that functionally drive or protect from AD and interrogating their respective gene networks for therapeutic targets	
5P01 AG003991-33 (Morris; Cruchaga PI Project 3)	1/1/97 – 6/30/19
NIA	\$1,224,236
Healthy Aging and Senile Dementia	
The overarching aim of this program project is to determine the indicators that characterize the progression from cognitive normality to the earliest stages of cognitive impairment caused by Alzheimer disease (AD)	
2P50 AG005681-32 (Morris; Cruchaga PI Genetics Core)	6/15/97- 4/30/20
NIA	\$1,754,838
Alzheimer’s Disease Research Center	
The Knight Alzheimer Disease Research Center will coordinate and support integrated cutting-edge transdisciplinary research initiatives to examine the mechanisms that underlie the transition from preclinical to symptomatic AD and that enhance the understand of the pathobiological disorder	
1U24 AG056270-01 (Mayeux; Cruchaga PI Subc Columbia U)	8/1/17 – 5/31/22
NIA	\$60,000
The National Institute on Aging (NIA) Late Onset of Alzheimer’s Disease (LOAD) Family Based Study (FBS)	
The goal of this project is to validate and quantify the clinical impact of these newly identified genetic risk facts using the multiplex families recruited through the NIA-LOAD	
5R01 NS092865-02 (Xu)	2/1/16 – 11/30/20
NINDS	\$196,875
Postmortem Validation of Biomarkers for Imaging Parkinson Disease	
This study will leverage human brain tissues collected previously to determine the relationship between postsynaptic dopaminergic markers and related transmitter systems and the loss of nigrostriatal neurons	
2 U19 AG032438-07 (Bateman)	9/15/08 – 3/31/19
NIA	\$2,876,685
Dominantly Inherited Alzheimer Network	
This renewal continues to address the three original DIAN hypotheses with increased emphasis on longitudinal data and maintain current aims, maintenance of the established international DIAN registry of individuals with attention to preparing and adjusting for participants who participate in current and planned prevention trials. New scientific studies are planned, many funded independently of the DIAN grant and conducted within the DIAN infrastructure at no cost to DIAN	

5R01 AR067715-02 (Gurnett, Dobbs) 6/20/15 – 5/31/20
NIAMSD \$402,784
Genetic Risk Factors for Severe Scoliosis
The goal of this project is to identify genetic risk factors for severe adolescent idiopathic scoliosis (AIS)

1 R01 AG053267-01 (Bateman) 9/1/17-5/31/22 0.6 calendar
NIA \$3,393,066
DIAN-TU: Next Generation Prevention Trial
The DIAN-TU Next Generation Prevention Trial grant will add a new drug arm to the DIAN-TU trial platform and implement key design changes that allow the platform to adaptively test next generation drugs with diverse mechanisms of action, dose escalations to maximize efficacy and a disease progression model to more rapidly establish clinical benefit in an asymptomatic population.

U01AG058922 (Cruchaga) Score: 38. Not Percentile. (Special RFA)
NIA \$ 3,535,211
The Familial Alzheimer Sequencing (FASe) Project
The aim of this research is to identify rare functional variants with large effect size on risk for Alzheimer's disease

RF1AG058501 (Cruchaga) Score: 40. Percentile: 30. Funding line 28
NIA \$ 3,791,754
Genetic Modifiers of Cerebrospinal Fluid TREM2 In Alzheimer's Disease

1 R01 AG057777-01A1 (Harari) 4/1/18-3/31/23 0.6 calendar
NIA \$495,221
Mendelian Randomization for Unbiased Biomarker Discovery for AD and other Complex Traits
This project aims to reveal novel intermediate traits and genetic variants involved in Alzheimer's Disease (AD) and other complex traits. This will lead to therapeutic targets and provide guidelines for accurate and practical assessment of AD status.

b. Non-governmental:

(Cruchaga, Harari, Karch)
The Chan Zuckerberg Initiative: CZI Neurodegeneration Challenge \$1,050,000
The goal of this project is to elucidate the mechanisms by which common variants near *MS4A* alter TREM2 function and drive neurodegenerative disease. We hypothesize that *MS4A* regulates intracellular trafficking of TREM2 and that disrupted interaction between sTREM2 and *MS4A* impair signaling, resulting in microglial dysfunction and neurodegeneration.

(Cruchaga) 10/1/15 – 9/30/18
Alzheimer's Association \$150,000
Understanding the Role of APP, PSEN1, PSEN2, TREM2 and PLD3 in AD
This study will promote further understanding of the underlying biological mechanism of genetic risk factors associated with Alzheimer's disease and related dementia

Bateman (program director) 8/1/15 – 7/31/18
GHR Foundation & Anonymous Foundation \$4,500,000
The Dominantly Inherited Alzheimer's Network Trials Unit (DIAN-TU);
Proposal for Sustainable Funding
The goals of the DIAN-TU sustainable funding grants are to 1) manage the DIAN expanded registry for identification of additional participants, researchers and site; 2) review potential compounds to

test in DIAN-TU trials; 3) design the next generation of DIAN-TU trials with expertise and input from pharma, regulators, researchers, consultants and DIAN/DIAN-TU participants and families; 4) manage DIAN-TU scientific proposals to aid trial design; 5) host DIAN pharma consortium meetings and 6) perform exome sequencing of families negative for current known ADAD mutations.

(Cruchaga) 09/1/18 – 9/30/19
Alector \$1,027,925
Proteomic signature of Alzheimer' disease risk variant carriers and disease progression

(Cruchaga) 09/1/18 – 9/30/19
Biogen \$338,184
Identifying genetic variants associated with stroke outcomes

COMPLETED

(Cruchaga PI) 7/7/16-7/6/17
Biogen
Identifying Genetic Variants Associated with Amyloid Biomarkers
This collaborative research will identify genetic variants associated with Amyloid biomarkers.

(Cruchaga PI) 10/1/14-9/30/16
Michael J. Fox Foundation for Parkinson's Research
Using Genetics, Biomarker and Mendelian Randomization to Identify Common Pathways and Genes Implicated on Alzheimer's and Parkinson's Disease
This project will identify informative biomarkers for AD and Parkinson's disease, and will identify genetic variants and pathways in common between PD and AD.

(Cruchaga PI) 10/1/14-9/30/16
Hope Center. Washington University
Identification and functional characterization of rare variants in DNAJC5/CSP gene associated with Alzheimer Disease
Hope Center Pilot Grant: We recently found that the DNAJC5 gene (DnaJ (Hsp40) homolog, subfamily C, member 5) is a Mendelian dementia-causing gene of an early-onset and rare form of dementia. In this project, our aim is to identify rare genetic variants in DNAJC5, and to determine whether those rare variants play a role in the risk for AD.

(Cruchaga PI) 7/1/13-6/30/15
American Federation for Aging Research, Inc.
Identification of Additional Risk Variants and Functional Characterization of TREM2 and PLD3
This study will identify pathogenic mutations in APP, PSEN1 and PSEN2 and the association of APOE genotype with risk which will lead to a better understanding of the pathobiology of AD, the development of novel animal models and therapies for AD.

(Cruchaga PI) 7/1/13-6/30/16
BrightFocus Foundation
Identification of Novel CSF Endophenotypes and Genetic Variants for AD by Mendelian Randomization
This study will focus on a significant and innovative concept for identifying novel AD endophenotypes and their potential use for genetic studies.

(Cruchaga PI) 12/1/13-4/30/15

Anonymous Foundation

Exome Sequencing in Alzheimer's Disease Families to Identify Novel Variants Implicated in Disease

The goal of this project is to use next-generation sequencing in LOAD families to identify novel genes involved on AD.

Cruchaga (PI)

1/1/2011-1/30/2013

Hope Center. Washington University

Hope Center Pilot Grant: Identification and functional characterization of the genetic variant causing Kufs' disease with early dementia in a family with an autosomal dominant inheritance.

The goal of this proposal is to identify the mutation causing the Kufs' disease in a family with an autosomal dominant pattern of inheritance.

Cruchaga (PI)

3/1/2011-3/30/2012

Washington University

ADRC Pilot Grant: Identification of genetic variants associated with cerebrospinal fluid granulin levels.

The goal of this proposal is to identify genetic variants implicated in frontotemporal dementia through the use of quantitative traits.

(Cruchaga PI)

7/1/11-6/30/13

McDonnell Center for Cellular and Molecular Neurobiology

\$40,000

Identification of Common and Rare Variants Associated with Alzheimer's Disease

The goal of this project is to identify novel variants, in the genes identified by recent genome-wide association studies by using the next-generation sequencing technology to identify the functional variants that drive the association with risk for Alzheimer's disease.

(Cruchaga PI)

10/1/11-9/30/13

Alzheimer's Association

Exome-Sequencing of Late-onset Alzheimer's Disease Families

This study will seek to identify mutations in three genes, which will have an incalculable value to increasing the understanding of the pathologic events and pathways involved in Alzheimer's disease (AD), and to, potentially, identify new therapeutic targets for AD.

(Cruchaga PI)

9/30/09-8/31/13

5 P30 NS069329-02

NINDS

Translational Neuroscientist and The Hope Center

The goal of this center is to develop better ways to diagnose and treat neurodegenerative diseases through an understanding of the underlying molecular mechanisms of disease.

Teaching Title and Responsibilities:

2009 - Present

Lecturer. Bio5285.

Fundamentals of Mammalian Genetics. Washington University

2012 - Present

Lecturer. Bio5491.

Advanced Genetics. Washington University

2018- Present

Lecturer. L41-5483.

Human Linkage and Association Analysis. Washington University

Graduate students:

- 1.- Sheng-Chih (Peter) Jin. 2011-2014. Genetic epidemiology program. Deep sequencing of genes associated with the Alzheimer's Disease. Graduation date: August 2014. Current Position: Postdoc at Yale.
- 2.- Yuetiva Deming. 2014-2017. Neurosciences. Using potential biomarkers for AD to identify common and rare variants implicated in AD. Graduation date: August 2017. Current Position: Postdoc Washington University.
- 3.- Zeran Li: 2015-present. Neurosciences.
- 4.- Umber Dube: 2015-Present. MD-PhD program.

Postdocs:

- 1.- Bruno Benitez, MD. 2011-2013. Current Position: instructor Washington University
- 2.- Jenny Lord, PhD. 2013-2014: Current Position: Postdoc at Sanger Institute
- 3.- Jorge del Aguila, PhD. 2014-Present.
- 4 - Victoria Fernandez, PhD. 2015-Present
- 5- Laura Ibanez, PhD. 2015-Present.
- 6- Yuetiva Deming Robles, PhD 2017- 2018. Current Position: Postdoc University Wisconsin

Work Study:

Zach Deutch, 9/19/11 - 12/9/11
Catherine Bi, 1/13/15 - 5/1/15
Catherine Bi, 8/31/15 - 12/15/15
Catherine Bi, 1/19/16 - 4/15/16
Catherine Bi, 9/6/16 - 12/5/16
Catherine Bi, 1/27/17 - 5/2/17
Elly Yeonsu Sim, 2/18/17 – 12/5/18

Summer:

Brooke Sadler, 5/19/10 - 8/16/10
Brooke Sadler, 5/18/11 - 8/11/11
Zach Deutch, 6/1/11 - 7/29/11
Jiyeon Choi, 6/17/13 - 8/16/13
Alexander Lu, 5/12/14 - 8/15/14
Jessica Hayes, 6/1/15 - 7/31/15
Alexander Lu, 5/12/14 - 8/15/14
Catherine Bi, 5/17/16 - 7/30/16
Marissa Turkin, 5/15/17 - 8/12/17

BP Endure:

Jessica Jimenez, 6/1/15 – 8/1/15
Joseph Bradley, 6/3/16 - 8/5/16
Joseph Bradley, 6/2/17 - 8/5/17

Bio 200/500:

Jiyeon Choi, 1/18/13 - 5/11/13.
Alexander Lu, 1/27/14 - 5/7/14
Jessica Hayes, 1/20/15 - 2/27/15
Alexander Lu, 9/1/14 - 12/1/14
Marissa Turkin, 8/29/16 - 12/2/16

Rotation:

Sheng Peter Chih Jin, 3/14/11 - 5/1/11
 Mo Lee Villagran, 7/31/12 - 9/28/12
 Yuetiva Robles, 8/22/13 - 10/7/13
 Zeran Li, 9/2/15 - 11/2/15
 Umber Dube, 10/26/15 - 1/2/16
 Jiang Zhu, 3/1/17 - 4/14/17
 Geralle Powell, 3/24/17 - 5/19/17

AMGEN:

Drexel Neumann, 6/1/12 - 8/3/12
 Dwani Patel, 5/31/13 - 8/2/13
 Dwani Patel, 6/1/14 - 7/25/14
 Angela Hacker, 6/1/15 - 8/1/15
 Alexander Gabrielli, 6/2/17 - 8/5/17
 Dalton Huey, 5/31/18 – 8/3/18

Mentees:

- 1.- **PI: Heitsch, L** **07/1/2013 – 6/30/2014** **Amount \$50,000**
Emergency Medicine Foundation (EMF) Career Development Grant
“Genetic architecture of human brain ischemia”
 Role: Co-mentor

- 2.- **PI: Heitsch, L** **01/01/2014-12/31/2015** **Amount \$155,000 (\$77,000 per year)**
NCRP Summer 2013 Mentored Clinical & Population Research Award (AHA)
“Genetics of early outcomes after ischemic stroke”
 Role: Co-mentor

3. **PI: Heitsch, L** **01/16/2017-01/01/2022** **Amount \$1,000,000 (\$180,727 per year)**
NINDS
“Genetics of Early Neurological Instability after Ischemic Stroke (GENISIS)”
 Role: Co-mentor

4. **PI: Dhar, R** **01/16/2018-01/01/2023** **Amount \$1,000,000 (\$180,727 per year)**
NINDS K23NS099440
Genetics and Prediction of Cerebral Edema After Hemispheric Stroke
 Role: Co-mentor

Thesis Committee:**Completed**

- 1.- **Jillian Gwen Buchan.** Genomic Analysis of Human Spinal Deformity and Characterization of a Zebrafish Disease Model. May 2014. Division of Biology & Biomedical Sciences. Molecular Genetics and Genomics. Washington University.
- 2.-**Matter Lane:** Network exploration of correlated multivariate protein data for Alzheimer’s disease association. March 31.2017. Master thesis. Department of Mathematics and Computer Science. UMSL.
- 3.- **Daniel Luke Kober.** Structural, Biophysical, and Functional Studies of TREM2 in Neurodegenerative Disease. May 2017. Division of Biology and Biomedical Sciences. Neurosciences. Washington University.

4.- Michael Yip-Hin Chan. A Parallelized Implementation of Cut-and-Solve and a Streamlined Mixed-Integer Linear Programming Model for Finding Genetic Patterns Optimally Associated with Complex Diseases. November 2018. Master thesis. Department of Mathematics and Computer Science. UMSL.

5.- Maitane Ezquerro. Molecular genetic diagnosis of patients from the Basque Country with Inherited Retinal Dystrophies. November 2018. Facultad de Ciencia y Tecnología. Departamento de Genética, Antropología Física y Fisiología Animal. Programa de Doctorado de Biología Molecular y Biomedicina. Universidad del País Vasco. Noviembre 2018. International thesis committee member.

Ongoing

1.- Alexandra Scott. Investigating the functional impacts of both common and rare structural variation using WGS and RNA-seq data from the GTEx cohort. Division of Biology and Biomedical Sciences. Neurosciences. Washington University.

2.- Lilian Antunes. Identify rare coding variants associated with severe adolescent idiopathic scoliosis. Division of Biology and Biomedical Sciences. Neurosciences. Washington University.

3.-Austin McCullough. Characterization and modeling of individual variations in AD pathological trajectories. Division of Biology and Biomedical Sciences. Neurosciences. Washington University. Chair of the thesis committee

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Book chapters:

b. Invited publications

Cruchaga C, Kauwe JSK, Goate AM. Alzheimer's Disease. *Principles of Psychiatric Genetics.* John I. Nurnberger Jr MD (Editor), Wade Berrettini MD (Editor) Cambridge University Press. November 12, 2012 | ISBN-10: 0521896495 | ISBN-13: 978-0521896498

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