

Curriculum Vitae

Wayne N. Frankel, Ph.D., Professor
Columbia University Irving Medical Center

EDUCATION

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| 1982 | B.A. (Biology), The University of Pennsylvania, Philadelphia, PA |
| 1984 | M.S. (Genetics), Albert Einstein College of Medicine, Bronx, NY |
| 1987 | Ph.D. (Genetics), Albert Einstein College of Medicine, Bronx, NY |

AWARDS AND HONORS

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| 1982-1987 | Scholarship, Immunology & Immunooncology Training Fellowship, Albert Einstein College of Medicine, Bronx, NY |
| 1987-1990 | Leukemia Society of America Fellowship, Tufts University School of Medicine, Boston, MA |
| 1990-1993 | Leukemia Society of America Special Fellowship, Tufts University School of Medicine, The Jackson Laboratory, Bar Harbor, ME |
| 1994-1997 | Klingenstein Fellowship in the Neurosciences, The Jackson Laboratory, Bar Harbor, ME |
| 2014-2021 | Javits Neuroscience Investigator Award (MERIT), National Institute of Neurological Disorders and Stroke |
| 2024 | GRI Patient Impact Award, CureGRIN |

RESEARCH POSITIONS

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| 1982-1987 | Graduate Student, Department of Genetics, Albert Einstein College of Medicine, Bronx, NY (advisor: T. V. Rajan) |
| 1987-1991 | Postdoctoral Fellow, Tufts University School of Medicine, Boston, MA (mentor: John M. Coffin). |
| 1992-1995 | Assistant Professor, The Jackson Laboratory, Bar Harbor, ME |
| 1995-2002 | Associate Professor, The Jackson Laboratory, Bar Harbor, ME |
| 2002-2015 | Professor, The Jackson Laboratory, Bar Harbor, ME |
| 2004-2015 | Adjunct Professor, The University of Maine, Orono, ME |
| 2011-2015 | Professor, Tufts University School of Medicine, Boston, MA |
| 2011-2015 | Professor, Tufts University School of Medicine, Sackler School of Graduate Biomedical Sciences, Boston, MA |
| 2015-2021 | Adjunct Professor, The Jackson Laboratory, Bar Harbor, ME |
| 2015-2023 | Professor, Institute for Genomic Medicine, Columbia University Irving Medical Center, NY, NY |
| 2015-2024 | Professor, Department of Genetics and Development, Columbia University Irving Medical Center, NY, NY |
| 2023-2024 | Director, Center for Translational Research in Neurodevelopmental Disease, Columbia University Irving Medical Center, NY, NY |
| 2024-present | Clinical Professor of Neurology (in the Department of Genetics and Development), Columbia University Irving Medical Center, NY, NY |

PROFESSIONAL ACTIVITIES

Editorial and Advisory Board Membership

1994-2010	Editorial Board, <i>Mammalian Genome</i>
1995-2003	Scientific Advisory Board, Digital Gene Technologies, La Jolla, CA
1996-1997	Editorial Board, <i>Genomics</i>
1997-2005	Associate Editor, <i>Genomics</i>
1997-1998	Secretariat, International Mammalian Genome Society
2000-2002	Founding Editor-in-Chief, <i>Gene Expression Patterns</i>
2001-2005	Member, <i>Faculty of 1000</i> , Animal Genetics section
2002-present	Editorial Board, <i>Genes, Brains and Behavior</i>
2005-2008	Founding Editor-in-Chief, <i>PLOS Genetics (Public Library of Science)</i>
2005-2009	Scientific Advisory Board, The EuroHear Consortium
2007-2008	Editorial Board, <i>Epilepsia</i>
2009-2010	Advisory Board <i>Faculty of 1000 Reports</i>
2008-2012	Member, Board of Scientific Counselors, NIH–NIDCD
2010-2012	Chair, Board of Scientific Counselors, NIH–NIDCD
2008-present	Associate Editor, <i>PLOS Genetics (Public Library of Science)</i>
2017-2023	Member, (area co-lead from 2022) Epilepsy Research Benchmarks Stewards Committee, (<i>American Epilepsy Society</i>)
2017-2019	Advisory Board, Alliance for Genome Resources (AGR)
2024-	Contributing Editor, <i>Epilepsy Currents</i>

Specific Extramural Grant Review and Planning Groups:

1993	NICHD Special Review Committee Study Section
1994, 1998	NIA Special Review Committee Study Section
1995	NIAAA Special Review Committee Study Section
1995	NIDCD Special Review Committee Study Section
1996, 1999	Temporary Member, Mammalian Genetics Study Section NIMH, Special Review Committee Study Section
1998	NCI Advisory, Preclinical Models for Cancer Workgroup, Mouse Genomics and Genetics
1998	NCI Review Board, Intramural Program
1998	NIH Advisory, Priority Setting for Mouse Genomics/Genetics Workshop
1998	<i>Ad Hoc</i> Reviewer, ALTX-3 Study Section
1999-2000	NIH BDCN-4 Study Section
1999	Princeton Mouse Genome Sequencing Meeting
1999	Temporary Member, NIH Biol-1 Study Section
1999	NIH/NINDS Advisory, Future Genetic Strategies for Epilepsy Workshop
1999	Genetics in the New Millennium: Directions for the Future, Bar Harbor
2000	NIH Advisory, Priority Setting Mouse Genomics/Genetics Follow-up
2000	NIMH Advisory, Setting Priorities for Phenotyping the Mouse Nervous System and Behavior
2000	Genetics in the New Millennium: A Neuroscience Prospective, Bar Harbor
2001	NIH/NINDS Advisory, Models for Epilepsy and Epileptogenesis, Bethesda
2001	NINDS Special Review Committee, Innovations in Translational Epilepsy Research Study Section
2001	Mouse Analysis Working Group Meeting (Mouse Genome Sequence Consortium), Whitehead Institute Genome Center, Cambridge, MA

- 2001 Chair, Mouse Sequencing Liaison Group (NHGRI-based committee to improve and maintain communication between the genome sequencing centers and the mouse research community)
- 2001 Reviewer, Quality of Life Integrated Projects in Functional Genomics Relating to Human Health (grant solicitation from European Commission)
- 2001 Reviewer, NIH/NINDS Gene Discovery Panel (RFA), Reviewer
- 2001 *Ad Hoc* Reviewer, NIH Social Sciences, Nursing, Epidemiology and Methods (SNEM) Integrated Review Group, SNEM-2
- 2002-2007 Scientific Reviewer, CURE (Citizens United for Research in Epilepsy)
- 2002 *Ad Hoc* Reviewer, NIH/CSR Special Review Committee for Mammalian Genetics
- 2003 *Ad Hoc* Reviewer, Veterans Administration, NEUD subpanel
- 2003 Reviewer, NIH/NINDS Special Review Committee Brain Disorders in the Developing World: Research Across the Lifespan (ZNS1 SRB-H 01)
- 2003 Subcommittee Member, Medical Research Council, Review of the MRC Mammalian Genetics Unit, Harwell, Oxford, UK
- 2004 Reviewer, NIH/NINDS Special Review Committee Model Validation for Antiepileptogenic and Resistant Epilepsy Therapies
- 2005 Panel Reviewer, Genome Canada Competition II. Montreal
- 2006 Reviewer, NIH/NINDS Neuroscience Blueprint Interdisciplinary Center Core Grant (RFA)
- 2006 *Ad Hoc* Reviewer, NIH Clinical Neuroplasticity and Neurotransmitters Study Section
- 2006 *Ad Hoc* Reviewer, NIH Genetics of Health and Disease Study Section
- 2006 *Ad Hoc* Reviewer, NIH/NINDS Workshop, Model Development in Epileptogenesis and Therapy-Resistant Epilepsy and Models of Geriatric Epilepsy
- 2007 Panel Reviewer, Genome Canada International Scientific Review Committee (ISRC) for Competition III. Vancouver, BC.
- 2007 *Ad Hoc* Reviewer, NIH Molecular, Cellular, and Developmental Neuroscience Study Section
- 2009 *Ad Hoc* Reviewer, Genetics IRG, Challenge grants
- 2010-2014 Member, NIH/GHD Study Section
- 2011 Member, Neurotechnology and Neurogenetics Member Conflict Study Section (ZRG1 ETTN A02)
- 2014 *Ad Hoc* Reviewer, GNOM-G study section (CEGs grants)
- 2015 Member BDCN N90 Special Emphasis Panel
- 2016 *Ad Hoc* Reviewer, CNNT study section, Washington, DC
- 2016 *Ad Hoc* Reviewer, ZRG1 BDCN study section, Bethesda, MD
- 2017 *Ad Hoc* Reviewer, ZRG1 BDCN-W (05) study section, Bethesda, MD
- 2017 *Ad Hoc* Reviewer, ZHG1 HGR-P study section, Bethesda, MD
- 2019 Member, U42 Mutant Mouse Resource and Research Centers review panel, NIH
- 2020 Member, P50 Centers for Collaborative Research in Fragile X and FMR1-Associated Conditions
- 2021-2023 National Library of Medicine Board of Regents: Comparative Genomics Resource Working Group (NLM)

CONFERENCE ORGANIZATION

- 1994 Organizer (with A. Messer, R. Reeves, B. Tempel), Workshop in Mouse Molecular Neurogenetics, Bar Harbor
- 1995 Organizer (with A. Chakravarti, S. Ghosh, N. Schork), Biology and Genetics of Complex Mammalian Traits, Bar Harbor

- 1996 Organizer (with K. Herrup, J. Noebels, A. Silva), 2nd Biennial Workshop in Mouse Molecular Neurogenetics, Bar Harbor
- 1996 Organizer (with others), Knockouts and Mutants: Genetically Dissecting Brain & Behavior, San Diego (sponsored by Elsevier)
- 1999 Organizer (with S. Ackerman, M. MacDonald, G. Sutcliffe), 3rd Biennial Workshop in Mouse Molecular Neurogenetics, Bar Harbor
- 2002 Organizer, 4th Biennial Workshop in Mouse Molecular Neurogenetics, Bar Harbor
- 2004 Organizer, 5th Biennial JAX Neurogenetics Conference (formerly Workshop in Mouse Molecular Neurogenetics), Bar Harbor
- 2011 Organizer, Coming Together on Epilepsy Genetics: From Human to Model Organisms, and Back, Conference, Bar Harbor
- 2014 Co-chair (with S. Petrou), "Precision Therapeutics" session, Epilepsy Genetics in the Era of Precision Medicine, San Francisco, CA

INSTITUTIONAL COMMITTEES/SERVICE

The Jackson Laboratory (1992-2015)

- 1992-1993 Reinstated/Organized Genetics Interest Group (Weekly Journal Club)
- 1992-93; 06-07 Training and Education Committee
- 1993-95; 98-99 Scientific Advisory Committee
- 1995 Genetic Resources
- 1995-1997 Staff (Faculty) Recruiting
- 1998-1999 Courses and Meetings
- 1998-2000 TJL Mutagenesis/Physiogenomics Service (Staff Supervisor)
- 1998-2005 Research Grants Committee
- 1999-2001 Director Search Committee
- 2000-01; 05-09 Faculty Recruiting Committee
- 2001 Staff Recruiting Subcommittee, Biophysics
- 2003-2006 Associate Director and Chair of Research Search Committee
- 2003-2011 Promotions Committee
- 2005-2006 Chair, TJL Neuroscience Recruitment Planning Group
- 2005-2008 University of Maine/TJL Functional Genomics Ph.D. Program Recruiting Committee
- 2006-2020 Director, Training In Neurogenetics (NIH-NINDS T32 training grant)
- 2008-2012 Scientific Director of Postdoctoral Training
- 2008-2013 Postdoctoral Training Committee, Chair
- 2009-2014 Predoctoral Training Committee
- 2011-2014 Research Grants Committee
- 2011-2014 Internal Advisory Board, Jackson Aging Center
- 2011-2012 Human Resources Senior Director Search Committee
- 2012-2013 Education Cabinet
- 2013-2014 Radiation and Lab Safety Committee, Chair
- 2013-2015 Education Advisory Committee
- 2014-2015 Predoctoral and Postdoctoral Training Committee

Columbia (2015-present)

- 2017, 2018 Member, Pew Biomedical Scholars review committee
- 2018 Member, Research Initiatives in Science and Engineering (RISE) Competition review committee

2018-2021	Co-chair, Columbia Precision Medicine Initiative Supplemental Funding for Mouse Models of Human Disease
2019-2023	Institutional Biosafety Committee
2020	Member, Revson Fellows review committee

TEACHING

1985	Medical Genetics Instructor, Albert Einstein College of Medicine
1989	Infectious Diseases Instructor, Tufts University School of Medicine
1992-93	Lecturer, Short Course in Medical and Experimental Mammalian Genetics
1995-2000	Lecturer, Short Course in Medical and Experimental Mammalian Genetics
1995-96, 98	Lecturer, Grad-level Genetics Course, Tufts University School of Medicine
1996	Lecturer, Short Course: Modern Genetics for Neuroscientists (held at Winter Conference for Brain Research), Snowmass, CO
1996	Lecturer, Society for Neurosciences Annual Meeting Short Course: What's wrong with my mouse?
1997	Director, Grad-Level Course, Experimental Genetics of the Laboratory Mouse: Genetics, Development and the Brain, The Jackson Laboratory
1997	Lecturer, Grad-Level Genetics Course, Albert Einstein College of Medicine
2016	Lecturer, Genetic Approaches II, Genetics & Development Ph.D. program, Columbia University
2017-2023	Lecturer and Course director, Genetic Approaches to Biological Problems II, (Genetics & Development Ph.D. program required course), Columbia University
2017-2020	Lecturer, Molecular Genetics, (CUMC integrated Ph.D. program), Columbia University
2018	Lecturer, Seminar in Modern Biology (for first year undergraduates, Biological Sciences Department), Columbia University

PH.D. THESIS DEFENSE COMMITTEES (COMPLETED)

1994	Outside Examiner, Kenro Kusumi, Mass Institute of Technology (December).
1996	Committee Chair, Cathleen M. Lutz, University of Maine.
2002	Outside Examiner, Jennifer Hughes, Tufts University Sackler School (September)
2004	Outside Examiner, Christopher Tipper, Tufts University Sackler School (June)
2010	Outside Examiner, Mohd Zulqisti Mohd Zulkifli, University of Melbourne (June)
2011	Outside Examiner, Oya Cingoz, Tufts University (October)
2013	Outside Examiner, Tae Hwan Kim, University of Melbourne (September)
2014	Committee Chair, Christian D. Richard, University of Maine
2017	Committee member, Sedef Tinaztepe, Columbia University
2020	Committee member, Meghan Pantella, Columbia University
2021	Alex Whitebirch, Columbia University
2021	Osasumen Virginia Aimiwu, Columbia University (mentor)
2022	Devin Jones, Columbia University (mentor)
2022	Wanqi Wang, Columbia University (mentor)

INVITED SEMINARS AND PLATFORM PRESENTATIONS

1996	Invited Speaker, "Genetics of epilepsy in mice," Winter Conference for Brain Research, Snowmass (January)
1996	Platform Speaker, Klingenstein Fellows Annual Meeting, Banbury Center, Cold Spring Harbor (April)

- 1996 Invited Speaker, Workshop on Behavioral Phenotypes of Inbred Strains of Mice, NIH, Bethesda (April)
- 1996 Invited Speaker, "Genetics of epilepsy in mice...as of May 30th, 1996," 10th International Workshop, Molecular Genetics of the Mouse, Spa, Belgium (June)
- 1997 Invited Seminar, "Genetics of epilepsy in mice," Cornell Medical School (February)
- 1997 Invited Seminar, "Genetics of epilepsy in mice," Department of Human Genetics, University of Florida, Gainesville (February)
- 1997 Invited Seminar, "Gene hunting (and finding) in mouse models for epilepsy and other neurological disorders," Albert Einstein College of Medicine, Department of Molecular Genetics, Bronx (December)
- 1998 Platform Speaker, "Gene hunting in new and old mouse models of epilepsy" at Epilepsy Research Foundation: Workshop on the Use of Animal Models for Elucidating the Molecular Basis of Epilepsy, Oxford, UK (September)
- 1998 Invited Seminar, "Gene discovery for uncovering molecular mechanisms of neurological disorders" at Department of Biophysics and Physiology, University of Iowa (October)
- 1998 Platform Speaker, "Gene discovery using mouse models: Overview and strategies of positional cloning for neurological traits" at Knockouts and Mutants: Genetically Dissecting Brain and Behavior (sponsored by Brain Research Interactive) San Diego (November)
- 1998 Platform Speaker, "The mouse stargazer gene encodes a candidate α subunit for neuronal calcium channels," at 1998 Annual Meeting of the Society for Neurosciences, Los Angeles (November)
- 1998 Invited Seminar, "Gene hunting in simple and complex mouse models for epilepsy," at Parke-Davis Mouse Genetics Laboratory, Alameda (April)
- 1999 Platform Speaker, "Fidgetin – a novel AAA ATPase disrupted in fidget mutant mice," at Cellular Functions of AAA Proteins, Salk Institute, La Jolla (April)
- 1999 Invited Seminar and Lecture, "Positional cloning mouse genes: α -stargazin and α -fidgetin and α -voiding complex traits?" and "Complex traits in mouse, NHGRI, Bethesda (May)
- 1999 Platform Speaker, "In search of seizure susceptibility genes: QTLs vs Mutagenesis," at Mouse Initiatives: Advanced Functional Genomics, The Jackson Laboratory, Bar Harbor (July)
- 2000 Platform Speaker, "Electroconvulsive screens for mouse strains and mutants," at Mouse Behavioral Phenotyping, Banbury Center, Lloyd Harbor (August)
- 2000 Platform Speaker, "Production and screening of mouse neurological mutants: The Jackson Laboratory's Neuroscience Mutagenesis Facility," at Mutagenesis of the Mouse Genome, University of Georgia, Athens (GA) September
- 2000 Invited Seminar, "Mutation screens for epilepsy and other neurological disorders in mice" Center for Neurobiology and Behavior, University of Pennsylvania, Philadelphia (November)
- 2000 Invited Special Speaker, "Mutation screens for epilepsy and other neurological disorders in mice" The Fourth Annual Einstein Postdoctoral Symposium, Albert Einstein College of Medicine, Bronx (December)
- 2001 Invited Seminar, "Mutation screens for epilepsy and other neurological disorders in mice" Eccles Center for Human Genetics, University of Utah, Salt Lake City (February)
- 2001 Invited Seminar, "Mutation screens for epilepsy and other neurological disorders in mice" Department of Biochemistry, George Washington University School of Medicine, Washington, DC (February)
- 2001 Invited Speaker, "Mutation screens for epilepsy and other neurological disorders in mice" at Genetic Basis of Neurological and Behavioral Disorders (sponsored by The

- Klingenstein Fund), Cold Spring Harbor Laboratory, Cold Spring Harbor (March)
- 2001 Invited Seminar, "Mutation screens for epilepsy and other neurological disorders in mice" Program in Genetics, Tufts University School of Medicine, Boston (April)
- 2001 Invited Speaker, "The effects of natural variants on detection and mapping of novel seizure mutations in mutagenesis screens" at Identification of Genes Underlying ENU-Induced Mutant Phenotypes, McLaughlin Research Institute, Great Falls (July)
- 2001 Platform Speaker, "Mutation screens for epilepsy and other neurological disorders in mice" at Mouse Genome Initiatives III, The Jackson Laboratory, Bar Harbor (August)
- 2001 Invited Speaker, "QTLs meets mutagenesis in Bar Harbor," IBANGS Annual Meeting (International Behavioral and Neuronal Genetics Society), San Diego (November)
- 2001 Platform Speaker, "Electroconvulsive thresholds and antiepileptic drug responses for mouse mutants and strains" at Society for Neuroscience Annual Meeting, San Diego (November)
- 2001 Participant, "New mouse behavioral phenotyping assays and mutagenesis for brain and behavior phenotypes, Half Moon Bay (December)
- 2002 Invited Seminar, "Mutation screens for seizure and other neurological disorders in mice," Duke University Department of Neurobiology, Durham (January)
- 2002 Invited Participant, "International Mouse Mutagenesis Consortium" discussion meeting, London (February)
- 2002 Invited Speaker, "Electroconvulsive thresholds and antiepileptic drug response in mouse mutants and strains" in Nurturing the Genome: A Festschrift for Benson E. Ginsburg, University of Connecticut, Storrs (June)
- 2002 Invited Speaker, "Progress in the JAX Neuroscience Mutagenesis Facility as it approaches its 2nd birthday" in Workshop on ENU Mutagenesis: Planning for Saturation, Chantilly, VA (July)
- 2002 Invited Speaker, "Mouse sequencing and mutations" in Mouse Initiatives IV: Comparative Genomic Approaches to the Analysis of Gene Function and Human Disease, Bar Harbor, ME (August)
- 2002 Invited Seminar, "Progress in mutagenesis screens for new models of epilepsy and other neurological disorders in mice," Tufts University Department of Molecular Biology and Microbiology, Boston (September)
- 2002 Invited Speaker, "Seizure genetics in mice" in Neurogenetics of Childhood Epilepsy (Symposium at Annual Meeting of American Society of Human Genetics, Baltimore, MD (October)
- 2002 Platform Speaker, "Mutant mice with a low electroconvulsive threshold have a spontaneous deletion that includes the Kcnq2 and Chrna4 genes" at Society for Neuroscience Annual Meeting, Orlando, FL (November)
- 2002 Invited Participant, "Beyond the beginning: the future of genomics II. NHGRI-sponsored conference at Airlie Conference Center (November)
- 2003 Invited Speaker, "Screening and characterization of epilepsy related and other neurological mutations in mice" in Molecular Basis of CNS Disorders, Bonn, Germany (February)
- 2003 Platform Speaker, "Spike-wave seizures in the stargazer allelic series," in Spring Epilepsy Research Conference, Grand Cayman, CI (April)
- 2003 Invited Speaker, "New genetically-defined and readily-testable models for epilepsy and for therapy resistance in mice," Division of Neurology, University of Pennsylvania, Philadelphia (September)
- 2003 Platform Speaker, "Stateside ENU: overview of neuro-mutagenesis centers funded by the NIH," The mouse as an instrument for hearing research, Bar Harbor (September)
- 2003 Invited Speaker, "Evolving the electroconvulsive test as a forward-genetic screen for epilepsy research in mice" in Phenotype screens for mice developing an integrated platform; Eumorphia Annual Meeting, Slough/London, UK (October)

- 2003 Invited Speaker, "Forward genetics approaches to new mouse models for epilepsy research and therapy resistance," Wellcome Trust Center for Human Genetics, Oxford, UK (December)
- 2004 Invited Speaker, "Effect of genetic background on induced and spontaneous mouse models for epilepsy" in joint plenary session of Human Genome Sequence Variation and the Inherited Basis of Common Disease and Natural Variation and Quantitative Genetics in Model Organisms, Keystone Symposia, Breckenridge (January)
- 2004 Invited Speaker, "Neurological mutations and modeling for epilepsy and other brain disorders in mice," Vermont Genetics Network Third Annual Retreat, Burlington (August)
- 2004 Invited Speaker, "Genetic studies of new epilepsy mutations and models in mice", University of Tennessee Neuroscience Institute, Memphis (October)
- 2005 Invited Speaker, "Genetic studies of epilepsy in mice: Complexities of simple traits" in Distinguished Scientist Seminar Series at Human and Molecular Genetics Center, Medical College of Wisconsin, Milwaukee (March)
- 2005 Invited Speaker, "New mouse models and genes for epilepsy in mice," at Stanford University, Stanford (May)
- 2006 Invited Speaker, "Genetics of new and old mouse models for epilepsy," Emory University, Atlanta, GA (March)
- 2006 Invited Speaker, "Compromised mRNA processing and epilepsy in Brunol4 mutant mice," American Society of Human Genetics Meeting, New Orleans, LA (October)
- 2006 Invited Speaker, "New and old genetic mouse models for epilepsy," Department of Human Genetics, University of Michigan Medical School (October)
- 2007 Invited Speaker, "New and old genetic models for epilepsy in mice," University of Wisconsin, Madison, WI (April)
- 2007 Plenary session speaker, "Science, Journalism and Cultural Politics," in the conference "Rethinking Communication in Changing Global Contexts", Orono, ME (June)
- 2007 Keynote speaker, The Jackson Laboratory Discovery Strategies Conference: The Laboratory Mouse in the Development of New Therapeutic Approaches to the Treatment of Neurological Disease, Bar Harbor, ME (June)
- 2008 Invited Speaker, "Genetics of absence epilepsy in a common mouse strain," UCI Epilepsy Research Center Symposium, "Hot Topics in Epilepsy" (March)
- 2008 Invited Speaker, Gordon Research Conference, "Mechanisms of Epilepsy & Neuronal Synchronization: Neuroplasticity in Epilepsy," Waterville, ME, (August)
- 2008 Invited Speaker, Mouse Genetics & Genomics: Development & Disease Conference, "New and old genetic models for epilepsy in mice" Cold Spring Harbor Laboratory, Cold Spring Harbor, NY, (November)
- 2009 Invited Speaker, Cornell University, "New and old genetic models for epilepsy in mice," Ithaca, NY, (April)
- 2009 Invited Speaker, Cambridge University Laboratory of Molecular Biology, "New and old genetics models for epilepsy in mice," Cambridge, UK (August).
- 2009 Platform Speaker, International Mouse Genome Conference, "Genetics of absence epilepsy in the C3H superfamily of mouse strains," La Jolla, CA (November)
- 2011 Invited Speaker, University of Maryland School of Pharmacy, "Genetic models of epilepsy in mice," Baltimore, MD (February)
- 2011 Invited Speaker, Vanderbilt University Medical Center, "Genetic models of epilepsy in mice," Nashville, TN (April)
- 2012 Invited Speaker, University of California –Irvine, "Dissecting genetically complex epilepsies in mice: New and old mutants and mechanisms," Irvine, CA (January)
- 2014 Invited Speaker, Audiogenic Epilepsy Congress, "Audiogenic Epilepsy: from models to clinic" Salamanca, Spain (September)

- 2015 Invited Speaker, Université de Montréal, “Neurobiology of Epilepsy: from Gene to Networks,” Montréal, Canada (May)
- 2017 Invited Speaker, Human Genetics in NYC Symposium, “At the Crossroads Where Human and Mouse Epilepsy Genetics Meet,” Mt. Sinai Medical Center, NY (January)
- 2018 Invited Speaker, Stanford University, “Genetic epileptic encephalopathies modeled in mice: Surprises and opportunities”, Palo Alto, CA (May)
- 2019 Invited Speaker, University of New England, “Towards precision medicine in new mouse models of severe childhood epilepsy,” Biddeford, ME (July)
- 2019 Invited Speaker, Emory University Center for Functional Evaluation of Rare Variants, “Mouse Models of GRIN Gain-Of-Function or Dominant Genetic Variants,” Atlanta, GA (September)
- 2020 Invited Speaker, Winter Conference in Brain Research, “Modeling non-ion channel genetic epilepsies in mice,” Big Sky, MT (January)
- 2023 GRICongress Boston 2023, Keynote Speaker, “Assessing gene therapy in mouse models of gain-of-function or dominant-negative childhood seizure disorders” (March)
- 2024 GRICongress 2024 (virtual), Invited Speaker, “Knockdown & Replace” Gene Therapy in a Mouse Model of Severe Childhood Epilepsy” (April)

TRAINEES (POSTDOCS, GRADUATE STUDENTS, SUMMER OR SEMESTER INTERNS)

- 1992-1994 Eric W. Johnson, Ph.D., Postdoctoral Fellow (Iverson Genetics).
- 1993, 1995 Jennifer Gervais (High School), JAX Summer Student Program
- 1993-1997 Cathleen M. Lutz, Predoctoral Fellow, Ph.D. 1997, University of Maine.
- 1994 Audrey Fu (Rice University), JAX Summer Student Program
- 1994-1998 Gregory A. Cox, Ph.D., Postdoctoral Fellow
- 1995 Richard Letts (University of Birmingham, UK), TJL Summer Student Program
- 1995-1999 Marie E. Legare, DVM, Ph.D., Postdoctoral Fellow.
- 1996 Jason Tarbett (University of Maine), JAX Summer Student Program
- 1996 Sara Hipkens (high school) JAX Summer Student
- 1997 Chandrika Rajan (University of Chicago), JAX Summer Student Program
- 1997 Christopher Berry (Duke University), JAX Summer Student Program
- 1998 Elizabeth Good (high school), JAX Summer Student Program
- 1998 Hilary Stanley (high school), JAX Summer Student Program
- 1999-2004 Arne Nystuen, Ph.D., Postdoctoral Fellow
- 2001 Terri Myers (high school), JAX Summer Student Program
- 2001-2006 Yan Yang, Ph.D., Postdoctoral Fellow, The Jackson
- 2003-2004 Amanda Muscat, College Intern (College of the Atlantic); BRIN Fellowship
- 2004-2006 Monica Roberts, Ph.D., Postdoctoral Fellow, The Jackson Laboratory
- 2005 Emily Patek, Rotation Student, University of Maine Ph.D. Program
- 2007-2010 Satoko Tokuda, DVM, Ph.D., Postdoctoral Fellow, The Jackson Laboratory
- 2008 Michael Stanley (high school), The Jackson Laboratory Summer Student
- 2009-2013 Jacy Wagnon, Postdoctoral Fellow, The Jackson Laboratory
- 2010-2012 Wenzhi Sun, Postdoctoral Fellow/Associate Research Scientist
- 2010 Zak Strassberg (high school), Undergraduate Intern
- 2011 Sherie Zhao (University of Virginia), JAX Summer Student Program
- 2011 Zak Strassberg (McGill University), JAX Summer Student Program
- 2011 Alexandra Buckley (Penn State), JAX Summer Student Program
- 2011-2014 Christian Richard, Ph.D. Student, University of Maine

2011-2014	Tracy McGarr, MS Student, University of Maine
2012	Katie Long (University of Michigan), JAX Summer Student Program
*2013	Bethany Berry (Williams College), JAX Summer Student Program
2013	Casey Poore (Brown University), JAX Summer Student Program
2014-2017	Narayan Subramanian, Postdoctoral Fellow, The Jackson Laboratory
2015	Daniel Hawkins (Georgia Tech), JAX Summer Student Program
2015	Jacqueline Bridges (HS junior Phila. area), JAX Summer Student Program
*2016-2019	Ariadna Amador, Ph.D. (Scripps RI), Postdoctoral Fellow, Columbia University
2017-2021	Megha Sah, Ph.D. (U. Conn, Storrs), Postdoctoral Fellow, Columbia University
2017-2022	JiaJie Teoh, Ph.D. (Osaka University), Postdoctoral Fellow, Columbia University
*2017, 2018	Elysia Colon (HS junior, Oceanside High School), Summer Intern
*2017-2021	Virginia Aimuwu (CUMC), Genetics & Development Ph.D. training program
2017-2022	Devin Jones (CUMC), Genetics & Development Ph.D. training program
2018-2023	Wanqi Wang (CUMC), Genetics & Development Ph.D. training program
2018-2019	Jaclyn Dron (Ardsley High School), Summer Intern
2018	Sadi Boddyu (UC Berkeley), Amgen Scholars Program, Summer Intern
2019	Tommy Bocian (Princeton Day School), Summer Intern
2019	Ada Lee (Stuyvesant High School), Summer Intern

*underrepresented minority

PATENTS

1996	In vitro method for identifying a clinical disorder associated with <i>Nhe1</i> mutation (US 5,811,244)
1998	Genes encoding neuronal voltage-gated calcium channel gamma subunits (US 6,365,337)

Active Research Support

R01NS031348-30 (PI: Frankel) 01/15/1993 - 07/30/2026

NIH/NINDS

Genetic Determinants of Epilepsy in Murine Systems (PI: Frankel)

In the current iteration of my longstanding NIH grant that uses mice to study genetic epilepsies, using three conditional mouse models of Developmental and Epileptic Encephalopathy developed during the prior (R37) award period, we will 1. Assess seizure activity and comorbid behaviors in mouse pups. For this, in we will employ new methods to assessing the onset and development of key DEE phenotypes in mice at ages that are more appropriate to the human condition. 2. Determine critical windows, and the role of neuron type, in disease development. We will exploit the conditional feature of our models by crossing to specific cre-driver lines to activate mutations in neuron subtypes or at select ages to better understand the cellular and temporal onset and development of key DEE phenotypes in young and adult mice. This will be complemented by an examination of synaptic mechanisms that underlie the in vivo effects of DEE variants in neuron subtypes. 3. Test different approaches to gene therapy. Recognizing that the genetic and mechanistic heterogeneity of genetic DEEs will require a menu of approaches for treatment, we will employ tailored approaches to somatic manipulation of RNA expression in each of three distinct DEE mouse models. This research will be done in collaboration with the laboratories of Jennifer Gelinis (Columbia), Matthew Weston (U. Vermont) and Scott Harper (Nationwide Children's Hospital).

The United States-Israel Binational Science Foundation (PIs: Frankel/Rubenstein)

Mechanisms of GRIN2D Developmental Delay and Epilepsy

10/01/2020-09/30/2024

Funding at Columbia in this study of a novel mouse model for *GRIN2D* related DEE, is to test pharmacological therapies, focusing on NMDAR receptor antagonists and combination drug therapies in the mouse model

CureGrin Foundation (PI: Frankel) 4/1/2023-3/30/2024

RNAi Therapy in a Mouse Model of GRIN2A Developmental and Epileptic Encephalopathy

The major goal of this project is to test whether the approach of targeting abnormal genes directly using RNA interference approach is effective in an established mouse model of GRIN2A disease.

PUBLICATIONS:

- Frankel, W., et al. (1985). "Retroviral insertional mutagenesis of a target allele in a heterozygous murine cell line." *Proc Natl Acad Sci U S A* 82(19): 6600-6604.
- Potter, T. A., et al. (1987). "Spontaneous deletion at the B2m locus: evidence for site-specific genetic rearrangement." *J Immunol* 138(4): 1270-1274.
- Potter, T. A., et al. (1987). "Mitotic recombination between homologous chromosomes generates H-2 somatic cell variants in vitro." *Proc Natl Acad Sci U S A* 84(6): 1634-1637.
- Coffin, J. M., et al. (1989). "Genetics of endogenous murine leukemia viruses." *Ann N Y Acad Sci* 567: 39-49.
- Frankel, W. N., et al. (1989). "Effect of proviral insertion on transcription of the murine B2mb gene." *J Virol* 63(6): 2623-2628.
- Frankel, W. N., et al. (1989). "Genetic identification of endogenous polytropic proviruses by using recombinant inbred mice." *J Virol* 63(9): 3810-3821.
- Frankel, W. N., et al. (1989). "Genetic analysis of endogenous xenotropic murine leukemia viruses: association with two common mouse mutations and the viral restriction locus Fv-1." *J Virol* 63(4): 1763-1774.
- Frankel, W. N., et al. (1990). "A linkage map of endogenous murine leukemia proviruses." *Genetics* 124(2): 221-236.
- Gerstein, R. M., et al. (1990). "Isotype switching of an immunoglobulin heavy chain transgene occurs by DNA recombination between different chromosomes." *Cell* 63(3): 537-548.
- Rajan, T. V., et al. (1990). "Rate and mechanism of generation of beta 2-microglobulin mutants from a heterozygous murine cell line." *J Immunol* 145(5): 1598-1602.
- Blank, R., et al. (1991). "Mouse chromosome 4." *Mamm Genome* 1 Spec No: S51-78.
- Frankel, W. N., et al. (1991). "Linkage of Mls genes to endogenous mammary tumour viruses of inbred mice." *Nature* 349(6309): 526-528.
- Lamont, C., et al. (1991). "Characterization of endogenous and recombinant proviral elements of a highly tumorigenic AKR cell line." *J Virol* 65(9): 4619-4628.
- Rise, M. L., et al. (1991). "Genes for epilepsy mapped in the mouse." *Science* 253(5020): 669-673.
- Beutner, U., et al. (1992). "Mls-1 is encoded by the long terminal repeat open reading frame of the mouse mammary tumor provirus Mtv-7." *Proc Natl Acad Sci U S A* 89(12): 5432-5436.
- Frankel, W. N., et al. (1992). "Characterization of the endogenous noncotropic murine leukemia viruses of NZB/B1NJ and SM/J inbred strains." *Mamm Genome* 2(2): 110-122.

- Marshall, J. D., et al. (1992). "The AXB and BXA set of recombinant inbred mouse strains." *Mamm Genome* 3(12): 669-680.
- Messer, A., et al. (1992). "Mapping of the motor neuron degeneration (Mnd) gene, a mouse model of amyotrophic lateral sclerosis (ALS)." *Genomics* 13(3): 797-802.
- Prochazka, M., et al. (1992). "NOR/Lt mice: MHC-matched diabetes-resistant control strain for NOD mice." *Diabetes* 41(1): 98-106.
- Taylor, B. A., et al. (1992). "Mouse chromosome 10." *Mamm Genome* 3 Spec No: S153-161.
- Abbott, C. M., et al. (1993). "Encyclopedia of the mouse genome III. October 1993. Mouse chromosome 4." *Mamm Genome* 4 Spec No: S58-71.
- Birkenmeier, E. H., et al. (1993). "Sulfated glycoprotein-2 (Sgp-2) maps to mouse chromosome 14." *Mamm Genome* 4(2): 131-132.
- Bowes, C., et al. (1993). "Localization of a retroviral element within the rd gene coding for the beta subunit of cGMP phosphodiesterase." *Proc Natl Acad Sci U S A* 90(7): 2955-2959.
- Lueders, K. K., et al. (1993). "Genomic mapping of intracisternal A-particle proviral elements." *Mamm Genome* 4(2): 69-77.
- Rinchik, E. M., et al. (1993). "Molecular analysis of viable spontaneous and radiation-induced albino (c)-locus mutations in the mouse." *Mutat Res* 286(2): 199-207.
- Taylor, B. A. and W. N. Frankel (1993). "A new strain congenic for the Mtv-7/MIs-1 locus of mouse chromosome 1." *Immunogenetics* 38(3): 235-237.
- Taylor, B. A., et al. (1993). "Encyclopedia of the mouse genome III. October 1993. Mouse chromosome 10." *Mamm Genome* 4 Spec No: S154-163.
- Asada, Y., et al. (1994). "A mutation in the Ter gene causing increased susceptibility to testicular teratomas maps to mouse chromosome 18." *Nat Genet* 6(4): 363-368.
- Danielson, P. E., et al. (1994). "Chromosomal mapping of mouse genes expressed selectively within the central nervous system." *Genomics* 19(3): 454-461.
- Frankel, W. N. and J. M. Coffin (1994). "Endogenous nonectropic proviruses mapped with oligonucleotide probes from the long terminal repeat region." *Mamm Genome* 5(5): 275-281.
- Frankel, W. N., et al. (1994). "The stumbler mutation maps to proximal mouse chromosome 2." *Mamm Genome* 5(11): 659-662.
- Frankel, W. N., et al. (1994). "Genetic epilepsy model derived from common inbred mouse strains." *Genetics* 138(2): 481-489.
- Lisitsyn, N. A., et al. (1994). "Direct isolation of polymorphic markers linked to a trait by genetically directed representational difference analysis." *Nat Genet* 6(1): 57-63.
- Lueders, K. K. and W. N. Frankel (1994). "Mapping of mouse intracisternal A-particle proviral markers in an interspecific backcross." *Mamm Genome* 5(8): 473-478.
- Neumann, P. E., et al. (1994). "Multifactorial inheritance of neural tube defects: localization of the major gene and recognition of modifiers in ct mutant mice." *Nat Genet* 6(4): 357-362.
- Rowe, L. B., et al. (1994). "Maps from two interspecific backcross DNA panels available as a community genetic mapping resource." *Mamm Genome* 5(5): 253-274.
- Frankel, W. N. (1995). "Of rats, mice, and men?" *Nat Genet* 9(1): 3-4.
- Frankel, W. N. (1995). "Taking stock of complex trait genetics in mice." *Trends Genet* 11(12): 471-477.
- Frankel, W. N., et al. (1995). "Congenic strains reveal effects of the epilepsy quantitative trait locus, EI2, separate from other EI loci." *Mamm Genome* 6(12): 839-843.

- Frankel, W. N., et al. (1995). "New seizure frequency QTL and the complex genetics of epilepsy in EL mice." *Mamm Genome* 6(12): 830-838.
- Hasenkrug, K. J., et al. (1995). "Chromosome mapping of Rfv3, a host resistance gene to Friend murine retrovirus." *J Virol* 69(4): 2617-2620.
- Letts, V. A., et al. (1995). "A curly-tail modifier locus, mct1, on mouse chromosome 17." *Genomics* 29(3): 719-724.
- Messer, A., et al. (1995). "Genetics of primary and timing effects in the mnd mouse." *Am J Med Genet* 57(2): 361-364.
- Naggert, J. K., et al. (1995). "Genomic analysis of the C57BL/Ks mouse strain." *Mamm Genome* 6(2): 131-133.
- Fletcher, C. F., et al. (1996). "Absence epilepsy in tottering mutant mice is associated with calcium channel defects." *Cell* 87(4): 607-617.
- Frankel, W. N. and N. J. Schork (1996). "Who's afraid of epistasis?" *Nat Genet* 14(4): 371-373.
- Hamilton, B. A., et al. (1996). "Disruption of the nuclear hormone receptor RORalpha in staggerer mice." *Nature* 379(6567): 736-739.
- Reuss, F. U., et al. (1996). "Genetics of intracisternal-A-particle-related envelope-encoding proviral elements in mice." *J Virol* 70(9): 6450-6454.
- Cox, G. A., et al. (1997). "Sodium/hydrogen exchanger gene defect in slow-wave epilepsy mutant mice." *Cell* 91(1): 139-148.
- Crawley, J. N., et al. (1997). "Behavioral phenotypes of inbred mouse strains: implications and recommendations for molecular studies." *Psychopharmacology (Berl)* 132(2): 107-124.
- de Lecea, L., et al. (1997). "Cloning, mRNA expression, and chromosomal mapping of mouse and human preprocrystatin." *Genomics* 42(3): 499-506.
- Letts, V. A., et al. (1997). "Genetic and physical maps of the stargazer locus on mouse chromosome 15." *Genomics* 43(1): 62-68.
- Lutz, C. M., et al. (1997). "Neuropeptide Y receptor genes on human chromosome 4q31-q32 map to conserved linkage groups on mouse chromosomes 3 and 8." *Genomics* 41(3): 498-500.
- Lutz, C. M., et al. (1997). "Neuropeptide Y receptor genes mapped in human and mouse: receptors with high affinity for pancreatic polypeptide are not clustered with receptors specific for neuropeptide Y and peptide YY." *Genomics* 46(2): 287-290.
- Wang, H., et al. (1997). "Paroxysmal discharges in the EL mouse, a genetic model of epilepsy." *Brain Res* 760(1-2): 266-271.
- Wong, B. R., et al. (1997). "TRANCE is a novel ligand of the tumor necrosis factor receptor family that activates c-Jun N-terminal kinase in T cells." *J Biol Chem* 272(40): 25190-25194.
- Cox, G. A., et al. (1998). "Identification of the mouse neuromuscular degeneration gene and mapping of a second site suppressor allele." *Neuron* 21(6): 1327-1337.
- de Lecea, L., et al. (1998). "The hypocretins: hypothalamus-specific peptides with neuroexcitatory activity." *Proc Natl Acad Sci U S A* 95(1): 322-327.
- Frankel, W. N. (1998). "Mouse strain backgrounds: more than black and white." *Neuron* 20(2): 183.
- Kusumi, K., et al. (1998). "The mouse pudgy mutation disrupts Delta homologue Dll3 and initiation of early somite boundaries." *Nat Genet* 19(3): 274-278.
- Letts, V. A., et al. (1998). "The mouse stargazer gene encodes a neuronal Ca²⁺-channel gamma subunit." *Nat Genet* 19(4): 340-347.

- Lorenzon, N. M., et al. (1998). "Altered calcium channel currents in Purkinje cells of the neurological mutant mouse leaner." *J Neurosci* 18(12): 4482-4489.
- Beamer, W. G., et al. (1999). "Quantitative trait loci for bone density in C57BL/6J and CAST/EiJ inbred mice." *Mamm Genome* 10(11): 1043-1049.
- Fletcher, C. F. and W. N. Frankel (1999). "Ataxic mouse mutants and molecular mechanisms of absence epilepsy." *Hum Mol Genet* 8(10): 1907-1912.
- Frankel, W. N. (1999). "Detecting genes in new and old mouse models for epilepsy: a prospectus through the magnifying glass." *Epilepsy Res* 36(2-3): 97-110.
- Super, H. J., et al. (1999). "Fine mapping of the friend retrovirus resistance gene, Rfv3, on mouse chromosome 15." *J Virol* 73(9): 7848-7852.
- Upadhyaya, P., et al. (1999). "Genetic modifiers of polycystic kidney disease in intersubspecific KAT2J mutants." *Genomics* 58(2): 129-137.
- Cox, G. A., et al. (2000). "The mouse fidgetin gene defines a new role for AAA family proteins in mammalian development." *Nat Genet* 26(2): 198-202.
- Frankel, W. N. (2000). "Mouse mutagenesis is 'in'. Sharing experience and resources in large-scale functional genomics: mutagenesis of the mouse genome, Georgia Genetics Symposium II, Athens, Georgia, 6-9 September 2000." *Trends Genet* 16(12): 540.
- Legare, M. E., et al. (2000). "A major effect QTL determined by multiple genes in epileptic EL mice." *Genome Res* 10(1): 42-48.
- Legare, M. E. and W. N. Frankel (2000). "Multiple seizure susceptibility genes on chromosome 7 in SWXL-4 congenic mouse strains." *Genomics* 70(1): 62-65.
- Letts, V. A., et al. (2000). "A new spontaneous mouse mutation in the Kcne1 gene." *Mamm Genome* 11(10): 831-835.
- Nadeau, J. H. and W. N. Frankel (2000). "The roads from phenotypic variation to gene discovery: mutagenesis versus QTLs." *Nat Genet* 25(4): 381-384.
- Barclay, J., et al. (2001). "Ducky mouse phenotype of epilepsy and ataxia is associated with mutations in the Cacna2d2 gene and decreased calcium channel current in cerebellar Purkinje cells." *J Neurosci* 21(16): 6095-6104.
- Bryda, E. C., et al. (2001). "High-resolution genetic and physical mapping of modifier-of-deafwaddler (mdfw) and Waltzer (Cdh23v)." *Genomics* 73(3): 338-342.
- Frankel, W. N., et al. (2001). "Electroconvulsive thresholds of inbred mouse strains." *Genomics* 74(3): 306-312.
- Hamilton, B. A. and W. N. Frankel (2001). "Of mice and genome sequence." *Cell* 107(1): 13-16.
- Kang, M. G., et al. (2001). "Biochemical and biophysical evidence for gamma 2 subunit association with neuronal voltage-activated Ca²⁺ channels." *J Biol Chem* 276(35): 32917-32924.
- Nadeau, J. H., et al. (2001). "Sequence interpretation. Functional annotation of mouse genome sequences." *Science* 291(5507): 1251-1255.
- Naf, D., et al. (2001). "Mouse models for the Wolf-Hirschhorn deletion syndrome." *Hum Mol Genet* 10(2): 91-98.
- Nystuen, A., et al. (2001). "A null mutation in inositol polyphosphate 4-phosphatase type I causes selective neuronal loss in weeble mutant mice." *Neuron* 32(2): 203-212.
- Garvey, S. M., et al. (2002). "The muscular dystrophy with myositis (mdm) mouse mutation disrupts a skeletal muscle-specific domain of titin." *Genomics* 79(2): 146-149.
- Klein, J. A., et al. (2002). "The harlequin mouse mutation downregulates apoptosis-inducing factor." *Nature* 419(6905): 367-374.

- Mouse Genome Sequencing, C., et al. (2002). "Initial sequencing and comparative analysis of the mouse genome." *Nature* 420(6915): 520-562.
- Letts, V. A., et al. (2003). "Phenotypic heterogeneity in the stargazin allelic series." *Mamm Genome* 14(8): 506-513.
- Yang, Y., et al. (2003). "Spontaneous deletion of epilepsy gene orthologs in a mutant mouse with a low electroconvulsive threshold." *Hum Mol Genet* 12(9): 975-984.
- Buchner, D. A., et al. (2004). "Three ENU-induced neurological mutations in the pore loop of sodium channel Scn8a (Na(v)1.6) and a genetically linked retinal mutation, rd13." *Mamm Genome* 15(5): 344-351.
- Bult, C., et al. (2004). "A genome end-game: understanding gene function in the nervous system." *Nat Neurosci* 7(5): 484-485.
- Clark, A. T., et al. (2004). "Implementing large-scale ENU mutagenesis screens in North America." *Genetica* 122(1): 51-64.
- Concepcion, D., et al. (2004). "Mutation rate and predicted phenotypic target sizes in ethylnitrosourea-treated mice." *Genetics* 168(2): 953-959.
- Goldowitz, D., et al. (2004). "Large-scale mutagenesis of the mouse to understand the genetic bases of nervous system structure and function." *Brain Res Mol Brain Res* 132(2): 105-115.
- Hertzano, R., et al. (2004). "Transcription profiling of inner ears from Pou4f3(ddl/ddl) identifies Gfi1 as a target of the Pou4f3 deafness gene." *Hum Mol Genet* 13(18): 2143-2153.
- Kitami, T., et al. (2004). "Genetic and phenotypic analysis of seizure susceptibility in PL/J mice." *Mamm Genome* 15(9): 698-703.
- O'Brien, T. P. and W. N. Frankel (2004). "Moving forward with chemical mutagenesis in the mouse." *J Physiol* 554(Pt 1): 13-21.
- Otto, J. F., et al. (2004). "Mice carrying the szt1 mutation exhibit increased seizure susceptibility and altered sensitivity to compounds acting at the m-channel." *Epilepsia* 45(9): 1009-1016.
- Yang, Y. and W. N. Frankel (2004). "Genetic approaches to studying mouse models of human seizure disorders." *Adv Exp Med Biol* 548: 1-11.
- Frankel, W. N. (2005). "Introducing PLoS Genetics." *PLoS Genet* 1(1): e21.
- Frankel, W. N., et al. (2005). "Development of a new genetic model for absence epilepsy: spike-wave seizures in C3H/He and backcross mice." *J Neurosci* 25(13): 3452-3458.
- Letts, V. A., et al. (2005). "A targeted mutation in Cacng4 exacerbates spike-wave seizures in stargazer (Cacng2) mice." *Proc Natl Acad Sci U S A* 102(6): 2123-2128.
- Shin, H. W., et al. (2005). "An enzymatic cascade of Rab5 effectors regulates phosphoinositide turnover in the endocytic pathway." *J Cell Biol* 170(4): 607-618.
- Wooley, C. M., et al. (2005). "Gait analysis detects early changes in transgenic SOD1(G93A) mice." *Muscle Nerve* 32(1): 43-50.
- Yang, Y., et al. (2005). "Functional characterization of fidgetin, an AAA-family protein mutated in fidget mice." *Exp Cell Res* 304(1): 50-58.
- Kearney, J. A., et al. (2006). "Severe epilepsy resulting from genetic interaction between Scn2a and Kcnq2." *Hum Mol Genet* 15(6): 1043-1048.
- Otto, J. F., et al. (2006). "A spontaneous mutation involving Kcnq2 (Kv7.2) reduces M-current density and spike frequency adaptation in mouse CA1 neurons." *J Neurosci* 26(7): 2053-2059.
- Sher, R. B., et al. (2006). "A rostrocaudal muscular dystrophy caused by a defect in choline kinase beta, the first enzyme in phosphatidylcholine biosynthesis." *J Biol Chem* 281(8): 4938-4948.

- Yang, Y., et al. (2006). "Interaction between fidgetin and protein kinase A-anchoring protein AKAP95 is critical for palatogenesis in the mouse." *J Biol Chem* 281(31): 22352-22359.
- Zhou, X., et al. (2006). "Auditory brainstem responses in 10 inbred strains of mice." *Brain Res* 1091(1): 16-26.
- Yang, Y., et al. (2007). "Complex seizure disorder caused by Brunol4 deficiency in mice." *PLoS Genet* 3(7): e124.
- Beyer, B., et al. (2008). "Absence seizures in C3H/HeJ and knockout mice caused by mutation of the AMPA receptor subunit Gria4." *Hum Mol Genet* 17(12): 1738-1749.
- Frankel, W. N. and G. S. Barsh (2008). "PLoS Genetics turns three: looking back, looking ahead." *PLoS Genet* 4(7): e1000135.
- Howell, V. M., et al. (2008). "A targeted deleterious allele of the splicing factor SCNM1 in the mouse." *Genetics* 180(3): 1419-1427.
- Miki, T., et al. (2008). "Two novel alleles of tottering with distinct Ca(v)2.1 calcium channel neuropathologies." *Neuroscience* 155(1): 31-44.
- Frankel, W. N. (2009). "Genetics of complex neurological disease: challenges and opportunities for modeling epilepsy in mice and rats." *Trends Genet* 25(8): 361-367.
- Frankel, W. N., et al. (2009). "Szt2, a novel gene for seizure threshold in mice." *Genes Brain Behav* 8(5): 568-576.
- Papale, L. A., et al. (2009). "Heterozygous mutations of the voltage-gated sodium channel SCN8A are associated with spike-wave discharges and absence epilepsy in mice." *Hum Mol Genet* 18(9): 1633-1641.
- Tokuda, S., et al. (2009). "Genetic complexity of absence seizures in substrains of C3H mice." *Genes Brain Behav* 8(3): 283-289.
- Boumil, R. M., et al. (2010). "A missense mutation in a highly conserved alternate exon of dynamin-1 causes epilepsy in fitful mice." *PLoS Genet* 6(8).
- Hawkins, N. A., et al. (2011). "Neuronal voltage-gated ion channels are genetic modifiers of generalized epilepsy with febrile seizures plus." *Neurobiol Dis* 41(3): 655-660.
- Paz, J. T., et al. (2011). "A new mode of corticothalamic transmission revealed in the Gria4(-/-) model of absence epilepsy." *Nat Neurosci* 14(9): 1167-1173.
- Tokuda, S., et al. (2011). "A novel Akt3 mutation associated with enhanced kinase activity and seizure susceptibility in mice." *Hum Mol Genet* 20(5): 988-999.
- Wagon, J. L., et al. (2011). "Etiology of a genetically complex seizure disorder in Celf4 mutant mice." *Genes Brain Behav* 10(7): 765-777.
- Nellaker, C., et al. (2012). "The genomic landscape shaped by selection on transposable elements across 18 mouse strains." *Genome Biol* 13(6): R45.
- Wagon, J. L., et al. (2012). "CEL4 regulates translation and local abundance of a vast set of mRNAs, including genes associated with regulation of synaptic function." *PLoS Genet* 8(11): e1003067.
- Park, H. J., et al. (2013). "Elevated Id2 expression results in precocious neural stem cell depletion and abnormal brain development." *Stem Cells* 31(5): 1010-1021.
- Sun, W., et al. (2013). "Aberrant sodium channel activity in the complex seizure disorder of Celf4 mutant mice." *J Physiol* 591(1): 241-255.
- Anderson, L. L., et al. (2014). "Antiepileptic activity of preferential inhibitors of persistent sodium current." *Epilepsia* 55(8): 1274-1283.

- Frankel, W. N., et al. (2014). "Unraveling genetic modifiers in the *gria4* mouse model of absence epilepsy." *PLoS Genet* 10(7): e1004454.
- Letts, V. A., et al. (2014). "Hidden in plain sight: spike-wave discharges in mouse inbred strains." *Genes Brain Behav* 13(6): 519-526.
- Neef, J., et al. (2014). "Modes and regulation of endocytic membrane retrieval in mouse auditory hair cells." *J Neurosci* 34(3): 705-716.
- Oliva, M. K., et al. (2014). "Physiological and genetic analysis of multiple sodium channel variants in a model of genetic absence epilepsy." *Neurobiol Dis* 67: 180-190.
- Tyler, A. L., et al. (2014). "A genetic interaction network model of a complex neurological disease." *Genes Brain Behav* 13(8): 831-840.
- Zhang, W., et al. (2014). "Loss of MeCP2 from forebrain excitatory neurons leads to cortical hyperexcitation and seizures." *J Neurosci* 34(7): 2754-2763.
- Asinof, S. K., et al. (2015). "Independent Neuronal Origin of Seizures and Behavioral Comorbidities in an Animal Model of a Severe Childhood Genetic Epileptic Encephalopathy." *PLoS Genet* 11(6): e1005347.
- Dhindsa, R. S., et al. (2015). "Epileptic encephalopathy-causing mutations in DNM1 impair synaptic vesicle endocytosis." *Neurol Genet* 1(1): e4.
- Jackson, H. M., et al. (2015). "DBA/2J genetic background exacerbates spontaneous lethal seizures but lessens amyloid deposition in a mouse model of Alzheimer's disease." *PLoS One* 10(5): e0125897.
- Richard, C. D., et al. (2015). "SWDreader: a wavelet-based algorithm using spectral phase to characterize spike-wave morphological variation in genetic models of absence epilepsy." *J Neurosci Methods* 242: 127-140.
- Asinof, S., et al. (2016). "Dynamin 1 isoform roles in a mouse model of severe childhood epileptic encephalopathy." *Neurobiol Dis* 95: 1-11.
- McSweeney, K. M., et al. (2016). "Inhibition of microRNA 128 promotes excitability of cultured cortical neuronal networks." *Genome Res* 26(10): 1411-1416.
- Wolfson, R. L., et al. (2017). "KICSTOR recruits GATOR1 to the lysosome and is necessary for nutrients to regulate mTORC1." *Nature* 543(7645): 438-442.
- Gelfman, S., et al. (2018). "meaRtools: An R package for the analysis of neuronal networks recorded on microelectrode arrays." *PLoS Comput Biol* 14(10): e1006506.
- Lin, J., et al. (2019). "PRAS: Predicting functional targets of RNA binding proteins based on CLIP-seq peaks." *PLoS Comput Biol* 15(8): e1007227.
- Teoh JJ, Subramanian N, Pero M, Bartolini F, Amador A, Kanber A, Williams D, Petri S, Yang M, Allen A, Beal J, Haut S, Frankel WN (2019). *Arfgef1* haploinsufficiency in mice alters neuronal endosome composition decreases membrane surface postsynaptic GABA_A receptors. *Neurobiology of Disease* 134:104632. doi 10.1016/j.nbd.2019.104632.
- Binder DK, Boison D, Eid T, Frankel WN, Mingorance A, Smith BN, Dacks PA, Whittemore V, Poduri A; (2020). AES/NINDS Epilepsy Benchmarks Stewards Epilepsy Benchmarks Area II: Prevent Epilepsy and Its Progression. *Epilepsy Curr.* Jan-Feb;20(1_suppl):14S-22S. doi: 10.1177/1535759719895274. Epub 2020 Jan 15. PMID: 31937124
- Sah M, Shore AN, Petri S, Kanber A, Yang M, Weston MC, Frankel WN. (2020). Altered excitatory transmission onto hippocampal interneurons in the IQSEC2 mouse model of X-linked neurodevelopmental disease. *Neurobiol Dis.* 2020 Jan 21;137:104758. PMID: 31978606.
- Aimiuwu OV, Fowler AM, Sah M, Teoh JJ, Kanber A, Pyne NK, Petri S, Rosenthal-Weiss C, Yang M, Harper SQ, Frankel WN. RNAi-based gene therapy rescues

developmental and epileptic encephalopathy in a genetic mouse model. *Mol. Therapy* 2020 DOI:<https://doi.org/10.1016/j.ymthe.2020.04.007>

Amador A, Bostick CD, Olson H, Peters J, Camp CR, Krizay D, Chen W, Han W, Tang W, Kanber A, Kim S, Teoh J, Sah M, Petri S, Paek H, Kim A, Lutz CM, Yang M, Myers SJ, Bhattacharya S, Yuan H, Goldstein DB, Poduri A, Boland MJ, Traynelis SF, Frankel WN. Modelling and treating GRIN2A developmental and epileptic encephalopathy in mice.

McCabe MP, Shore AN, Frankel WN, Weston MC. Altered Fast Synaptic Transmission in a Mouse Model of DNM1-Associated Developmental Epileptic Encephalopathy. *eNeuro*. 2021 Mar 10;8(2):ENEURO.0269-20.2020. doi: 10.1523/ENEURO.0269-20.2020. PMID: 33372033; PMCID: PMC7986544.

Tang M, Park SH, Petri S, Yu H, Rueda CB, Abel ED, Kim CY, Hillman EM, Li F, Lee Y, Ding L, Jagadish S, Frankel WN, De Vivo DC, Monani UR. An early endothelial cell-specific requirement for Glut1 is revealed in Glut1 deficiency syndrome model mice. *JCI Insight*. 2021 Feb 8;6(3):e145789. doi: 10.1172/jci.insight.145789. PMID: 33351789; PMCID: PMC7934852.

Shore AN, Colombo S, Tobin WF, Petri S, Cullen ER, Dominguez S, Bostick CD, Beaumont MA, Williams D, Khodagholy D, Yang M, Lutz CM, Peng Y, Gelinis JN, Goldstein DB, Boland MJ, Frankel WN, Weston MC. The paradoxical effects of K⁺ channel gain-of-function are mediated by GABAergic neuron hypoexcitability and hyperconnectivity. *Cell Rep*. 2020;33(4):108303. Epub 2020/10/29. doi: 10.1016/j.celrep.2020.108303. PubMed PMID: 33113364

Wang W, Frankel WN. Overlaps, gaps, and complexities of mouse models of Developmental and Epileptic Encephalopathy. *Neurobiol Dis*. 2021 Jan;148:105220. doi: 10.1016/j.nbd.2020.105220. Epub 2020 Dec 7. PMID: 33301879; PMCID:PMC8547712.

Kapur M, Ganguly A, Nagy G, Adamson SI, Chuang JH, Frankel WN, Ackerman SL. Expression of the Neuronal tRNA n-Tr20 Regulates Synaptic Transmission and Seizure Susceptibility. *Neuron*. 2020. Epub 2020/08/28. doi: 10.1016/j.neuron.2020.07.023. PubMed PMID: 32853550.

Gill BJA, Khan FA, Goldberg AR, Merricks EM, Wu X, Sosunov AA, et al. Single Unit Analysis and Wide-Field Imaging Reveal Alterations in Excitatory and Inhibitory Neurons in Glioma *Brain* 2022; accepted.

Teng S, Zhen F, McRae BR, Zhu E, Frankel WN, Peng Y. Sensory regulation of absence seizures in a mouse model of Gnb1 encephalopathy. *iScience*. 2022 Nov 9;25(11):105488. doi: 10.1016/j.isci.2022.105488. eCollection 2022 Nov 18.

Wu X, Sosunov AA, Lado W, Teoh JJ, Ham A, Li H, Al-Dalahmah O, Gill BJA, Arancio O, Schevon CA, Frankel WN, McKhann GM 2nd, Sulzer D, Goldman JE, Tang G. Synaptic hyperexcitability of cytomegalic pyramidal neurons contributes to epileptogenesis in tuberous sclerosis complex. *Cell Rep*. 2022 Jul 19;40(3):111085. doi: 10.1016/j.celrep.2022.111085. PMID: 35858542

Colombo S, Reddy HP, Petri S, Williams DJ, Shalomov B, Dhindsa RS, Gelfman S, Krizay D, Bera AK, Yang M, Peng Y, Makinson CD, Boland MJ, Frankel WN, Goldstein DB, Dascal N.

Epilepsy in a mouse model of GNB1 encephalopathy arises from altered potassium (GIRK) channel signaling and is alleviated by a GIRK inhibitor. *Front Cell Neurosci.* 2023 May 18;17:1175895. doi: 10.3389/fncel.2023.1175895. eCollection 2023. PMID: 37275776

Dugger SA, Dhindsa RS, Sampaio GA, Ressler AK, Rafikian EE, Petri S, Letts VA, Teoh J, Ye J, Colombo S, Peng Y, Yang M, Boland MJ, Frankel WN, Goldstein DB. Neurodevelopmental deficits and cell-type-specific transcriptomic perturbations in a mouse model of HNRNPU haploinsufficiency. *PLoS Genet.* 2023 Oct 2;19(10):e1010952. Doi 10.1371/journal.pgen.1010952. eCollection 2023 Oct. PMID: 37782669

Shore AN, Li K, Safari M, Qunies AM, Spitznagel BD, Weaver CD, Emmitte KA, Frankel WN, Weston MC. Heterozygous expression of a *Kcnt1* gain-of-function variant has differential effects on SST- and PV-expressing cortical GABAergic neurons. *bioRxiv [Preprint]*. 2024 May 29:2023.10.11.561953. doi: 10.1101/2023.10.11.561953. PMID: 37873369

Wanqi Wang, Damian J. Williams, Jia Jie Teoh, Divyalakshmi Soundararajan, AamirZuberi, Cathleen M. Lutz, Wayne N. Frankel, Christopher D. Makinson. Impaired axon initial segment structure and function in a model of *ARHGEF9* Developmental and Epileptic Encephalopathy. *Proc. Natl Acad Sci* (accepted)

Jones DJ, Soundararajan D, Taylor NK, Aimiwu OV, Mathkar P, Shore A, Teoh JJ, Wang W, Sands TT, Weston MC, Harper SQ, Frankel WN. Effective knockdown - replace gene therapy in a novel mouse model of DNM1 developmental and epileptic encephalopathy. *Mol Ther.* 2024 Aug 9:S1525-0016(24)00531-8. doi: 10.1016/j.ymthe.2024.08.009. Online ahead of print. PMID: 39127888