

# Joshua M. Shulman, M.D., Ph.D.



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- **Principal Investigator, Laboratory for Integrative Functional Genomics**

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## CONTACT INFORMATION

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## PROFESSIONAL STATEMENT

Recent advances have made the discovery of genetic susceptibility loci for complex human phenotypes a reality, including nervous system disorders. The critical next step will be to definitively identify the responsible genes and understand their functions in both health and disease. Our research integrates genetic investigation in human subjects and model organisms, with the goal of understanding brain function and aging, and improving the treatment of neurologic disease. We focus on Alzheimer's disease and Parkinson's disease, two incurable neurodegenerative disorders and experimental paradigms for the age-dependent failure of brain cognitive and motor control in humans.

**Human Genetics:** The clinical manifestation of neurodegenerative disease is the culmination of a multi-tiered pathogenic cascade that evolves over decades—understanding how genetic variants impact this causal chain is essential. Although 2% of the population over age 65 are clinically diagnosed with Parkinson's disease, the defining pathology of disease (alpha-synuclein Lewy bodies) is discovered in 20% of brains from population-based autopsy studies. We are therefore investigating the impact of genomic variation on directly measured Lewy pathology, neuronal loss in the midbrain substantia nigra, and progressive motor impairment, leveraging human subject cohorts with detailed clinical and pathological data. We also participate in collaborative studies for the functional genetic dissection of Alzheimer's disease, focusing on the responsible neuropathology, amyloid neuritic plaques and Tau neurofibrillary tangles.

**Drosophila Genetics:** Despite the promise of current human genetic methods, such as genome-wide association studies, they often fail to identify disease susceptibility genes with certainty, instead highlighting broad genomic regions. We are taking advantage of the rapid and powerful genetics available in the fruit fly *Drosophila melanogaster* in order to accelerate the validation of responsible genes and an understanding of their functions in disease pathogenesis. Expression of human amyloid-beta, Tau, or alpha-synuclein proteins in the fly nervous system recapitulates many core features of Alzheimer's disease and Parkinson's disease pathogenesis. We are testing candidate human susceptibility genes for functional genetic interactions in these fly models of neurodegeneration. Implicated molecular pathways are probed in greater depth, using both *Drosophila* and human genetic approaches. Our strategy has recently identified cell adhesion converging on the cytoskeleton as likely important for Tau-mediated neurodegeneration and Alzheimer's disease susceptibility, and we are now following up these insights to elucidate the detailed mechanisms.

## SPECIALITY

- Neurogenetics
- Parkinson's Disease
- Movement Disorders

## EDUCATION

- M.D., Harvard Medical School & M.I.T. Health Sciences and Technology, Boston, Mass.
- Ph.D., University of Cambridge, United Kingdom
- Internship, Internal Medicine, Massachusetts General Hospital, Boston, Mass.
- Residency, Neurology, Brigham and Women's Hospital & Massachusetts General Hospital, Boston, Mass.
- Clinical Fellowship, Movement and Memory Disorders, Brigham and Women's Hospital & Massachusetts General Hospital, Boston, Mass.
- Research Fellowship, Neurogenetics, Brigham and Women's Hospital & Broad Institute of Harvard/M.I.T., Boston, Mass.

## WEBSITE

- Laboratory for Integrative Functional Genomics  
(<https://www.bcm.edu/departments/neurology/research/labs/integrative-functional-genomics>)

## CLINIC APPOINTMENTS

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## CONSULT

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## CLINIC LOCATION

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## CLINICAL INTERESTS

Parkinson's disease and related movement disorders

## RESEARCH AREAS

- Functional genomics of Alzheimer's disease and Parkinson's disease
- Integrative genetic analyses in humans and *Drosophila*

## GRANTS (NIH FUNDING)

- Shulman JM (Principal Investigator). Functional validation of the CD2AP susceptibility network in Alzheimer's disease. National Institute on Aging R01 grant (PA-13-302); \$270,048 awarded in direct costs, May 1, 2016–April 30, 2021.
- Shulman JM (Principal Investigator). Tau-spliceosome interactions in Alzheimer's disease. National Institute on Aging R01 grant (PAR-15-358); \$507,222 awarded in direct costs, Sept. 15, 2016–April 30, 2021.
- Shulman JM (Principal Investigator). Functional Validation of Parkinsons Disease Susceptibility Genes in Drosophila. National Institute of Neurological Disorders and Stroke R21 grant (PA-13-303); \$275,000 awarded in direct costs, Sept. 1, 2014–Aug. 31, 2016.
- Shulman JM (Principal Investigator). Exploring the Genetics of Alzheimer's Disease in Humans and Drosophila. National Institute on Aging K08 grant (PA-06-512); \$236,250 awarded in direct costs, Aug. 15, 2009–July 31, 2014.

## JOURNAL PUBLICATIONS

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2. Li J, Amoh BK, McCormick E, Tarkunde A, Zhu KF, Perez A, et al. Integration of transcriptome-wide association study with neuronal dysfunction assays provides functional genomics evidence for Parkinson's disease genes. *Hum Mol Genet*. 2023;32(4):685-95. PMID: 36173927.
3. Wu T, Deger JM, Ye H, Guo C, Dhindsa J, Pekarek BT, et al. Tau polarizes an aging transcriptional signature to

- excitatory neurons and glia. *elife*. 2023;12:. PMID: 37219079.
4. Ye H, Robak LA, Yu M, Cykowski M, Shulman JM. Genetics and pathogenesis of Parkinson's syndrome. *Annu Rev Pathol*. 2023;18:95-121. PMID: 36100231.
  5. Yu M, Ye H, De-Paula RB, Mangleburg CG, Wu T, Lee TV, et al. Functional screening of lysosomal storage disorder genes identifies modifiers of alpha-synuclein neurotoxicity. *PLoS Genet*. 2023;19(5):e1010760. PMID: 37200393.
  6. Hill EJ, Robak LA, Al-Ouran R, Deger J, Fong JC, Vandeventer PJ, et al. Genome sequencing in the Parkinson disease clinic. *Neurol Genet*. 2022;8(4):e200002. PMID: 35747619.
  7. Lagisetty Y, Bourquard T, Al-Ramahi I, Mangleburg CG, Mota S, Soleimani S, et al. Identification of risk genes for Alzheimer's disease by gene embedding. *Cell Genom*. 2022;2(9):. PMID: 36268052.
  8. Ye F, Funk Q, Rockers E, Shulman JM, Masdeu JC, Pascual B, et al. In Alzheimer-prone brain regions, metabolism and risk-gene expression are strongly correlated. *Brain Commun*. 2022;4(5):. PMID: 36092303.
  9. Abreha MH, Ojelade S, Dammer EB, McEachin ZT, Duong DM, Gearing M, et al. TBK1 interacts with tau and enhances neurodegeneration in tauopathy. *J Biol Chem*. 2021;296:100760. PMID: 33965374.
  10. Alfradique-Dunham I, Al-Ouran R, von Coelln R, Blauwendraat C, Hill E, Luo L, et al. Genome-wide association study meta-analysis for Parkinson disease motor subtypes. *Neurol Genet*. 2021;7(2):e557. PMID: 33987465.
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  12. Guo Q, Dammer EB, Zhou M, Kundinger SR, Gearing M, Lah JJ, et al. Targeted quantification of detergent-insoluble RNA-binding proteins in human brain reveals stage and disease specific co-aggregation in Alzheimer's disease. *Front Mol Neurosci*. 2021;14:623659. PMID: 33815056.
  13. Hill EJ, Mangleburg CG, Alfradique-Dunham I, Ripperger B, Stillwell A, Saade H, et al. Quantitative mobility measures complement the MDS-UPDRS for characterization of Parkinson's disease heterogeneity. *Parkinsonism Relat Disord*. 2021;84:105-11. PMID: 33607526.
  14. Petyuk VA, Yu L, Olson HM, Yu F, Clair G, Qian WJ, et al. Proteomic profiling of the substantia nigra to identify determinants of Lewy body pathology and dopaminergic neuronal loss. *J Proteome Res*. 2021;20(5):2266-82. PMID: 33900085.
  15. Blauwendraat C, Reed X, Krohn L, Heilbron K, Bandres-Ciga S, Tan M, et al. Genetic modifiers of risk and age at onset in GBA associated Parkinson's disease and Lewy body dementia. *Brain*. 2020;143(1):234-48. PMID: 31755958.
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  17. Mangleburg CG, Wu T, Yalamanchili HK, Guo C, Hsieh YC, Duong DM, et al. Integrated analysis of the aging brain transcriptome and proteome in tauopathy. *Mol Neurodegener*. 2020;15(1):56. PMID: 32993812.
  18. Robak LA, Du R, Yuan B, Gu S, Alfradique-Dunham I, Kondapalli V, et al. Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. *Neurol Genet*. 2020;6(5):e498. PMID: 32802956.
  19. Wan YW, Al-Ouran R, Mangleburg CG, Perumal TM, Lee TV, Allison K, et al. Meta-analysis of the Alzheimer's disease human brain transcriptome and functional dissection in mouse models. *Cell Rep*. 2020;32(2):107908. PMID: 32668255.
  20. Yao T, Sweeney E, Nagorski J, Shulman JM, Allen GI. Quantifying cognitive resilience in Alzheimer's Disease: The Alzheimer's Disease Cognitive Resilience Score. *PLoS One*. 2020;15(11):e0241707. PMID: 33152028.
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  22. Al-Ouran R, Wan YW, Mangleburg CG, Lee TV, Allison K, Shulman JM, et al. A portal to visualize transcriptome profiles in mouse models of neurological disorders. *Genes (Basel)*. 2019;10(10):. PMID: 31561642.
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- Alzheimer's Disease Risk Gene, Is Required for Synaptic Transmission and Proteostasis. *Cell Rep.* 2019;28(7):1799-813 e5. PMID: 31412248.
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## POSTER and PLATFORM PRESENTATIONS

1. Zhao J, Gu M, Yu M, Barros De Paula R, Bellen H, Shulman J. Dissecting oligogenic mechanisms of lysosomal susceptibility in Parkinson's disease. Presented at the AD/PD, 2024 International Conference on Alzheimer's and Parkinson's Diseases and related neurological disorders in Lisbon, Portugal (March 5-9, 2024).
2. Shulman J, Vanegas-Arroyave N, Fong J, Elsea S, Lin C, Saade H, et al. Precision neurology for dementia. Presented at the American Academy of Neurology (AAN), 75<sup>th</sup> Annual Meeting in Boston, Mass. (April 22-28, 2023).
3. Fong JC, Chavez F, Silos K, Castro GC, Arroyo-Miranda ML, Kunik ME, et al. "Hablemos de Alzheimer": Words Mexican Americans use to talk about Alzheimer's disease genetic testing. Implications for precision medicine education. Presented at the 2022 Alzheimer's Association International Conference (AAIC), San Diego, Calif. (July 30 - Aug. 4, 2022).
4. Li-Kroeger D, Al-Ramahi I, Marella A, Amoh BK, Smith N, Botas J, et al. A matrisomal proteomics network modulates tau and amyloid-beta toxicity in fly AD models. Presented at the 2022 Alzheimer's Association International Conference (AAIC), San Diego, Calif. (July 30 - Aug. 4, 2022).
5. Wu T, Guo C, Deger JM, Dhindsa J, Al-Ouran R, Shulman JM. Cell-specific responses to Tau pathology in the aging brain. Presented at the 2022 Alzheimer's Association International Conference (AAIC), San Diego, Calif.

- (July 30 - Aug. 4, 2022).
6. Ye H, Bagchi P, Wu T; Dammer EB, Seyfried NT, Shulman JM. Retromer subunit, VPS29, regulates vacuolar-type ATPase and lysosomal acidification in the aging brain. Presented at the 2022 Alzheimer's Association International Conference (AAIC), San Diego, Calif. (July 30 - Aug. 4, 2022).
  7. Lee TV, Allison K, Seyfried NT, Levey AI, Shulman JM. Functional validation of the cytoskeletal protein Plectin in Alzheimer's disease. Program No. 224.07. 2021 Neuroscience Meeting Planner. Society for Neuroscience, 2021. Online.
  8. Paveskovic M, Ojelade S, Pekarek B, Kochukov M, Arenkiel B, Shulman J. Alzheimer's disease risk gene CD2AP modulates synaptic structure and plasticity. Program No. 212.11. 2021 Neuroscience Meeting Planner. Society for Neuroscience, 2021. Online.
  9. Cary GA, Gockley J, Lehallier B, Leal K, Greenwood AK, Shulman JM, et al. Integrating multimodal data to support Alzheimer's disease target prioritization. Presented at the 2021 Alzheimer's Association International Conference (AAIC), Virtual Meeting (July 26-30, 2021).
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  11. Wu T, Guo C, Shulman JM. Elucidating cellular contributions to tau-mediated neurodegeneration using Drosophila and single cell transcriptomics. Presented at the 2021 Alzheimer's Association International Conference (AAIC), Virtual Meeting (July 26-30, 2021).
  12. Johnson ECB, Dammer EB, Duong D, Ping L, Zhou M, Yin L, et al. A consensus proteomic analysis of Alzheimer's disease brain and cerebrospinal fluid reveals early changes in energy metabolism associated with microglia and astrocyte activation. Presented at the 2020 Alzheimer's Association International Conference (AAIC), Virtual Meeting (July 27-31, 2020).
  13. Hill E, Deger J, Robak L, Rao S, Saade H, Ripperger B, et al. Addressing challenges of clinical comprehensive genetic testing in Parkinson's disease. *Neurology*. 2020;94(15 Suppl):5267.
  14. Johnson ECB, Dammer EB, Duong DM, Yin L, Betarbet R, Shulman JM, et al. A consensus Alzheimer's disease brain protein co-expression network identifies robust biological alterations associated with early- and late-stage disease. *Ann Neurol*. 2019;86(Suppl 24):S242.
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