

JACK HUMPHREY

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APPOINTMENTS

Departments of Neuroscience, Genetics and Genomic Sciences, Mount Sinai School of Medicine, New York

2025-present: Assistant Professor (research-track)

2021-2025: Instructor

EDUCATION AND TRAINING

Departments of Neuroscience, Genetics and Genomic Sciences, Mount Sinai School of Medicine, New York

2019-2021: Postdoctoral research fellow

Mentor: Prof Towfique Raj

Institute of Neurology & Genetics Institute, University College London, UK

2014-2019: PhD in Clinical Neuroscience

Thesis title: *RNA dysregulation in models of frontotemporal dementia and amyotrophic lateral sclerosis*

Mentors: Prof Adrian Isaacs, Dr Vincent Plagnol, Prof Pietro Fratta

Inivata, Cambridge, UK

January-March 2018: Computational Scientist, Technology Development

Stanford University, USA

February-May 2017: Visiting Student Researcher, Department of Genetics

Mentors: Prof Jonathan Pritchard, Dr Yang Li, Dr David A Knowles

University College London, UK

2010-2014: MSci Natural Sciences (4-year undergraduate degree); First class honours

Brain, Behaviour and Cognition (major) and Synthetic Organic Chemistry (minor)

SELECTED PUBLICATIONS

Number of publications & preprints: 50; Citations: 8,159; h-index: 30 (July 2026) - [Google Scholar](#)

Humphrey J, Brophy E, Kosoy R, Zeng B, Coccia E, Mattei D, Ravi A, Naito T, Efthymiou AG, Navarro E, De Sanctis C, Flores-Almazan V, Muller BZ, Snijders GJLJ, Allan A, Münch A, Kitata RB, Kleopoulos SP, Argyriou S, Malakates P, Psychogiou K, Shao Z, Francoeur N, Tsai CF, Gritsenko MA, Monroe ME, Paurus VL, Weitz KK, Shi T, Sebra R, Liu T, de Witte LD, Goate AM, Bennett DA, Haroutunian V, Hoffman GE, Fullard JF, Roussos P, Raj T. (2025) **Long-read RNA sequencing atlas of human microglia isoforms elucidates disease-associated genetic regulation of splicing.** *Nature Genetics* - **sole first author**

Humphrey J, Venkatesh S, Hasan R, Herb JT, de Paiva Lopes K, Küçükali F, Byrska-Bishop M, Evani US, Narzisi G, Fagegaltier D; NYGC ALS Consortium; Slegers K, Phatnani H, Knowles DA, Fratta P, Raj T. (2023) **Integrative transcriptomic analysis of the amyotrophic lateral sclerosis spinal cord implicates glial activation and suggests new risk genes.** *Nature Neuroscience* - **sole first author, co-corresponding**

Brown A, Wilkins OG, Keuss MJ, Hill SE, Zanollo M, Lee WC, Lee FCY, Masino L, Qi YA, Bryce-Smith S, Bampton A, Gatt A, Phatnani H, NYGC ALS Consortium, Schiavo G, Fisher EMC, Raj T, Secrier M, Lashley T, Ule J, Buratti E, **Humphrey J**, Ward ME, Fratta P (2022) **TDP-43 loss and ALS-risk SNPs drive mis-splicing and depletion of UNC13A.** *Nature* - **senior author**

Humphrey J, Oku A, Byrska-Bishop M, Basile AO, Evani US, Corvelo A, Tokolyi A, BP K, Réal A, Kim Y, Bond ML, Clarke WE, Fu R, Geiger H, Chang S, Naito T, Jang B, Musunuri R, Dredge WH, Al-Abri R, Hoover BN, Manaa D, McClintock J, Singh FP, Pedersen MH, Runnels A, Propp N, Fennessey S, Won HH, Zody MC, Narzisi G, Robine N, Lappalainen T, Fagegaltier D, Gürsoy G, Knowles DA, Raj T; NYGC ALS Consortium; Harms MB, Phatnani H. **The New York Genome Center ALS Consortium resource integrates postmortem tissue transcriptomics and whole genome sequencing to empower biological discovery.** *medRxiv* - **co-corresponding**

ACTIVE GRANT FUNDING

2026: Competing Team, Longitude Prize in ALS

Position: Consultant; Lead PI: Bernie Zipprich, Mosaic Neuroscience

2025-2028 BrightFocus Foundation Alzheimer's Disease Research Program

Identifying TDP-43 pathology burden in a mixed cohort of aging brains

Position: PI; Budget: \$300,000 over 3 years.

2025-2028 My Name's Doddie ALS Discovery Network

Single Cell Analysis of TDP-43 Dysfunction in ALS

Position: Co-PI; Budget: \$1.28M over 3 years between 3 groups (\$409k direct costs)

2025-2028 My Name's Doddie ALS Discovery Network / Target ALS Basic Biology Consortium

Investigating glial-immune crosstalk in modulating sporadic ALS survival in postmortem and longitudinal samples

Position: Co-PI; Budget: \$2.28M over 3 years between 4 groups (\$536k direct costs).

2025-2026 Robert Packard Center for ALS Research Pilot Grant Program

ALS patient-specific transcriptional networks and disease subtypes with multi-tissue factor analysis

Position: PI; Budget: \$75,000 over 1 year.

PREVIOUS FUNDING

2021-2026: NINDS U54 Center without Walls for Frontotemporal Dementia

Discovery of novel TDP-43 splicing targets: the Achilles heel for FTD and towards sensitive biomarkers and therapeutic targets;

Position: Member of Genomics Core; Budget: \$1.25M over 5 years

2023-2025 CurePSP Pipeline Grant

Understanding the roles of risk loci implicated in genome-wide association studies of PSP.

Position: Co-Principal Investigator; Budget: \$100,000 over 2 years.

2023-2024: ALS Association Seed Grant

Mapping the genetics of TDP-43 binding in ALS motor neurons

Position: Principal Investigator; Budget: \$50,000 over 1 year.

2022-2024: Mount Sinai Alzheimer's Disease Research Center (NIA P30-AG066514)

Developmental Project Funding Award

Assessing the genetic contribution of RNA editing within myeloid cells to Alzheimer's disease

Position: Principal Investigator; Budget: \$100,000 over 2 years.

SUBMITTED GRANTS

2026-2031 NIA R01 (Submitted June 2025, 37%; July 2026 resubmission)

Genetic regulation of TDP-43 RNA-protein interactions in ADRD

Position: PI

2027-2029 NINDS R21 (Submitted June 2026)

Mechanisms Linking circRNA Biology to TDP-43 Dysfunction in ALS/FTD

Position: Co-Investigator; PI: Christopher Donnelly, UTSW

2026-2031 NINDS R01 (Submitted Feb 2026; ND; October 2026 resubmission)

Identifying the Genetic Basis of ALS Through RNA and Chromatin Mechanisms

Position: MPI (Lead MPI: Yini Li, Purdue)

2026-2031 NIA R21 (Submitted October 2025; 28%; October 2026 resubmission)

The role of myeloid RNA editing in Alzheimer's disease genetics

Position: Principal Investigator

2026-2031 NINDS R01 (Submitted June 2025, ND; October 2026 resubmission)

Mapping the Molecular Cascade of ALS Progression Across the Motor Axis Using Single-Cell Multiomic Profiling

Position: Co-Investigator; PI: Veronique Belzil, Vanderbilt University Medical Center

2027-2029 ALS Finding a Cure (Submitted June 2026)

Identifying genetic modifiers of C9orf72

Position: Lead MPI (MPI: Andrew Sharp, MSSM)

SUPERVISION AND MENTORING

Current direct supervision in the Humphrey Lab:

Alice Maria Giani, Postdoctoral Fellow, funded by Target ALS / My Name's Doddie
Brooke Friedman, Graduate Student, funded by NIH T32
Faith Singh, Data Analyst, funded by the Packard Center / My Name's Doddie
Judy Liang, Data Analyst, funded by Longitude Prize

Previous supervision, Mount Sinai

All Master's students and analysts were primarily supervised by me in learning and applying computational methods. Graduate students were co-mentored by myself and Towfique Raj.
Irika Sinha, Postdoctoral Fellow (2026), now Postdoc at Allen Institute
Kailash Babu Panneerselvam, PhD Biomedical Sciences (2022-)
Winston Cuddleston, PhD Biomedical Sciences (2022-2024), now Postdoc, U Washington.
Rahat Hasan, MS Biomedical Data Science (2020-2022), now Bristol Myers-Squibb.
Hyomin Seo, MS Biomedical Data Science (2021-2023), Data Analyst (2023-2024), now Analyst, Mount Sinai.
Benjamin Muller, Data Analyst (2021-2023), now Informatics Scientist, Abbvie.
Ashvin Ravi, Data Analyst (2021-2023), now PhD student, UCSF.
Erica Brophy, Data Analyst (2021-2025), now Analyst at Memorial Sloan Kettering.
Alexander Chung, Collin Spencer, Jake Herb, Sanan Venkatesh, PhD rotations (2020-2024)
Abdullah Uddin, Volunteer (2021)

External Supervision

Ashely Harlock, BS, Graduate Student, Mount Sinai (thesis committee member)
Ali Oku, MS, Data Analyst, New York Genome Center (2022-present)
Anna-Leigh Brown, PhD, Pietro Fratta lab, University College London (2019-2023)
Oliver Ziff, PhD, Rickie Patani lab, University College London (2021-2023)
Gijsje Snijders, PhD, Lotje de Witte lab, Mount Sinai (2020-2023)
Callum Harvey, John Cooper-Knock lab, University of Sheffield (thesis examiner 2024)

TEACHING & OUTREACH

2023-present: *Core 2 Cellular and Molecular Neuroscience - "mRNA Splicing"*

2026-present: *Core 4 Molecular Pathogenesis of Neurological and Psychiatric Disorders - "ALS and FTD"*

I teach two lectures a year to Neuroscience graduate students at Mount Sinai. For the Core 2 lecture, I set an exam question which focuses on the design and application of RNA splicing therapeutics.

2014-2018: Taster Lecturer, UCL Widening Participation & UK Undergraduate Recruitment

I created and delivered a six-session course on *Introduction to Neuroscience* for a class of high school students from under-represented minority backgrounds, aged 17-18. I delivered the course each summer from 2015 to 2017. Additionally, I created a 45-minute interactive lecture on genetics entitled "Gene Detectives" and delivered it to groups aged 13 to 18 both in person and as a webinar from 2014-2018.

2016-2018: Placement host, In2ScienceUK

I took in high school students from under-represented minority backgrounds for a week of work experience in my office. I took them to meetings and taught them R programming and data visualisation.

2017: Instructor, Software Carpentry workshop in R programming, UCL

INDUSTRY EXPERIENCE

Scientific Advisory Board member, Mosaic Neuroscience (2026 - present)

Computational Scientist, Inivata, Cambridge (January – March 2018)

I worked with the Technical Development and Computational Genetics teams at Inivata, a cancer diagnostics startup. I was tasked with improving the design and analysis software of their circulating tumour DNA assay analytics. While there I developed tools to analyse and troubleshoot their historical sequencing data, which allowed me to re-design and extend one of their key products. While there I wrote extensive documentation and unit tests for the company's in-house software packages in R.

PEER REVIEW

Editorial Board Member, Molecular Neurodegeneration (2026-present).

Ad hoc reviewer: Nature Genetics, Nature Neuroscience, PLoS Genetics, The American Journal of Human Genetics, Nature Communications, Frontiers in Neurology, Scientific Data, Mammalian Genome, Molecular

Neurodegeneration, Neurobiology of Disease, and Brain Communications.

Grant reviewer: US Department of Defense, Association for Frontotemporal Dementia, ALS Canada/Brain Canada, ALS Association, Motor Neuron Disease Association (UK), and LiveLikeLou.

SOFTWARE

New York Genome Center ALS Consortium Browser (R/Shiny)

An interactive R/Shiny web app that allows users to explore and visualise gene expression associations in five post-mortem brain tissues. Website: <http://alsbrowser.nygenome.org/>

LeafCutter (R/Python; with Yang Li and David Knowles): A set of tools to perform differential splicing analyses without requiring a set of reference transcripts, allowing it to be used to find novel or cryptic splicing. I help maintain the project and assist our many users on Github. Project: github.com/davidaknowles/leafcutter

Leafviz & sQTLviz (R/Shiny): Interactive visualisation apps built with the R Shiny framework to visualise complex splicing changes between biological conditions (leafviz) or between genotypes (sQTLviz).

leafviz demo: leafcutter.shinyapps.io/leafviz code: github.com/jackhump/leafviz

sQTLviz demo: rajlab.shinyapps.io/sQTLviz_CMC code: github.com/jackhump/sQTLviz

CryptEx (Bash, AWK, R): Pipeline for novel splicing discovery in RNA-seq data. github.com/jackhump/CryptEx

INVITED TALKS (PAST 5 YEARS)

Aug 2026 *Neural Stem Cell Institute, Albany, NY*
May 2026 *Vanderbilt ALS Center, Vanderbilt University Medical Center, Nashville, TN*
Mar 2026 *Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA*
Mar 2026 *Weinberg ALS Center, Thomas Jefferson University, Philadelphia, PA*
Jan 2026 *Institute of Medical Sciences, University of Aberdeen, UK*
Dec 2025 *University Medical Center Utrecht, Netherlands*
Dec 2025 *Swammerdam Institute for Life Sciences, University of Amsterdam, Netherlands*
Dec 2025 *Centre for Molecular Neurology, VIB Antwerp, Belgium*
Nov 2025 *Dept. of Neuroscience, University of Montreal, Montreal, Canada*
Sep 2025 *Appel Forum on Neurodegeneration, Weill Cornell School of Medicine, NY*
Apr 2025 *Dept. Neuroscience, Baylor College of Medicine, Houston, TX*
Apr 2025 *St Jude Children's Research Hospital, Memphis, TN*
Mar 2025 *Dept. Molecular, Cellular, and Developmental Biology, University of Michigan, Ann Arbor, MI*
Feb 2025 *Dept. Neuroscience, Case Western Reserve University, Cleveland, OH*
Apr 2024 *Dept. Neurology, Vanderbilt University Medical Center, Nashville, TN*
Jan 2024 *Dept. Neurodegenerative Disease, Van Andel Institute, Grand Rapids, MI*
Nov 2023 *Dept. Genetics, Emory University, Atlanta, GA*
Nov 2023 *Center for Motor Neuron Biology & Disease, Columbia University Medical Center, NY*
Oct 2023 *Center for Human Genetics and Genomics, New York University Medical Center, NY*
Jul 2023 *Dept. Neuroscience, Mayo Clinic, Jacksonville, FL*
Jul 2023 *Center for Neurodegenerative Disease, Emory University, Atlanta, GA*
Feb 2023 *NIH Center for Alzheimer's and Related Dementias, Bethesda, MD*
Dec 2022 *UK Dementia Research Institute, King's College London, UK*
Sep 2021 *Laboratory of Neurogenetics, National Institutes of Health (virtual)*

CONFERENCE PRESENTATIONS

May 2026: Target ALS Annual Meeting, Boston, MA

Investigating Glial-Immune Crosstalk in Postmortem Samples

Mar 2026: Packard Center for ALS Research Annual Meeting, Baltimore, MD

Further adventures in TDP-43 cryptic splicing with single-cell and isoform approaches

Sep 2025: 2nd biennial conference on TDP-43 function and dysfunction in disease, Italy

Jul 2025: Gordon Research Conference on Amyotrophic Lateral Sclerosis, ME

Aug 2024: Gordon Research Conference on Neurobiology of Disease, Barcelona, Spain

Jul 2024: Live Like Lou ALS Research Symposium, Ann Arbor, MI

Functional genomics across neurodegenerative disease uncovers novel genes, pathways, and cell-types

May 2025, May 2026: Target ALS Annual Meeting, Boston, MA

Glial-immune Crosstalk in Modulating Sporadic ALS Survival

Dec 2024: André-Delambre ALS Research Symposium, Montreal, Canada

The New York Genome Center ALS Consortium combines postmortem tissue transcriptomics with whole genome sequencing to empower biological discovery

Nov 2024: American Society of Human Genetics, Denver, Colorado

The New York Genome Center ALS Consortium combines postmortem tissue transcriptomics with whole genome sequencing to empower biological discovery

Mar 2023: 17th International Conference on Alzheimer's and Parkinson's Diseases, Gothenburg, Sweden

Long-read RNA-seq of human microglia reveals novel isoforms and elucidates genetic regulation of splicing associated with Alzheimer's and Parkinson's disease

Oct 2022: American Society of Human Genetics, Los Angeles, USA

Invited session: Long-read RNA-seq to illuminate splice-driven mechanisms of human genetic diseases

Sep 2022: Emerging Scholars in Genome Sciences Symposium, University of Virginia, USA

Long-read RNA-seq of human microglia reveals novel isoforms and elucidates Alzheimer's-associated genetic regulation of splicing

Aug 2022: Gordon Research Conference on Neurobiology of Brain Disorders, Barcelona, Spain

Long-read RNA-seq of human microglia reveals novel isoforms and elucidates Alzheimer's-associated genetic regulation of splicing

Apr 2022: UCL Queen Square Motor Neuron Disease Centre Seminar, London

Genetic regulation and transcriptomic response to amyotrophic lateral sclerosis in the human spinal cord

Mar 2021: 15th International Conference on Alzheimer's and Parkinson's Diseases (virtual)

Atlas of genetic effects in human microglia transcriptome across brain regions, aging and disease pathologies

PUBLIC ENGAGEMENT

May 2026: Reporting the Research, Vanderbilt ALS Research Center

Invited speaker to present to ALS patients and carers.

Apr 2026: EverythingALS Expert Talk: Unlocking the Origins of ALS Through Large-Scale Patient Data

Online seminar given to a group of ALS patients and caregivers. Recorded and hosted on [YouTube](#).

May 2025: Featured in Target ALS ALS Awareness Month campaign

My contributions to ALS research in New York City were highlighted on LinkNYC billboards across the city during ALS Awareness Month. Details [here](#).

Sep 2024: The ALS Clinical Research Learning Institute, Johns Hopkins, Baltimore, MD

I presented my work to a panel of ALS patient activists.

AWARDS

- 2023** Reviewer's Choice Abstract Award, American Society of Human Genetics
- 2021** Junior Faculty Award, 15th International Conference on Alzheimer's and Parkinson's Diseases
- 2019** Healey Center GRC travel fellowship, GRC on ALS and Related Motor Neuron Diseases, VT
- 2016** UCL Bogue Fellowship, Funded visit to Stanford University, USA, as a visiting student researcher.
- 2012** DAAD RISE Scholar, Funded visit to the University of Giessen in Germany for undergraduate research

PROFESSIONAL AFFILIATIONS

I am a member of the **Deep Dementia Phenotyping (DEMON) Network**, organised by the UK Dementia Research Institute and Alzheimer's Research UK, and a member of the **American Society of Human Genetics**.

INTERESTS

Music: I play bass and guitar in occasional bands for weddings and parties.

Spanish: I spent time in summer 2012 and 2014 travelling in Spain, including a spell as a goat herder, to improve my conversational Spanish.

REFERENCES

Towfique Raj | Professor, Department of Neuroscience, Mount Sinai | *Postdoc mentor* | towfique.raj@mssm.edu
Alison Goate | Department Head, Department of Genetics and Genomic Sciences; Chair, Loeb Center for Alzheimer's Disease Research, Mount Sinai | *Head of Department* | alison.goate@mssm.edu
Pietro Fratta | Professor, Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology | *PhD co-supervisor* | p.fratta@ucl.ac.uk

FULL BIBLIOGRAPHY

 First or co-first author
  Corresponding or co-corresponding author
  Both

2026:

 Fodder K, Murthy M, de Silva R, Raj T, Farrell K, **Humphrey J**, Bettencourt C. (2026) **Cross-disease genetic and epigenetic architecture of the MOBP locus shows convergence in ALS-PSP.** *bioRxiv* - **co-corresponding author**

 **Humphrey J**, Oku A, Byrska-Bishop M, Basile AO, Evani US, Corvelo A, Tokolyi A, BP K, Réal A, Kim Y, Bond ML, Clarke WE, Fu R, Geiger H, Chang S, Naito T, Jang B, Musunuri R, Dredge WH, Al-Abri R, Hoover BN, Manaa D, McClintock J, Singh FP, Pedersen MH, Runnels A, Propp N, Fennessey S, Won HH, Zody MC, Narzisi G, Robine N, Lappalainen T, Fagegaltier D, Gürsoy G, Knowles DA, Raj T; NYGC ALS Consortium; Harms MB, Phatnani H. (2026) **The New York Genome Center ALS Consortium resource integrates postmortem tissue transcriptomics and whole genome sequencing to empower biological discovery.** *medRxiv* - **sole first; co-corresponding author**

Jang B, BP K, Tokolyi A, Dredge WH, Ravi A, Jung SH, Naito T, Kim B, Kim MS, Cho M, Park MS, Rosen M, Blanchard J, **Humphrey J**, Knowles DA, Won HH, Raj T. (2026) **A meta-analysis of single-nucleus expression quantitative trait loci linking genetic risk to brain disorders.** *Nature Genetics*.

Gatt A, Buhidma Y, Fodder K, **Humphrey J**, Foti SC, Frias B, Benson BC, Gami-Patel P, Gittings LM, Toomey CE, Lashley T. (2026) **Transcriptomic and pathological analysis of the hnRNP network reveals glial involvement in frontotemporal lobar degeneration pathological subtypes.** *Brain Communications*.

Sreeram A, Baron DM, Brusati A, Stallworth K, **Humphrey J**, Landers JE. (2026) **Global transcriptional changes across multiple isogenic C9orf72 patient iPSC-derived neurons.** *iScience*.

Yang A, Rodriguez M, Kozlenkov A, Vadukapuram R, Hurd Y, Dracheva S, **Humphrey J**, Breen MS. **Long-read transcriptomics of purified human cortical cell types exposes glial isoform complexity and disease-relevant transcript architecture.** *bioRxiv*

2025:

 **Humphrey J**, Brophy E, Kosoy R, Zeng B, Coccia E, Mattei D, Ravi A, Naito T, Efthymiou AG, Navarro E, De Sanctis C, Flores-Almazan V, Muller BZ, Snijders GJLJ, Allan A, Münch A, Kitata RB, Kleopoulos SP, Argyriou S, Malakates P, Psychogyiou K, Shao Z, Francoeur N, Tsai CF, Gritsenko MA, Monroe ME, Paurus VL, Weitz KK, Shi T, Sebra R, Liu T, de Witte LD, Goate AM, Bennett DA, Haroutunian V, Hoffman GE, Fullard JF, Roussos P, Raj T. (2025) **Long-read RNA sequencing atlas of human microglia isoforms elucidates disease-associated genetic regulation of splicing.** *Nature Genetics*. - **sole first author**

 Snijders GJLJ, de Paiva Lopes K, Sneeboer MAM, Muller BZ, Gigase FAJ, D'Urso D, Vialle RA, Missall R, Kubler R, Raj T, **Humphrey J**, de Witte LD. (2025) **The human microglia responsome: a resource for microglia states in health and disease.** *Brain Behav Immun*. - **co-corresponding author**

Bryce-Smith S, Brown AL, Mehta PR, Mattedi F, Mikheenko A, Barattucci S, Zanovello M, Dattilo D, Yome M, Hill SE, Qi YA, Wilkins OG, Sun K, Ryadnov E, Wan Y; NYGC ALS Consortium; Vargas JNS, Birsa N, Raj T, **Humphrey J**, Keuss M, Ward M, Secier M, Fratta P. (2024) **TDP-43 loss induces cryptic polyadenylation in ALS/FTD.** *Nature Neuroscience*

Kosoy R, Fullard JF, Bendl J, Kleopoulos SP, Shao Z, Argyriou S, Mathur D, Psychogyiou K, Malakates P, Vicari J, Ma Y, **Humphrey J**, Brophy E, Raj T, Katsel P, Voloudakis G, Lee D, Bennett DA, Haroutunian V, Hoffman GE, Roussos P. **Alzheimer's disease transcriptional landscape in ex vivo human microglia.** (2025) *Nature Neuroscience*.

Koromina M, Ravi A, Panagiotaropoulou G, Schilder BM, **Humphrey J**, ..., Ripke S, Raj T, Coleman JRI, Mullins N. (2024) **Fine-mapping genomic loci refines bipolar disorder risk genes.** (2025) *Nature Neuroscience* (author list shortened)

Kopell BH, Kaji DA, Liharska LE, ..., **Humphrey J**, ..., Beckmann ND, Charney AW. (2025) **A study of RNA splicing and protein expression in the living human brain.** *PLoS One* (author list shortened).

Rambarack N, Fodder K, Murthy M, Toomey C, de Silva R, Heutink P, **Humphrey J**, Raj T, Lashley T, Bettencourt C. **DNA methylation as a contributor to dysregulation of STX6 and other frontotemporal lobar degeneration genetic risk-associated loci.** *Acta Neuropathol Commun*.

Seo H, Cuddleston WH, Fu T, Navarro E, Parks M, Allan A, Efthymiou AG, Breen MS, Xiao X, Raj T, **Humphrey J** (2025) **Cytosine-to-uracil RNA editing is upregulated by pro-inflammatory stimulation of myeloid cells** *bioRxiv* - co-corresponding author

Dredge WH, BP K, Réal A, Muller BZ, Jang B, Fu T, Seo H, Won H, Breen MS, Xiao X, Knowles DA, **Humphrey J**, Raj T (2025) **Meta-analysis of genetic regulation of RNA editing in the human brain identifies new genes underlying neurological disease.** *medRxiv* - co-corresponding author

Réal A, BP K, Dredge WH, Muller BZ, Jang B, Tokolyi A, Won H, **Humphrey J**, Raj T, Knowles DA. (2025) **Mapping genetic effects on splicing in ten thousand post-mortem brain samples reveals novel mediators of neurological disease risk.** *medRxiv*

Lee C-Y, Ravi A, Huang T-C, Wyman B, Chriss A, **Humphrey J**, Knowles DA, Cotsapas C, Huckins L, Raj T, Brennand K (2025) **Regulatory landscape of Alzheimer's disease variants in human microglia.** *medRxiv*

2024:

Farrell K, **Humphrey J**, Chang T, Zhao Y, Leung YY, Kuksa PP, Patil V, Lee WP, Kuzma AB, Valladares O, Cantwell LB, Wang H, Ravi A, De Sanctis C, Han N, Christie TD, Afzal R, Kandoi S, Whitney K, Krassner MM, Ressler H, Kim S, Dangoor D, Iida MA, Casella A, Walker RH, Nirenberg MJ, Renton AE, Babrowicz B, Coppola G, Raj T, Höglinger GU, Müller U, Golbe LI, Morris HR, Hardy J, Revesz T, Warner TT, Jaunmuktane Z, Mok KY, Rademakers R, Dickson DW, Ross OA, Wang LS, Goate A, Schellenberg G, Geschwind DH; PSP Genetics Study Group; Crary JF, Naj A. **Genetic, transcriptomic, histological, and biochemical analysis of progressive supranuclear palsy implicates glial activation and novel risk genes.** (2024) *Nature Communications*.

Ressler HW, **Humphrey J**, Vialle RA, Babrowicz B, Kandoi S, Raj T, Dickson DW, Ertekin-Taner N, Crary JF, Farrell K. **MAPT haplotype-associated transcriptomic changes in progressive supranuclear palsy.** (2024) *Acta Neuropathologica Communications*.

Pasquini L, Pereira FL, Seddighi S, Zeng Y, Wei Y, Illán-Gala I, Vatsavayai SC, Friedberg A, Lee AJ, Brown JA, Spina S, Grinberg LT, Sirkis DW, Bonham LW, Yokoyama JS, Boxer AL, Kramer JH, Rosen HJ, **Humphrey J**, Gitler AD, Miller BL, Pollard KS, Ward ME, Seeley WW. (2024) **Frontotemporal lobar degeneration targets brain regions linked to expression of recently evolved genes.** *Brain*

Navarro E, Efthymiou AG, Parks M, Riboldi GM, Vialle RA, Udine E, Muller BZ, **Humphrey J**, Allan A, Argyrou CC, Lopes KP, Münch A, Raymond D, Sachdev R, Shanker VL, Miravite J, Katsnelson V, Leaver K, Frucht S, Bressman SB, Marcora E, Saunders-Pullman R, Goate A, Raj T. (2024) **LRRK2 G2019S variant is associated with transcriptional changes in Parkinson's disease human myeloid cells under proinflammatory environment.** *bioRxiv*

2023:

Humphrey J, Venkatesh S, Hasan R, Herb JT, de Paiva Lopes K, Küçükali F, Byrska-Bishop M, Evani US, Narzisi G, Fagegaltier D; NYGC ALS Consortium; Slegers K, Phatnani H, Knowles DA, Fratta P, Raj T. (2023) **Integrative transcriptomic analysis of the amyotrophic lateral sclerosis spinal cord implicates glial activation and suggests new risk genes.** *Nature Neuroscience*. - sole first author; co-corresponding author

Ziff OJ, Neeves J, Mitchell J, Tyzack G, Martinez-Ruiz C, Luisier R, Chakrabarti AM, McGranahan N, Litchfield K, Boulton SJ, Al-Chalabi A, Kelly G, **Humphrey J**, Patani R (2022) **Integrated transcriptome landscape of ALS identifies genome instability linked to TDP-43 pathology.** *Nature Communications* - senior author

Fodder K, Murthy M, Rizzu P, Toomey CE, Hasan R, **Humphrey J**, Raj T, Lunnon K, Mill J, Heutink P, Lashley T, Bettencourt C. **Brain DNA methylomic analysis of frontotemporal lobar degeneration reveals OTUD4 in shared dysregulated signatures across pathological subtypes.** *Acta Neuropathologica*.

Bettencourt C, Skene N, Bandres-Ciga S, Anderson E, Winchester LM, Foote IF, Schwartzentruber J, Botia JA, Nalls M, Singleton A, Schilder BM, **Humphrey J**, Marzi SJ, Toomey CE, Kleiflat AA, Harshfield EL, Garfield V, Sandor C, Keat S, Tamburin S, Frigerio CS, Lourida I; Deep Dementia Phenotyping (DEMON) Network; Ranson JM, Llewellyn DJ. **Artificial intelligence for dementia genetics and omics.** *Alzheimers & Dementia*

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2022:

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