

David A. Knowles

<https://daklab.github.io/>

Positions held

2019-present	CORE FACULTY MEMBER New York Genome Center
2025-present	ASSOCIATE PROFESSOR (Computer Science)
2019-2024	ASSISTANT PROFESSOR (Computer Science) AFFILIATE MEMBER (Data Science Institute) INTERDISCIPLINARY APPOINTEE (Systems Biology) Columbia University
2014-2018	POSTDOCTORAL RESEARCHER (Genetics, Pathology) Stanford University Co-advisors: Prof. Jonathan Pritchard, Prof. Sylvia Plevritis
2012-2014	POSTDOCTORAL RESEARCHER (Computer Science) Stanford University Advisor: Prof. Daphne Koller
2008-2012	PHD Candidate, Roger Needham Scholar, Wolfson College, University of Cambridge Machine Learning Group, Cambridge University Engineering Department
2006	Summer Undergraduate Research Fellow California Institute of Technology

Education

2008-2012	PHD Engineering (Machine Learning) University of Cambridge Thesis: <i>Bayesian non-parametric models and inference for sparse and hierarchical latent structure</i> Advisor: Prof. Zoubin Ghahramani
2007-2008	MSc Bioinformatics and Systems Biology - Distinction Imperial College London Thesis: <i>Statistical tools for ultra-deep pyrosequencing of fast evolving viruses.</i> Advisor: Prof. Susan Holmes, Statistics Department, Stanford University.
2003-2007	MENG Engineering - Distinction University of Cambridge Thesis: <i>A non-parametric extension to Independent Components Analysis.</i> Advisor: Prof. Zoubin Ghahramani. BA Natural Sciences (Physics) - First Class

Honours & Awards

2017	Stanford Cancer Systems Biology Symposium — Poster Award
2014	The International Society for Bayesian Statistics Travel Award for best invited Bayesian paper
2014	The International Society for Bayesian Statistics Dennis V. Lindley Prize for innovative research in Bayesian Statistics

2007	Charles Lamb University prize for first place in Information Engineering Sir Joseph Larmor Silver Plate and three other college prizes (Cargill, Cunningham and College)
2005	Wright Prize for ranking 5/600 in Natural Sciences Earle Year Prize for top 4 students across all subjects at St. John's College Hollinshead-Howles Prize for top Natural Scientist at St. John's College BP Prize for Advanced Physics
2004	Gaskell Year Prize for ranking 9/600 in Natural Sciences Hollinshead-Howles Prize for Part IA
2003	Top 50 nationally in Royal Society of Chemistry Olympiad
2001	Top 50 nationally in Mathematics Olympiad

Fellowships

2008-2012	Roger Needham Scholar, <i>Wolfson College, University of Cambridge</i> , funded by Microsoft Research
2006	Summer Undergraduate Research Fellow, <i>California Institute of Technology</i>

Industry Positions

2013-2014	Contract research scientist Ventana Roche Medical Systems <i>Deep learning/computer vision based prognosis of early stage breast cancer.</i>
2009-2012	Contract Software Engineer Microsoft Research Cambridge <i>Extending the probabilistic programming language Infer.NET.</i>
2009	Research Intern Microsoft Research Cambridge <i>Non-conjugate Variational Message Passing.</i> Supervisor: Thomas P. Minka.
2005	Equity Research Intern UBS Investment Bank, London
2004	Software Engineering Intern Data Connection Limited, London

Teaching

2019-ongoing	Machine learning in functional genomics						
	Course #	Semester	Enrollment	Class score		Instructor score	
				Mean	Median	Mean	Median
	COMS4995	Fall 2019	39	4.15	5.0	4.50	5.0
	COMS4995	Spring 2021	63	4.35	5.0	4.46	5.0
	COMS4762	Spring 2022	71	4.04	4.0	4.24	5.0
	COMS4762	Fall 2022	72	4.33	5.0	4.50	5.0
	COMS4762	Fall 2023	90	4.32	4.0	4.53	5.0
	COMS4762	Fall 2024	115	4.0	4.0	4.0	5.0
2022-ongoing	BMEN4500 Functional Genomics Methods & Applications guest lecturer						

2019	Columbia University Functional Genetics Boot Camp
2019	Weill Cornell Medical College Computational Genomics
2017	CBIO 244: Lecture Series in Cancer Systems Biology
2013-2015	Statistical, Mathematical, and Computational Consulting (SMACC) hosted by Stanford ICME
2013	Stanford Statistics guest lecture on Bayesian nonparametrics
2009-2011	Statistics advisor, Cambridge University Statistics Clinic
2009-2011	Supervisor, Cambridge University Engineering Department, <i>Digital Signal Processing</i>
2009-2011	Demonstrator, Cambridge University Engineering Department, <i>C++ programming</i>
2007-2009	Private Mathematics tutor, Camtutors

Mentoring

UNDERGRADUATES

2025-ongoing	Nicole Cui, Columbia University
2024-2025	Molly Durawa, Barnard College
Summer 2024	Sharvani Vadlamani, Columbia University
Summer 2024	Rohan Nidumbur, University of Maryland (co-mentored by Gao Wang, Columbia)
Summer 2024	Clara Yu, Caltech
Summer 2023	Fionna Feerick, Stevens Institute of Technology (NYGC/MacMillan diversity fellow)
2022-2024	Gilead Turok, Columbia University
2022-2024	Eric Kim, Columbia University
2022-2023	Ekene Ezeunala, Columbia University
2022-2023	Siddhant Sanghi, Columbia University (now a PhD student at UC Davis with Gerald Quon)
2022-2025	Gillian Bartels-Quansah (Science Path Scholar), Barnard College
2022-2024	Sophia Sowinski, Barnard College
Summer 2022	Mariya Semchuk (S Jay Levy diversity fellow), City College of New York
2021-2024	Stella Park, Columbia University
2021-2022	Daniel Um, Columbia University (now a Machine Learning Analyst at Bracebridge Capital)
2021-2022	Claire Johnson, Barnard (now a PhD student at Harvard Medical School with Eliezer Van Allen)
2019-2023	Collin Wang, Columbia University (now at Jane Street Capital)
2019-2020	Kevin Wang, Columbia University
2019-2021	Udai Nagpal, Columbia University (now at Goldman Sachs)

MASTERS STUDENTS

2025-ongoing	Nicholas Keung, Columbia University
2024-ongoing	Smriti Vaidyanathan, Columbia University
2024-2025	Shree Raghavendra, Columbia University
2023-ongoing	Julia Lewandowski, Columbia University
2023-ongoing	Zaikang Lin, Columbia University
2022-2024	Xingpei Zhang, Columbia University
2022-2024	Ting Chen, Columbia University
2022-2023	Joshua Segovia (DREAM Bridge student), Columbia University
2022-2023	Shouvik Mani, Columbia University with Elham Azizi (now a PhD student at Stanford University)
2021-2022	Hyun Dong Lee, Columbia University
2021-2022	Chloé Terwagne, Université Libre de Bruxelles (now a PhD student with Greg Findlay at The Francis Crick Institute, London).
2020-2021	Nasrine Metic, École polytechnique fédérale de Lausanne (now a PhD student with Mirjana Efre-mova at Barts Cancer Institute, London).
2019-2021	Stephen Malina, Columbia University (now Machine Learning Team Lead at Dyno Therapeutics).

PHD STUDENTS

2024-ongoing	Arghamitra Talukder, Computer Science, Columbia University (co-mentored by Itsik Pe'er)
2023-ongoing	Daniel Meyer, Computer Science, Columbia University
2022-ongoing	Sei Chang, Computer Science, Columbia University
2022-ongoing	Anjali Das, Computer Science, Columbia University
2021-ongoing	Jiayu Su, Systems Biology, Columbia University (co-mentored by Raúl Rabadán)
2020-ongoing	Shashaank Narayanan, Computer Science, Columbia University (co-mentored by David Sulzer)
2021-2025	Karin Isaev, Systems Biology, Columbia University. Now at Pioneer Labs.
2019-2025	Andrew Stirn, Computer Science, Columbia University. Now at Synthesize Bio.

POSTDOCTORAL SCHOLARS

2024-ongoing	Aaron Zweig, NYGC/Columbia (co-mentored by Elham Azizi, Columbia)
2024-ongoing	Alex Tokolyi, NYGC
2023-ongoing	Isabella Grabski, NYGC (co-mentored by Rahul Satija, NYU)
2023-ongoing	Tatsuhiko Naito, NYGC (co-mentored by Towfique Raj, MSSM)
2022-ongoing	Aline Réal, NYGC
2021-ongoing	Scott Adamson, NYGC (co-mentored by Tuuli Lappalainen)
2019-2025	Megan Schertzer, NYGC/Columbia. Now a Senior Staff Scientist at UVA.
2019-2024	Brielin Brown, DSI fellow, NYGC/Columbia (co-mentored by Tuuli Lappalainen). Now Assistant Professor at the University of Pennsylvania in the Department of Biostatistics, Epidemiology and Informatics.

PREDOCTORAL RESEARCH SCIENTISTS

2024-2025	Stella Park, NYGC
2022-2024	Claire Harbison, NYGC (joint with Tuuli Lappalainen)
2021-2024	Jui-Shan (Teresa) Lin, NYGC
2021-2022	Peter Halmos, NYGC (now a PhD student with Ben Raphael at Princeton).

POSTDOCTORAL RESEARCH SCIENTISTS

2023-ongoing	Saikat Banerjee, PhD, NYGC
2020-ongoing	Chirag Lakhani, PhD, NYGC
2019-2021	Laura Pereira, PhD, NYGC (now Team Lead at Spark Therapeutics).

VISITING SCIENTISTS

2023-2024	Giacomo Cavalca, PhD student at University of Bologna. Co-mentored with Dr Gao Wang, Columbia Neurology.
Summer 2023	Aditya Sriram, PhD student at University of Pittsburgh.

Service

TO THE DEPARTMENT

2023, 2024	Co-organized CS PhD visit
2022-2024	Distributed Research Apprenticeships for Master's (DREAM) MS-to-PhD Bridge program mentor
2022	Organized junior faculty session at annual CS faculty retreat
2021-ongoing	MS advisor for ML track

TO THE SCHOOL OF ENGINEERING

2021 Reviewed for Columbia Precision Medicine Initiative Pilot Grants Program
2021-ongoing Reviewed for Columbia Engineering SIRS/STAR Funding

TO THE UNIVERSITY

2023-ongoing Advisor for Computational Biology BA.
2021-2023 Created the new Computational Biology BA degree (w/ Itsik Pe'er): developed curriculum, partnered with Biological Sciences, and advocated to the University.
2021 DSI molecular tumor board representative
2021-ongoing Reviewing for Data Science Institute MS applicants
2020 DSI capstone project advisor (with Johnson & Johnson)

To NYGC

2025-ongoing AI Taskforce Co-chair
2022 CSHL - NYGC faculty retreat planning committee
2022-ongoing Statistical and population genetics working group co-chair
2022-ongoing NYGC DEI summer internship program mentor & speaker
2021-ongoing Faculty representative on academic DEI committee
2021 Faculty recruitment committee co-chair
2020-ongoing Weekly NYGC-wide lab meeting organizer
2020-ongoing Annual postdoc & student award external judge organizer
2021 AI Applications to Disease Research/Fundamentals of AI Workshop

TO THE PROFESSION

2023-ongoing Co-chair of the annual Machine Learning in Computational Biology (MLCB) meeting mlcb.org/
2020-2021 Machine Learning in Computational and Systems Biology (MLCSB) Communities of Special Interest (COSI) board member, Organization: International Society for Computational Biology (ISCB)
2020-2022 Co-organizer of MLCB.

PHD COMMITTEES

2025-ongoing Feiyue Yang, Systems Biology, Columbia University
2024-ongoing Philippe Chlenski, Computer Science, Columbia University
2024-2025 Yining Liu, Computer Science, Columbia University
2024-ongoing Charlotte Rochereau, Systems Biology, Columbia University
2024-ongoing Christina Floristean, Computer Science, Columbia University
2024-ongoing Justin Hong, Computer Science, Columbia University
2024-ongoing Fion Shiau, Systems Biology, Columbia University
2023-ongoing Yige Zhao, Systems Biology, Columbia University
2024-2025 Achille Nazaret, Computer Science, Columbia University
2022-2024 Sydney Hart, New York University
2022-2024 Sitara Persad, Computer Science, Columbia University
2022-2023 Erica Wolin, Biological Sciences, Columbia University
2021-ongoing Izaak Coleman, Systems Biology, Columbia University
2022-ongoing Mert Ketenci, Computer Science, Columbia University
2021-2024 Guojie Zhong, Systems Biology, Columbia University
2021-2022 Jonah Einson, Systems Biology, Columbia University
2021-2024 Raiyan Rashid Khan, Computer Science, Columbia University
2020-2022 Daniel Li, Computer Science, Columbia University

2019-2022 Akash Sookdeo, New York University
 2019-2022 Neville Dusaj, Weill Cornell Medical Campus
 2020-2021 Jie Yuan, Computer Science, Columbia University
 2020-2021 Antonio Khalil Moretti, Computer Science, Columbia University
 2020 External Examiner for Clint Lombard, Electrical and Electronic Engineering, Stellenbosch University, South Africa.
 2020 Hooshmand Shokri Razaghi, Computer Science, Columbia University
 2019-2020 Hanna Levitin, Systems Biology, Columbia University
 2019 Gabriel Loaiza, Statistics, Columbia University

POSTDOCTORAL FELLOWSHIP & TRANSITION AWARD COMMITTEES

2025-ongoing Amanda Wilson, Landweber lab, Columbia (F32)
 2024-ongoing Iraj Eshghi, Imielinski lab, New York Genome Center (BWF CASI)
 2024-ongoing Mariela Cortes Lopez, Landau lab, New York Genome Center (K99)
 2021-2023 John Morris, Lappalainen lab, New York Genome Center (K99)
 2022-2024 Adam Widman, Landau lab, New York Genome Center
 2020-2022 Tim Stuart, Satija lab, New York Genome Center (K99)

Reviewing

2023 Poster judge for 1st NYC RNA Symposium at Rockefeller University
 2017 Advances in Neural Information Processing Systems (NeurIPS) Best Reviewer Award

JOURNALS

Nature, Science, Cell, Nature Genetics, Nature Communications, Nature Bioengineering, Nature Machine Intelligence, eLife, PLOS Genetics, PLOS Computational Biology, Bioinformatics, BMC Bioinformatics, npj Genomic Medicine, Transactions on Machine Learning Research, Science Advances, Genome Research, Genome Biology, Biological Psychiatry, Journal of Mathematical Biology, Journal of Machine Learning Research, Journal of Computational and Graphical Statistics, Journal of the Royal Statistical Society: Series B, IEEE Transactions on Pattern Analysis and Machine Intelligence, Annals of Applied Statistics. Machine Learning, Journal of the American Statistical Association, Statistics and Computing (Springer), Bayesian Analysis.

CONFERENCES

Advances in Neural Information Processing Systems (NeurIPS), International Conference on Machine Learning (ICML), International Conference on Artificial Intelligence and Statistics (AISTATS), Pacific Biocomputing Symposium (PBS), International Joint Conferences on Artificial Intelligence (IJCAI), International Conference on Learning Representations (ICLR), Intelligent Systems for Molecular Biology.

WORKSHOPS

PC member for NeurIPS Workshop on Computational Biology, PC member for NeurIPS Workshop on Optimization in Machine Learning

GRANT REVIEWING

2025 NIH Genomics, Computational Biology and Technology study section
 2025 Oak Ridge Associated Universities Ralph E Powe Junior Faculty Enhancement Awards

2022 NIH:NHGRI Special Emphasis Panel for Supporting Talented Early Career Researchers in Genomics

2022 Ad-hoc reviewer for NSF Directorate for Biological Sciences/Molecular and Cellular Biosciences Division/Systems and Synthetic Biology Team

2021 NIH/NIA special emphasis panel for PAR-19-269 “Cognitive Systems Analysis of Alzheimer’s Disease Genetic and Phenotypic Data”.

2021 NIH Director’s Early Independence Awards (DP5) Subject Matter Expert.

2020 US-Israel Binational Science Foundation reviewer.

TENURE AND PROMOTION REVIEWING

2023 Technical University of Denmark

2022 Institut Pasteur

2022 Rockefeller University

PROFESSIONAL AFFILIATIONS

Organizer for Machine Learning in Computational Biology (MLCB) 2019-2022, MLCB Co-chair 2023-ongoing, ISMB MLCSB COSI member, Early Stage Investigators in Cancer Systems Biology Steering Committee, International Society of Bayesian Analysis (ISBA) Chair of the Continuing Education Committee.

Selected Talks

2025 UCLA Computer Genomics Summer Institute invited tutorial

2025 Yale RNA AI meeting invited speaker

2025 Alzheimer’s disease Gordon Research Conference invited speaker

2025 Systems Biology session, Probabilistic Genomics (CSHL), co-chair and speaker

2025 Stony Brook Single Cell Genomics invited speaker

2024 Pacific Biosciences PRISM Boston invited speaker

2024 UCLA Computational Genomics Summer Institute

2024 Generative AI in Life Science, Copenhagen, invited speaker

2024 Brown/Lifespan Center for Clinical Cancer Informatics and Data Science

2024 Memorial Sloan Kettering Cancer Center csBio Seminar, invited speaker

2024 NYU Institute for Systems Genetics Nano Seminar, invited speaker

2023 MIT Bioinformatics Seminar Series

2023 Engineering in Medicine Symposium, Columbia University

2023 Quantitative Biology Seminar Series, Cold Spring Harbor Laboratories

2023 Genetics and Genomic Sciences External Speaker Seminar series, Mount Sinai School of Medicine

2022 Computational Genomics Summer Institute, UCLA

2022 Simons Institute *From Algorithms to Discovery* Conference

2021 Machine Learning in Computational Biology seminar series, University of Washington

2020 Mount Sinai Medical School Neurogenomics seminar series

2020 Einstein Developmental and Molecular Biology seminar series

2020 New York Stem Cell Foundation Invited Speaker

2020 NIH Single Cell Genomics Hackathon Invited Speaker

2019 Washington University Department of Genetics invited speaker

2019 NYC Human Genetics meeting invited speaker

2019 UCLA Computational Genomics Summer Institute

2018 UCLA Computational Genomics Winter Institute

2017 American Society of Human Genetics Annual Meeting

2017	Statistical and Computational Challenges in Large Scale Molecular Biology at the Banff International Research Station
2015	Highlights from Bayesian Analysis (Joint Statistical Meeting session), invited speaker
2015	Statistical and Computational Challenges In Bridging Functional Genomics, Epigenomics, Molecular QTLs, and Disease Genetics at the Banff International Research Station
2015	The Biology of Genomes meeting at Cold Spring Harbor Laboratory
2014	NeurIPS Variational Inference Workshop invited speaker
2013	RECOMB/ISCB Conference on Regulatory & Systems Genomics
2012	Stanford Biostatistics Workshop
2012	Collegio Carlo Alberto Statistics Seminar
2011	Cambridge Statistics Initiative One Day Special Meeting
2011	27th Conference on Uncertainty in Artificial Intelligence (UAI)
2011	NeurIPS Workshop on Predictive Models in Personalized Medicine
2011	Eighth Workshop on Bayesian Nonparametrics; Veracruz, Mexico
2009	NeurIPS Workshop on Computational Biology

Grants awarded

2024	The genomic and synaptic basis of language disorder and learned sound association. SIRS Blavatnik fund. Role: lead PI. Co-I: David Sulzer (Psychiatry, Neurology, Pharmacology).
2023-2026	Inferring cell-type specific gene regulatory and disease causal networks underlying cancer progression and drug resistance. Funding body: MacMillan Center for the Study of the Non-Coding Cancer Genome. Role: Lead principal investigator (PI). Co-I: Elham Azizi, Biomedical Engineering, Columbia University.
2024-2025	A deep learning based approach for phenome-wide association studies. Columbia Precision Medicine Pilot Award. Role: Co-I. Lead PI: Gamze Gürsoy (Columbia).
2022-2025	CAREER: Unifying short and long read RNA-seq analysis of alternative splicing using network flow models. Award number: DBI2146398. Funding body: National Science Foundation Biological Informatics program. Role: PI.
2022-2027	Delineating the network effects of mental disorder-associated variants using convex optimization methods. Award number: R01MH130879. Funding body: National Institute of Mental Health. Role: Lead PI. Co-PI: Niamh Mullins, Mount Sinai School of Medicine.
2022-2024	A CRISPR/Cas13 approach for identifying individual transcript isoform function in cancer. Award number: R21CA272345. Funding body: National Cancer Institute. Role: Lead PI. Co-I: Neville Sanjana, New York University.
2021-2022	Identifying genetic and transcriptomic drivers of Parkinson's disease progression. Award number: U01NS120256. Funding body: National Institute of Neurological Disorders and Stroke. Role: Co-Investigator.
2021-2023	Resolving complex alternative splicing of psychiatric disease genes using single-cell approaches. R01MH125579. Funding body: National Institute of Mental Health. Role: Co-I. Multi-PIs: Gang Fang (MSSM), Kristen Brennand (Yale).
2021-2023	Fine-mapping psychiatric disease variants that affect post-transcriptional gene regulation. Award number: R56MH127844. Funding body: National Institute of Mental Health. Role: Lead PI. With Kristen Brennand (Co-I, Yale) and William Fairbrother (Co-I, Brown).
2020-2025	Learning the Regulatory Code of Alzheimer's Disease Genomes. Award number: U01AG068880. Funding body: National Institute on Aging. Role: Multiple PI. With Towfique Raj, Mount Sinai School of Medicine.

GRANTS AWARDED TO TRAINEES

- 2025 **Geometric modeling of perturbed gene regulation dynamics across time and space.** Damon Runyon Quantitative Biology Fellowship Award. Trainee: Aaron Zweig.
- 2024 **A probabilistic framework for deconvolving causal mechanisms of cancer therapeutics with genetic perturbation screens.** Damon Runyon Quantitative Biology Fellowship Award. Trainee: Isabella Grabski.
- 2024 **The genomic and synaptic basis of language disorder and learned sound association.** NIH/NIDCD. Award number: F31DC022183. Trainee: Shashaank Narayanan.
- 2022 **Perturbation-response approaches to determining the regulatory networks underlying human complex traits.** NIH/NHGRI. Award number: K99-HG012373. Trainee: Brielin Brown (postdoc).
- 2021 **Measuring allele and isoform-specific RBP binding to improve predictive models of RNA splicing.** NIH/NIGMS. Award number: F32GM142213. Trainee: Megan Schertzer (postdoc).
- NSF Graduate Research Fellowships awarded to Udai Nagpal, Sei Chang, and Shashaank Narayanan. CSGrad4US fellowships awarded to Daniel Meyer and Arghamitra Talukder.

Publications

I have published in two fields, machine learning and genetics. For both, the first author(s) contributed most to the work, and the senior authors appear last. In machine learning, proceedings for the top conferences (i.e., NeurIPS, ICML, AISTATS) are as prestigious as any journal, whereas in genetics conference abstracts are not considered formal publications.

Total citations: **12391**. *h*-index: **39** (retrieved from Google scholar, 8/17/2025).

Lab members are highlighted in **bold**.

Under submission

- [1] **Saikat Banerjee**, Shane O’Connell, Sarah M C Colbert, Niamh Mullins, and **David A Knowles**. Convex approaches to isolate the shared and distinct genetic structures of subphenotypes in heterogeneous complex traits. *medRxiv* (2025).
- [2] Lindsay A Becker, Sofia A Quinodoz, Troy J Comi, Ofer Kimchi, **David A Knowles**[†], and Clifford P Brangwynne[†]. Genome-wide mapping of mesoscale neuronal RNA organization and condensation. *bioRxiv* (2025). [†]Co-corresponding.
- [3] Beomjin Jang, Kailash Bp, **Alex Tokolyi**, Winston H Cuddleston, Ashvin Ravi, Sang-Hyuk Jung, **Tatsuhiko Naito**, Beomsu Kim, Min Seo Kim, Minyoung Cho, Mi-So Park, Mikaela Rosen, Joel Blanchard, Jack Humphrey, **David A Knowles**, Hong-Hee Won, and Towfique Raj. SingleBrain: A meta-analysis of single-nucleus eQTLs linking genetic risk to brain disorders. *medRxiv* (2025).
- [4] **Chirag M Lakhani**, Jui-Shan T Lin, Anjali Das, **Tatsuhiko Naito**, Towfique Raj, and **David A Knowles**. Integration of deep learning annotations with functional genomics improves identification of causal Alzheimer’s disease variants. *medRxiv* (2025).
- [5] **Zaikang Lin**, Sei Chang, Aaron Zweig, Minseo Kang, Elham Azizi, and **David A Knowles**. Interpretable neural ODEs for gene regulatory network discovery under perturbations. *arXiv [cs.LG]* (2025).
- [6] Lingjun Meng, Alice Yu, Huanwei Huang, Jodie Meng, Yongjin Yoo, Katie Schaukowitch, Justyna A Janas, and **David A Knowles**. Oncogenic mutations disrupt lineage differentiation via epigenetic perturbations. *bioRxiv* (2025).

- [7] Alan M Moses, Jason E Stajich, Audrey P Gasch, and **David A Knowles**. Inferring fungal cis-regulatory networks from genome sequences. *bioRxiv* (2025).
- [8] **Tatsuhiko Naito**, Kosei Hirata, Beomjin Jang, **Chirag M Lakhani**, Alice Buonfiglioli, Wan-Ping Lee, Otto Valladares, Li-San Wang, Yukinori Okada, Hong-Hee Won, Eirini P Pappetrou, Samuele G Marro, **David A Knowles**, and Towfique Raj. Mosaic chromosomal alterations in blood are associated with an increased risk of Alzheimer’s disease. *medRxiv* (2025).
- [9] **Megan D Schertzer**, **Stella H Park**, **Jiayu Su**, Gloria M Sheynkman, and **David A Knowles**. Perplexity as a metric for isoform diversity in the human transcriptome. *bioRxiv* (2025).
- [10] **Jiayu Su**, Yiming Qu, **Megan Schertzer**, Haochen Yang, Jiahao Jiang, Tenzin Lhakhang, Theodore M Nelson, **Stella Park**, Qiliang Lai, Xi Fu, Seung-Won Choi, **David A Knowles**[†], and Raul Rabadan[†]. A computational framework for mapping isoform landscape and regulatory mechanisms from spatial transcriptomics data. *bioRxiv* (2025).
- [11] **Aaron Zweig**, **Zaikang Lin**, Elham Azizi, and **David A. Knowles**. Towards identifiability of interventional stochastic differential equations. *arXiv [cs.LG]* (2025).

Journal articles (genetics)

- [12] **Brielin C Brown**, John A Morris, Tuuli Lappalainen, and **David A Knowles**. Large-scale causal discovery using interventional data sheds light on the regulatory network architecture of blood traits. *To appear in Nature Communications* (2025).
- [13] **Anjali Das**, **Chirag Lakhani**, **Chloé Terwagne**, **Jui-Shan T Lin**, **Tatsuhiko Naito**, Towfique Raj, and **David A Knowles**. Leveraging functional annotations to map rare variants associated with Alzheimer’s disease with gruyere. *American Journal of Human Genetics* (2025).
- [14] Michael B Fernando, Yu Fan, Yanchun Zhang, **Alex Tokolyi**, Aleta N Murphy, Sarah Kam-mourh, P J Michael Deans, Sadaf Ghorbani, Ryan Onatzevitch, Adriana Pero, Christopher Padilla, Sarah Williams, Erin K Flaherty, Iya A Prytkova, Lei Cao, **David A Knowles**, Gang Fang, Paul A Slesinger, and Kristen J Brennand. Phenotypic complexities of rare heterozygous neurexin-1 deletions. *Nature* (2025).
- [15] **Megan D Schertzer**, **Andrew Stirn**, **Keren Isaev**, **Laura Pereira**, **Anjali Das**, **Claire Harbison**, **Stella H Park**, Hans-Hermann Wessels, Neville E Sanjana, and **David A Knowles**. Cas13d-mediated isoform-specific RNA knockdown with a unified computational and experimental toolbox. *Nature Communications* (2025).
- [16] Markus Terrey, Georgii Krivoshein, **Scott I Adamson**, Elena Arystarkhova, Laura Anderson, John Szvec, Shelby McKee, Holly Jones, Sara Perkins, Vijay Selvam, Pierre-Alexandre Picc, Dweet Chhaya, Ari Dehn, Aamir Zuberi, Stephen A Murray, Natalia S Morsci, Kathleen J Sweadner, **David A Knowles**, Else A Tolner, Arn M J M van den Maagdenberg, and Cathleen M Lutz. Alternating hemiplegia of childhood associated mutations in Atp1a3 reveal diverse neurological alterations in mice. *Neurobiology of disease* (2025).
- [17] Shane O’Connell, **Brielin C Brown**, Dara M Cannon, Pilib Ó Broin, **David A Knowles**, Nadine Parker, Dag Alnæs, Lars T Westlye, **Saikat Banerjee**, Leila Nabulsi, Emma Corley, Ole A Andreassen, and Niamh Mullins. Deriving Mendelian Randomization-based causal networks of brain imaging phenotypes and bipolar disorder. *Biological Psychiatry: Cognitive Neuroscience and Neuroimaging* (2024).
- [18] Cameron Y Park*, **Shouvik Mani***, Nicolas Beltran-Velez, Katie Maurer, Teddy Huang, Shuqiang Li, Satyen Gohil, Kenneth J Livak, **David A Knowles**, Catherine J Wu, and Elham Azizi. A Bayesian framework for inferring dynamic intercellular interactions from time-series single-cell data. *Genome research* (2024). *Equal contribution.

- [19] **Brielin C Brown**[†], **Collin Wang**, Silva Kasela, François Aguet, Daniel C Nachun, Kent D Taylor, Russell P Tracy, Peter Durda, Yongmei Liu, W Craig Johnson, David Van Den Berg, Namrata Gupta, Stacy Gabriel, Joshua D Smith, Robert Gerzsten, Clary Clish, Quenna Wong, George Papanicolaou, Thomas W Blackwell, Jerome I Rotter, Stephen S Rich, R Graham Barr, Kristin G Ardlie, **David A Knowles**[†], and Tuuli Lappalainen[†]. Multiset correlation and factor analysis enables exploration of multi-omics data. *Cell Genomics* (2023). [†]Co-corresponding.
- [20] Mariela Cortés-López, Paulina Chamely, Allegra G Hawkins, Robert F Stanley, Ariel D Swett, Saravanan Ganesan, Tarek H Mouhieddine, Xiaoguang Dai, Lloyd Kluegel, Celine Chen, Kiran Batta, Nili Furer, Rahul S Vedula, John Beaulaurier, Alexander W Drong, Scott Hickey, Neville Dusaj, Gavriel Mullokandov, Adam M Stasiw, **Jiayu Su**, Ronan Chaligné, Sissel Juul, Eoghan Harrington, **David A Knowles**, Catherine J Potenski, Daniel H Wiseman, Amos Tanay, Liran Shlush, Robert C Lindsley, Irene M Ghobrial, Justin Taylor, Omar Abdel-Wahab, Federico Gaiti, and Dan A Landau. Single-cell multi-omics defines the cell-type-specific impact of splicing aberrations in human hematopoietic clonal outgrowths. *Cell stem cell* (2023).
- [21] **Shashaank Narayanan**, Mahalakshmi Somayaji, Mattia Miotto, Eugene V Mosharov, Emily A Makowicz, **David A Knowles**, Giancarlo Ruocco, and David L Sulzer. Computational models of dopamine release measured by fast scan cyclic voltammetry in vivo. *PNAS nexus* (2023).
- [22] **Jiayu Su**, Jean-Baptiste Reynier, Xi Fu, Guojie Zhong, Jiahao Jiang, Rydberg Supo Escalante, Yiping Wang, Luis Aparicio, Benjamin Izar, **David A Knowles**, and Raul Rabadan. Smoother: a unified and modular framework for incorporating structural dependency in spatial omics data. *Genome biology* (2023).
- [23] Julia J Wattacheril, Srilakshmi Raj, **David A Knowles**, and John M Greally. Using epigenomics to understand cellular responses to environmental influences in diseases. *PLoS genetics* (2023).
- [24] Rockwell J Weiner, **Chirag Lakhani**, **David A Knowles**, and Gamze Gürsoy. LDmat: efficiently queryable compression of linkage disequilibrium matrices. *Bioinformatics* (2023).
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