



BRIAN M SCHILDER, PHD

Passionately pursuing transdisciplinary research to advance human health and knowledge.



Cold Spring Harbor Laboratory

Postdoctoral Research Scientist

Below are selected subsets of the [full CV](#). -



EDUCATION

2024



Imperial College London / The Alan Turing Institute

PhD; Computational Genomics & Machine Learning

📍 [London, UK](#)

Thesis: Multi-omic medicine: dissecting the cell type-specific and pleiotropic mechanisms underlying disease genomics at scale

2017



The George Washington University / Georgetown University

MPhil; Comparative Neuroscience & Genomics

📍 [Washington, DC, USA](#)

Thesis: The evolution of the hippocampus and adult neurogenesis: novel insights into the origins of human memory

2011



Brown University / Princeton University

ScB; Neurological Diseases & Disorders

📍 [Providence, RI, USA](#)



CORE SKILLS

Research

15+ years of deep expertise in genomics, AI, evolutionary biology and biomedicine. Strategically fuses concepts and methods across multiple domains.

- **Publication record:** **23** publications, **8** preprints and **13** awarded grants.
- **Reproducibility:** Global leader in promoting and enabling reproducible scientific practices. 📄 Writes 100% reproducible manuscripts programmatically. 📄
- **Bioinformatics:** Created **45** Python and R packages to address key challenges in biological research.
- **High-performance computing:** Highly parallelised analyses and AI model training (CPUs and GPUs).
- **Web development:** **6+** websites, web apps, and interactive reports.

CONTACT

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🌐 [LinkedIn](#)

🆔 [ORCID](#)

🔍 [Google Scholar](#)

🐙 [GitHub](#)

🐦 [Twitter](#)

📺 [YouTube](#)

🌐 [Personal Website](#)

🌐 [Lab Website](#)

SUMMARY

📅 **15+ years of research**

📄 **23 publications**

📄 **8 preprints**

🔧 **41 software packages**

📦 **11 databases & apps**

🗣️ **23 talks**

👤 **13+ years of teaching & team management**

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AI & Machine Learning

Proficient in developing and deploying AI/ML models (PyTorch, tensorflow, Keras, sklearn and H2O) to solve complex biological problems. Applied examples include:

- **Causal variant effect prediction:** Used functional impact predictions from DNA sequence models (DeepSEA, Basenji, IMPACT) to validate SNPs prioritised with Bayesian fine-mapping. 
- **Foundation models:** Used transformer trained on >36M cells and protein sequence embeddings to uncover cell type-specific mechanisms of disease.
- **LLM knowledge extraction:** Developed framework to extract quantitative metrics of phenotype severity from GPT-4. 
- **Disease genomics embeddings:** Developed VAE/graph models to reveal joint latent representation of genomic signatures across all diseases and phenotypes.
- **NLP:** Created a suite of proprietary Python packages for advanced topic modelling of the PubMed literature to provide business intelligence to the world's largest digital health, biotech, and pharma companies (as a consultant with [120/80 Group](#)).
- **Tensor decomposition:** Applied multi-condition factorisation to efficiently discover neurodegeneration-relevant *trans*-eQTLs 

Project Management

Efficient management strategies to define objectives, track progress and coordinate diverse teams.

- **Documentation:** Defines objectives and tracks progress with GitHub Projects. Includes useful documentation in Issues, inline code and shareable reports.
- **Version control:** Extensive and daily use of GitHub, containers (*Docker*, *Singularity*, *virtual machines*), environments (*conda*) and pipelines (*Nextflow*).
- **Team management:** Led numerous collaborative research projects and supervised researchers at various career stages.

Soft Skills

Advances science through effective problem formulation, collaboration and communication.





- **Problem formulation:** Rapid hypothesis generation, project design, and creative problem solving.
- **Communication:** Clear and concise distillation of complex results to a variety of audiences. Presented **25** conference posters.
- **Collaboration:** Diverse and global collaborative networking.



PUBLICATIONS

- 2023 • **rworkflows: automating reproducible practices for the R community**
Nature Communications (2023) 15(149); <https://doi.org/10.1038/s41467-023-44484-5>
BM Schilder, AE Murphy, NG Skene
 **News**
- Featured in *Nature Communications* Editors' Highlights
- 2023 • **Artificial intelligence for neurodegenerative experimental models**
Alzheimer's & Dementia (2023) <http://doi.org/10.1002/alz.13479>
SJ Marzi, **BM Schilder**, A Nott, C Sala Frigerio, S Willaime-Morawek, M Bucholc, DP Hanger, C James, PA Lewis, I Lourida, W Noble, F Rodriguez-Algarra, JA Sharif, M Tsalenchuk, LM Winchester, U Yaman, Z Yao, DEMON Network, JM Ranson, DJ Llewellyn
- 2023 • **Artificial intelligence for dementia genetics and omics**
Alzheimer's & Dementia (2023) <http://doi.org/10.1002/alz.13427>
C Bettencourt, NG Skene, S Bandres-Ciga, E Anderson, LM Winchester, IF Foote, J Schwartzentruber, JA Botia, M Nalls, A Singleton, **BM Schilder**, J Humphrey, SJ Marzi, CE Toomey, A Al Kleifat, EL Harshfield, V Garfield, C Sandor, S Keat, S Tamburin, C Sala Frigerio, I Lourida, DEMON Network, JM Ranson, DJ Llewellyn
- 2023 • **Artificial intelligence for dementia research methods optimization**
Alzheimer's & Dementia (2023) <http://doi.org/10.1002/alz.13441>
M Bucholc, C James, A Al Kleifat, A Badhwar, N Clarke, A Dehsarvi, CR Madan, SJ Marzi, C Shand, **BM Schilder**, S Tamburin, HM Tantiangco, I Lourida, DJ Llewellyn, JM Ranson
- 2023 • **EpiCompare: R package for the comparison and quality control of epigenomic peak files**
Bioinformatics Advances (2023) 13(1):vbad049; <https://doi.org/10.1093/bioadv/vbad049>
S Choi, **BM Schilder**, L Abbasova, AE Murphy, NG Skene

- 2022 • **Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors**
Biological Psychiatry (2022) 91(3):313-327; <https://doi.org/10.1016/j.biopsych.2021.05.029>
 N Mullins, J Kang, AI Campos,...**BM Schilder**, et al.
- 2022 • **Genetic analysis of the human microglial transcriptome across brain regions, aging and disease pathologies**
Nature Genetics (2022) <https://doi.org/10.1038/s41588-021-00976-y>
 K de Paiva Lopes, G JL Snijders, J Humphrey, A Allan, M Sneeboer, E Navarro, **BM Schilder**...T Raj
 **News**
 - Microglial transcriptomics meets genetics: new disease leads (Nature Reviews Neurology, 2022)
 - Mighty MiGA: Microglial Genomic Atlas Zeros in on Causal AD Risk Variants (ALZFORUM, 2022)
 - Can a Human Microglial Atlas Guide Brain Disorder Research? (Mount Sinai Health System, 2022)
 - Polygenic Scores Paint Microglia as Culprits in Alzheimer's (ALZFORUM, 2021)
- 2021 • **Multi-omic insights into Parkinson's Disease: From genetic associations to functional mechanisms**
Neurobiology of Disease (2021) 105580; <https://doi.org/10.1016/j.nbd.2021.105580>
BM Schilder, E Navarro, T Raj
- 2021 • **Fine-Mapping of Parkinson's Disease Susceptibility Loci Identifies Putative Causal Variants**
Human Molecular Genetics (2021) ddab294; <https://doi.org/10.1093/hmg/ddab294>
BM Schilder, T Raj
- 2021 • **echolocator: An Automated End-to-End Statistical and Functional Genomic Fine-Mapping Pipeline**
Bioinformatics (2021) btab658; <https://doi.org/10.1093/bioinformatics/btab658>
BM Schilder, J Humphrey, T Raj
- 2021 • **MungeSumstats: A Bioconductor Package for the Standardisation and Quality Control of Many GWAS Summary Statistics**
Bioinformatics (2021) 37(23):4593-4596; <https://doi.org/10.1093/bioinformatics/btab665>
 A Murphy, **BM Schilder**, NG Skene
- 2021 • **Dysregulation of mitochondrial and proteo-lysosomal genes in Parkinson's disease myeloid cells**
Nature Genetics (2021) <https://doi.org/10.1101/2020.07.20.212407>
 E Navarro, E Udine, K de Paiva Lopes, M Parks, G Riboldi, **BM Schilder**...T Raj
 **News**
 - Mount Sinai: Fighting Neurodegenerative Disorders (Mount Sinai Health System, 2019)
- 2021 • **Phenome-wide and eQTL Associations of COVID-19 Genetic Risk Loci**
iScience (2021) <https://doi.org/10.1016/j.isci.2021.102550>
 C Moon, **BM Schilder**, T Raj, K-I Huang
- 2021 • **Genome-Wide Association Study of over 40,000 Bipolar Disorder Cases Provides Novel Biological Insights**
Nature Genetics (2021) 53:817-829; <https://doi.org/10.1038/s41588-021-00857-4>
 N Mullins, AJ Forstner, KS O'Connell, B Coombes, JRI Coleman...**BM Schilder**... et al.
 **News**
 - Researchers identify 64 regions of the genome that increase risk for bipolar disorder (EurekAlert, 2021)
 - Largest Bipolar Disorder Genetics Study Doubles Genetic Risk Factors (Nordic Society of Human Genetics and Precision Medicine, 2021)
- 2020 • **Tensor decomposition of stimulated monocyte and macrophage gene expression profiles identifies neurodegenerative disease-specific trans-eQTLs**
PLOS Genetics (2020) 16(9):e1008549; <https://doi.org/10.1371/journal.pgen.1008549>
 S Ramdhani, E Navarro, E Udine, AG Efthymiou, **BM Schilder**, M Parks, A Goate, T Raj

- 2019 • **Evolutionary shifts dramatically reorganized the human hippocampal complex**
Journal of Comparative Neurology (2019) 528(17):3143-3170; <https://doi.org/10.1002/cne.24822>
 BM Schilder, HM Petry, PR Hof
- 2019 • **FAIRshake: Toolkit to Evaluate the Findability, Accessibility, Interoperability, and Reusability of Research Digital Resources**
Cell Systems (2019) 9; <https://doi.org/10.1016/j.cels.2019.09.011>
 D Clarke, L Wang, A Jones, M Wojciechowicz, D Torre, K Jagodnik, S Jenkins, P McQuilton, Z Flamholz, M Silverstein, BM Schilder...A Ma'ayan
 **News**
 - Chosen as 'Featured Frontmatter' article in *Cell Systems*
- 2019 • **Geneshot: search engine for ranking genes from arbitrary text queries**
Nucleic Acids Research (2019) 47(W1):W571-W577; <https://doi.org/10.1093/nar/gkz393>
 A Lachmann, BM Schilder, ML Wojciechowicz, D Torre, MV Kuleshov, AB Keenan, A Ma'ayan
 **News**
 - Geneshot: Piercing the Literature to Identify and Predict Relevant Genes (University of Pittsburgh Health Sciences Library System Update, 2019)
 - The Future of AI at the Hasso Plattner Institute for Digital Health at Mount Sinai (Mount Sinai Health System, 2020)
- 2018 • **eXpression2Kinases (X2K) Web: linking expression signatures to upstream cell signaling networks**
Nucleic Acids Research (2018) 46(W1):W171-W179; <https://doi.org/10.1093/nar/gky458>
 DJB Clarke, MV Kuleshov, BM Schilder, D Torre, ME Duffy, AB Keenan, A Lachmann, AS Feldmann, GW Gundersen, MC Silverstein, Z Wang
 **News**
 - Mount Sinai Faculty Spotlight: Ma'ayan Lab (Mount Sinai Health System, 2018)
- 2015 • **Defining elemental imitation mechanisms: A comparison of cognitive and motor-spatial imitation learning across object- and computer-based tasks**
Journal of Cognition and Development (2015) 17(2):221-243; <https://doi.org/10.1080/15248372.2015.1053483>
 F Subiaul, L Zimmerman, E Renner, BM Schilder, R Barr
- 2015 • **Take the monkey and run**
Journal of Neuroscience Methods (2015) 248:28-31; <http://doi.org/10.1016/j.jneumeth.2015.03.023>
 KA Phillips, MK Hambricht, K Hewes, BM Schilder, CN Ross, SD Tardif
 **News**
 - Monkeys on a Treadmill? A Conversation with Dr. Kimberley Phillips (Why Social Science?)
- 2014 • **Becoming a high-fidelity - super - imitator: what are the contributions of social and individual learning?**
Developmental Science (2014) 18(6):1025-1035; <http://doi.org/10.1111/desc.12276>
 F Subiaul, EM Patterson, BM Schilder, E Renner, R Barr
- 2014 • **Working memory constraints on imitation and emulation**
Journal of Experimental Child Psychology (2014) 128:190-200; <http://doi.org/10.1016/j.jecp.2014.07.005>
 F Subiaul, BM Schilder






PREPRINTS

- 2024 • **Zero-shot transfer learning of genomic disease signatures using single-cell foundation models**
arXiv (2024)
 BM Schilder
- 2024 • **Harnessing generative AI to annotate the severity of all phenotypic abnormalities within the Human Phenotype Ontology**
medRxiv (2024) <https://doi.org/10.1101/2024.06.10.24308475>
 KB Murphy, BM Schilder, NG Skene

- 2024 • **Navigating the Phenomic Landscape: systematic characterisation of the latent genomic space underlying all traits and diseases**
bioRxiv (2024) <http://dx.doi.org/10.13140/RG.2.2.12144.26880>
 BM Schilder, NG Skene
- 2024 • **Chromatin interactions and active histone mark signatures underpin TBXT expression in metastatic lung cancer**
SSRN (2024) <https://ssrn.com/abstract=4965385>
 RM Yaa, BM Schilder, RD Acemel, FC Wardle
- 2024 • **Integrative multi-omics analysis of glial signatures associated with accelerated cognitive decline in Alzheimer's disease**
bioRxiv (2024)
 E Schneegans, N Fancy, V Chau, TKD Cheung, E Adair, M Papageorgopoulou, BM Schilder, PM Matthews, JS Jackson
- 2023 • **Fine-mapping genomic loci refines bipolar disorder risk genes**
medRxiv (2023) <https://www.medrxiv.org/content/10.1101/2024.02.12.24302716v1>
 M Koromina, A Ravi, G Panagiotaropoulou, BM Schilder, ... S Ripke, T Raj, JRI Coleman, N Mullins
 **News**
 - Currently under journal review
- 2023 • **Identification of cell type-specific gene targets underlying thousands of rare diseases and subtraits**
medRxiv (2023) <https://doi.org/10.1101/2023.02.13.23285820>
 KB Murphy, R Gordon-Smith, J Chapman, M Otani, BM Schilder, NG Skene
- 2022 • **CUT&Tag recovers up to half of ENCODE ChIP-seq peaks**
bioRxiv (2022) <https://doi.org/10.1101/2022.03.30.486382>
 D Hu, L Abbasova, BM Schilder, A Nott, NG Skene, SJ Marzi



RESEARCH EXPERIENCE


- I
2024 • **Postdoctoral Research Scientist**
 Cold Spring Harbor Laboratory (Quantitative Biology)  Cold Spring Harbor, NY, USA
 • Advancing deep learning applications in genomics and biomedicine in the laboratory of Dr. Peter Koo.
 • Developing a genomic foundation model to map complex genome-phenome relationships and make highly accurate, personalized disease risk predictions.
- I
2019 • **Lead Data Scientist**
 120/80 Group  New York, NY, USA
 • Offers data-driven consultation services to a wide portfolio of high-profile digital healthcare, pharmaceutical and biotech companies.
 • Developed a suite of proprietary softwares to extract customised business intelligence from the published literature to generate customised and interpretable reports to clients.
 • Provides clients guidance on strategic AI implementation, data analysis, publication and transparency.
- 2020
I
2018 • **Bioinformatician II**
 Icahn School of Medicine at Mount Sinai (Department of Neuroscience / Department of Neurology / Department of Genetics & Genomics / Ronald M. Loeb Center for Alzheimer's Disease)  New York, NY, USA
 • Developed machine learning systems to integrate large-scale multi-omics datasets (e.g. whole-genome sequencing, bulk and single-cell RNA-seq, epigenomics, clinical data) to uncover the molecular mechanisms underlying neurodegenerative diseases (e.g. Alzheimer's, Parkinson's, ALS).
 • Computationally identified specific disease-causal variants, pathways and cell-types for subsequent functional wet lab validation (e.g. CRISPR-cas9 editing in patient-derived cell cultures, iPSCs and cerebral organoids).

- 2018
|
2017

Bioinformatician II

Icahn School of Medicine at Mount Sinai (Department of Pharmacological Sciences)

 - Conducted computational systems biology research. Integrated and analyzed large-scale genomic and biomedical data (e.g. Python, R, JavaScript).
 - Developed evolutionary algorithm to optimize gene network kinase regulator prediction (eXpression2Kinases).
 - Developed and deployed computational tools, software, databases and web applications for basic and clinical research, resulting in 3 peer-reviewed publications.


 [New York, NY, USA](#)

- 2013
|
2011

Research Assistant

The George Washington University (Department of Anthropology)

 - Performed dissection, histology, microscopy and quantitative stereology in post-mortem primate brain tissues.
 - Trained junior and senior personnel on lab protocols.


 [Washington, DC, USA](#)

- 2013
|
2011

Senior Lab Manager

The George Washington University (Department of Speech, Language & Hearing Sciences)

 - Organized and trained dozens of undergraduates to conduct weekly cognitive development research; designed and/or directly contributed to over 15 research projects in two years.


 [Washington, DC, USA](#)

- 2010

Paid Research Intern

Princeton University (Princeton Neuroscience Institute)

 - Investigated the neural basis of decision-making in humans.
 - Recruited participants, recorded EEG and analyzed data in MATLAB.

 [Princeton, NJ, USA](#)

\$ GRANTS

- Total (all grants): \$2,949,872**

Total (as primary applicant): \$311,382