BRIAN M SCHILDER, PHD

Passionately pursuing transdisciplinary research to advance human health and knowledge.

CSH 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

16+ 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 **14** 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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+44 073-0653-7736
🔗 **LinkedIn**
🆔 **ORCID**
G Google Scholar
GitHub
Twitter
YouTube
🌐 **Personal Website**
🌐 **Lab Website**

SUMMARY

🗂 **16+ years of research**
📄 **23 publications**
📑 **8 preprints**
🔧 **41 software packages**
🗄 **11 databases & apps**
📣 **23 talks**
🔁 **14+ years of teaching & team management**

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🧊 *Made with autoCV*

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.
- **Collaboration**: Diverse and global collaborative networking.
- **Communication**: Clear and concise distillation of complex results to a variety of audiences. Presented **25** conference posters.

📄 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 Khleifat, 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 Hambright, 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

2025 Gene expression patterns of the developing human face at single cell resolution reveal cell type contributions to normal facial variation and disease risk
bioRxiv (2025) https://www.biorxiv.org/content/10.1101/2025.01.18.633396v1
N Khouri-Farah, EW Winchester, **BM Schilder**, K Robinson, SW Curtis, NM Skene, E Leslie-Clarkson, J Cotney

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

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2024

Postdoctoral Research Scientist
Cold Spring Harbor Laboratory (Simons Center for 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.

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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
|
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)   ◉ New York, NY, USA
- 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.

2013 | 2011

Research Assistant

The George Washington University (Department of Anthropology)   ◉ Washington, DC, USA
- Performed dissection, histology, microscopy and quantitative stereology in post-mortem primate brain tissues.
- Trained junior and senior personnel on lab protocols.

2013 | 2011

Senior Lab Manager

The George Washington University (Department of Speech, Language & Hearing Sciences)
◉ Washington, DC, USA
- 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.

2010

Paid Research Intern

Princeton University (Princeton Neuroscience Institute)   ◉ Princeton, NJ, USA
- Investigated the neural basis of decision-making in humans.
- Recruited participants, recorded EEG and analyzed data in MATLAB.

$ GRANTS

Total (all grants): $3,049,872
Total (as primary applicant): $311,382