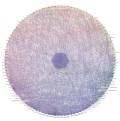

BRIAN M SCHILDER, PHD

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

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 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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+1 908-268-9859
UK
+44 073-0653-7736
LinkedIn
ORCID
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

- | 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.

- | 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): $2,949,872
Total (as primary applicant): $311,382