

# BRIAN M SCHILDERBRIAN M

# **SCHILDER**

Passionately pursuing transdisciplinary research to advance human health and knowledge.

- 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 a genomics, AI, evolutionary biology and biomedicine. Stategically 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 anayses and AI model training (CPUs and GPUs).
- Web development: 6+ websites, web apps, and interactive reports.

### CONTACT

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**UK** 

+44 073-0653-7736

in LinkedIn

ORCID

**G** Google Scholar

**G** GitHub

**▼** Twitter

YouTube

Personal Website

Lab Website

# SUMMARY

11 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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Education

✓ Skills

Publications

Preprints

**Experience** 

**S** Grants

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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:

- 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.
- Causal variant effect prediction: Used functional impact 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 neurodegenerationrelevant trans-eQTLs

# **Project Management**

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

- Documentation: Defines obectives 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 communcation.

- 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.

2023

2023

2023

# **PUBLICATIONS**

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 I

■ News

- Featured in Nature Communications Editors' Highlights

# 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

## 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

#### 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

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

2023

2023

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, Al 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 SchilderT 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 SchilderT Raj  Rews  - 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 ColemanBM 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

Evolutionary shifts dramatically reorganized the human hippocampal complex 2019 Journal of Comparative Neurology (2019) 528(17):3143-3170; https://doi.org/10.1002/cne.24822 BM Schilder, HM Petry, PR Hof FAIRshake: Toolkit to Evaluate the Findability, Accessibility, Interoperability, and Reusability of 2019 **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 Geneshot: search engine for ranking genes from arbitrary text queries 2019 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 - 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) eXpression2Kinases (X2K) Web: linking expression signatures to upstream cell signaling 2018 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) Defining elemental imitation mechanisms: A comparison of cognitive and motor-spatial imitation 2015 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 I

■ News - Monkeys on a Treadmill? A Conversation with Dr. Kimberley Phillips (Why Social Science?)

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

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



2014

2014

2024

2024

Zero-shot transfer learning of genomic disease signatures using single-cell foundation models arXiv (2024)

**BM Schilder** 

 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

Navigating the Phenomic Landscape: systematic characterisation of the latent genomic space underlying all traits and diseases

bioRxiv (2024)

BM Schilder, NG Skene

Active chromatin interaction and histone mark signatures underpin epigenetic TBXT expression in metastatic lung cancer

bioRxiv (2024)

RM Yaa, BM Schilder, RD Acemel, FC Wardle

Integrative multi-omics analysis of glial signatures associated with accelerated cognitive decline in Alzheimer's disease

bioRxiv (2024)

2024

2023

2023

2022

2019

2020

2018

2018

2017

E Schneegans, N Fancy, V Chau, TKD Cheung, E Adair, M Papageorgopoulou, **BM Schilder**, PM Matthews, JS Jackson

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

- Currently under journal review

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

CUT&Tag recovers up to half of ENCODE ChIP-seg 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

#### 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 Al implementation, data analysis, publication and transparency.

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).

# **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.

5

# **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

### 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