# Alan P. Boyle

#### **Education**

Doctor of Philosophy, Computational Biology and Bioinformatics
 Duke University, Durham, NC

 Bachelor of Science, summa cum laude, Biochemistry and Molecular Biology
 Bachelor of Science, summa cum laude, Computer Science
 Mississippi State University, Starkville, MS

### **Academic Appointments**

2025-present 2023-present	Professor with tenure, Department of Computational Medicine & Bioinformatics Professor, Department of Human Genetics Core Member, Rogel Cancer Center
2020–2023	Affiliate Member, Rogel Cancer Center
2021-present	Affiliate, Michigan Neuroscience Institute
2016-present	Member, Center for RNA Biomedicine
2020–2025	Associate Professor with tenure, Department of Computational Medicine & Bioinformatics Associate Professor, Department of Human Genetics
2015–2020 2014–2020	Assistant Professor, Department of Human Genetics Assistant Professor, Department of Computational Medicine & Bioinformatics University of Michigan, Ann Arbor, MI
2010–2014	Postdoctoral Scholar, Genetics Stanford University, Stanford, CA; Advisor: Dr. Michael Snyder
Spring 2010	Postdoctoral Associate, Computational Biology Duke University, Durham, NC; Advisor: Dr. Terrence S. Furey

### Scholarships, Fellowships, and Honors

2022	Valuing our Own Award, Michigan Medicine
2019	Endowment for the Basic Sciences Teaching Award
2018	First Place in CAGI5 Regulation Saturation Challenge
2017	NSF CAREER Award
2016	Institutional nominee for W.M. Keck Foundation Medical Science Research Program
2016	Institutional nominee for Searle Scholar Award
2015-2017	Alfred P. Sloan Foundation Fellowship in Computational & Evolutionary Molecular Biology
2013-2014	NIH Pathway to Independence Award (K99/R00) [1K99HG007356-01]
2012	AAAS/Science Program for Excellence in Science
2005–2008	NSF Graduate Research Fellowship
2005-2009	James B. Duke Fellowship
Summer 2004	Mayo Clinic Summer Undergraduate Research Fellow
2003	Barry M. Goldwater Memorial Scholarship
Summer 2003	The Institute for Genomic Research (TIGR) Summer Fellow
2001	Robert C. Byrd Honors Scholarship
2001	Mississippi State University Presidential Scholarship
2001	National Merit Scholarship

### **Grant Support**

#### Active

Alan P. Boyle May 16, 2025

U24 HG009293 (Multi-PI: Boyle, Cherry) 2017-2025

NIH/NHGRI

RegulomeDB: A Resource for the Human Regulome

This project seeks to expand and support a RegulomeDB, a database for prioritizing and predicting functional variants in the human genome.

U01 HG011952

(PI: Boyle)

NIH/NHGRI

2021-2026

Predicting the impact of genomic variation on cellular states

This project seeks to develop tools for interpretation of genomic variation on cellular state through modeling single cell data as part of the IGVF consortium.

2022-2026 R01 GM144484 (PI: Boyle)

NIH/NIGMS

Mobile element derived chromatin looping variability in human populations

This project seeks to study the impact of polymorphic LTR13 integrations on 3D chromatin conformation.

UG3 NS132084 (Multi-PI: Mills, Boyle, McConnell) 2023-2028

NIH/OD

Molecular and Computational Tools for Identifying Somatic Mosaicism in Human Tissues As part of the SMaHT consortium this project seeks to develop long-read methods to study somatic mosaicism in normal human tissues.

**Taubman Institute Innovation Projects** (co-PI: Todd, Boyle, Mills) 2022-2025

University of Michigan

Short Tandem repeats in precision health and human disease

The goal of this project is to develop any assay to measure STRs in human genomes and develop bioinformatic tools to predict STR expansions from genotypes.

K08 HL153799 (PI: Denstaedt; Consultant) 2021-2026

NIH/NHLBI

Predisposition for Lung Injury in Sepsis Survival

The goal of this project is to understand the biological mechanisms predisposing to these complications in order to prevent and treat them.

R01 NS122165 (PI: Castro; Co-I with Effort) 2021-2026

NIH/NINDS

Uncover the role of H3.3-G343R mutation in shaping the DNA damage response, anti-tumor immunity and mechanisms of resistance in glioma

This project seeks to study pediatric high-grade gliomas with H3.3-G343R, ATRX, and TP53 inactivating mutations to understand the impact of H3.3-G343R on the tumor immune microenvironment.

R01 CA260677 (PI: Malek; Co-I with Effort) 2022-2026

NIH/NCI

The Biology of Mutant STAT6 in Follicular Lymphoma

This project seeks to study STAT6 gene regulation in the context of B cell lymphoma.

R01 NS099280 (PI: Todd; Consultant) 2023-2028

NIH/NINDS

NIH/NIDCR

Hexanucleotide repeat translation in ALS and Frontotemporal Dementia

This project seeks to study RAN translation in ALS and FTD at a hexonucleotide expansion in C9orf72.

R01 DE032699 2024-2029 (PI: Brenner, Mills, Spector; Co-I with Effort)

Defining the Role of HPV Integration Structures in HNSCC Molecular Heterogeneity

This proposal seeks to determine how the structure of HPV integration events influences chromatin accessibility and adjacent gene expression in HPV-positive oropharyngeal squamous cell carcinoma, with the goal of identifying pivotal driver integrations that can predict treatment outcomes and guide therapy decisions to improve patient survival and reduce morbidity.

Completed

2013–2017 R00 HG007356 Pathway to Independence Award (K99/R00)

(PI: Boyle)

NIH/NHGRI

Global Discovery and Validation of Functional Regulatory Elements

This project seeks to extend current assays demonstrating function of genomic regions into an equivalent genome-wide assay.

2015-2017 FG-2015-65465

(PI: Boyle)

Alfred P. Sloan Foundation

Fellowship in Computational & Evolutionary Molecular Biology

2016–2020 R01 HL130705

(PI: Willer; Co-I with Effort)

NIH/NHLBI

Large-scale human genetics to understand molecular mechanisms of atrial fibrillation and related traits

This project seeks to provide new insights into atrial fibrillation mechanisms through wholegenome screening.

2017-2018 Eleanor and Larry Jackier U-M/Technion and Weizmann Collaborative Research Grant

(co-PI: Boyle, Mandel-Gutfreund)

Michigan - Israel Partnership for Research & Education

Identifying novel disease related mutations in the genomic environments around Trascription Factor binding sites

The goal of this project is to identify variants in the proximity of TF binding sites that have an indirect effect on their binding.

2017-2024 R35 HL135824

(PI: Willer; Co-I with Effort)

NIH/NHLBI

Using Genetics to Inform Mechanism of Cardiovascular Disease

The goal of this project is to uncover novel genetic discoveries and biological mechanisms underlying association with devastating cardiovascular diseases.

2019 NVIDIA GPU Grant

(PI: Boyle)

NVIDIA Corporation

2017-2022 | DBI-1651614

(PI: Boyle)

NSF/BIO/DBI

CAREER: Conservation of cohesin-containing cis regulatory modules in the human and mouse lineages

The goal of this project is the study of the turnover of cohesin binding sites in the human and mouse genomes.

2022-2022 R21 HG011493 S1

(Multi-PI: Boyle, Mills)

NIH/NIA

New technologies for accurate capture and sequencing of repeat-associated regions (Supplement)

This project seeks to map mobile elements in a set of Alzheimer's samples.

2019–2022 | Precision Health Investigators Award

(co-PI: Todd, Boyle, Mills)

University of Michigan

Short Tandem repeats in precision health and human disease

The goal of this project is to develop any assay to measure STRs in human genomes and develop bioinformatic tools to predict STR expansions from genotypes.

2022 NVIDIA GPU Grant

(PI: Boyle)

**NVIDIA Corporation** 

2021–2022 | Cancer Center Discovery

(PI: Boyle)

University of Michigan

Direct capture of complete HPV integration sites using long-read sequencing

This project seeks to develop methods to capture of complete HPV integration events in the human genome.

Alan P. Boyle May 16, 2025

W81XWH2010336 (PI: Aguilar; Co-I with Effort) 2020-2023

DoD/Army

Understanding & Enhancing the Regenerative Capacity of Skeletal Muscle to Trauma by Targeting Muscle-Nerve Synergy

This project seeks to study the single cell chromatin and RNA landscape in skeletal muscle repair.

R21 HG011493 2020-2023 (Multi-PI: Boyle, Mills)

NIH/NHGRI

New technologies for accurate capture and sequencing of repeat-associated regions

This project seeks to map mobile elements in a trio of cell lines and develop technologies for

improving this mapping.

2018-2024 R01 HD093570 (PI: Bielas; Co-I with Effort)

NIH/NICHD

Genetic Diagnosis of Neurodevelopmental Disorders in India

This study will establish whole-exome sequencing to study mendelian genetic disorders at the All

India Institute of Medical Sciences.

Michigan Alzheimer's Disease Center Developmental Project (PI: Zhou; Consultant) 2022-2024

University of Michigan

Explore the functional impact of transposable elements in Alzheimer's disease and related dementias

This project seeks to explore the connection between the somatic transposable elements in the human genome and Alzheimer's disease and related dementias.

R21 CA2578964 2022-2025 (PI: Boyle)

NIH/NCI

High-throughput inverted reporter assay for characterization of silencers and enhancer blockers This project seeks to develop tools for the study of negative regulatory elements in cancer development.

R01 HD104680 (PI: Hammoud; Co-I with Effort) 2021-2025

NIH/NICHD

Sperm Chromatin: Implications on organismal development and fertility This project seeks to explore protamine chromatin structure in mouse sperm.

#### **Professional Service**

#### Service

DHG Faculty Development Committee 2024-current

R01 Bootcamp Medical School Cohort Coach 2023-current

Somatic Mosaicism across Human Tissues (SMaHT) Consortium Steering Committee 2023-current

Impact of Genomic Variation on Function (IGVF) Consortium Steering Committee 2021-current

University of Michigan Biomedical Research Council (BMRC) (Standing Member) 2022-current

DHG M.S. Admissions Committee 2020-2022 Lab Safety Liaison for DCM&B 2018-current

DCM&B Preliminary Exam Abstract Review Committee (PARC) [Chair 2018–2022] 2017-2024

DHG Ph.D. Admissions Committee 2019-2020

2017-2020 DHG Faculty Recruitment and Promotions Committee

DCM&B Seminar Series Committee [Chair] 2016-2020

Cellular and Molecular Biology Admissions Committee 2018-2019

EBS Faculty IT Committee 2017-2019

DCM&B Faculty Recruitment Committee 2016-2019

DCM&B Admissions Committee 2015-2018

DHG Computational Support Committee 2015-2017

DCM&B Retreat Planing Committee Chair (including 1st annual) 2015, 2016, 2025

2014 Ad hoc admissions reviewer, University of Michigan DCM&B

Duke Computational Biology & Bioinformatics student committee 2008-2009

### Memberships

2018-current	Member, American Society of Human Genetics (ASHG)
2013-current	Member, International Society for Computational Biology (ISCB)
2012-current	Member, American Association for the Advancement of Science (AAAS)
2005-current	Member, Gamma Sigma Delta Agricultural Honor Society

### **Manuscript Reviewing Activity**

Since 2009	Ad hoc reviewer (>100 verified reviews) for the journals: Science, Nature Biotechnology, Nature
	Genetics, Genome Research, Genome Biology, Nature Neuroscience, Nature Communications,
	Nature Protocols, Bioinformatics, Nucleic Acids Research, BMC Biology, BMC Bioinformatics,
	PLOS Computational Biology, Oncotarget, Scientific Reports, Atherosclerosis, BioEssays, Gene
2023	Program Committee, Genome Sequence Analysis, ISMB/ECCB
2023	Program Committee, Biomedical Informatics, ISMB/ECCB
2018, 2020	Program Committee, Comparative and Functional Genomics, ISMB/ECCB
2018, 2019	Program Committee, Studies of Phenotypes and Clinical Applications, ISMB/ECCB
2019	Program Committee, General Computational Biology, ISMB/ECCB
2017	Program Committee, Regulatory Genomics Special Interest Group Meeting (RegGenSIG),
	ISMB/ECCB
2015–2018	Program Committee, Great Lakes Bioinformatics and Canadian Computational Biology Confer-
	ence (GLBIO/CCBC)
2015–2016	Program Committee, Algorithms for Computational Biology (ALCOB)
2013–2016	Program Committee, Gene Regulation and Transcriptomics, ISMB/ECCB
2012–2015	DNA Day Essay Contest Detailed Review Judge for ASHG
2012	Distinguished contributor as a leading reviewer for the journal Bioinformatics

### **Grant Reviewing Activity**

2025	NIH Study Section BDMA - Biodata Management and Analysis Study Section (Ad Hoc)
2024	NIH Study Section ZRG1 BBBT-D (50) - PAR Panel: Biodata Management and Common Fund
	Data Sets (Ad Hoc and co-Chair)
2024	NIH Study Section BDMA - Biodata Management and Analysis Study Section (Ad Hoc)
2023	NSF Review Panel - Molecular and Cellular Biosciences (MCB) - Genetic Mechanisms (Ad Hoc)
2023	NIH Study Section - Multi-Omics of Health and Disease - Data Analysis and Coordination Center
2023	NIH Study Section GVE - Genetic Variation and Evolution Study Section (Ad Hoc)
2022	NIH Study Section ZRG1 ISB-S (57) - Academic-Industrial Partnerships for Translation of Tech-
	nologies for Diagnosis and Treatment
2022	NASA Study Section E.11 Space Biology: Animal Studies - Omics Systems [21SBAS-OmisSys]
	(Ad Hoc)
2020	NIH/NIMH Study Section ZMH1 ERB-C (08) - Fine-Mapping Genome-Wide Associated Loci to
	Identify Proximate Causal Mechanisms of Serious Mental Illness
2019	NIH/NIMH Study Section ZMH1 ERB-C (01) - PsychENCODE: Non-Coding Functional Elements
	in the Human Brain and Their Role in the Development of Psychiatric Disorders
2018–2019	University of Michigan internal review for Searle Scholars Program
2015	UK Medical Research Council (RCUK MRC) - Methodology Research Panel (Ad Hoc)
2015	UK Biotechnology and Biological Sciences Research Council (RCUK BBSRC) (Ad Hoc)
2015	Michigan Institute for Clinical & Health Research (MICHR) Postdoctoral Translational Scholars
	Program (Ad Hoc)

## **Teaching and Mentorship**

## **Training Programs**

2024-present	Member, Systems & Integrative Biology Training Grant (SIB)
2021-present	Member, Biomedical Informatics and Data Science Training Program (BIDS-TP)
2017-present	Member, Cellular and Molecular Biology Program
2015-present	Member, Genome Science Training Program (GSTP)
2015-present	Member, Michigan Predoctoral Training Program in Genetics (GTP)
2014-present	Member, Program in Biomedical Sciences

2014–present | **Member**, Bioinformatics Training Program

**Teaching** (F = Fall Term, W = Winter Term, S = Summer Term)

W19, W20, W21, Bioinformatics Concepts and Algorithms (BIOINF 529) [Course Director] W22, W23, W24, Gene Structure and Regulation (HUMGEN 541) [3 lectures + 2 discussions / yr.] F15, F16, F17, F18, F19, F20, F21 Research Responsibility and Ethics (PIBS 503) [1 discussion / yr.] F19, F22 F21, W22, F24, Genetics Student Seminar (HUMGEN 821/822) [Mentor] W25 Experimental Genetics Systems (HUMGEN 632) [Course Director] F17, F18 Bioinformatics Journal Club (BIOINF 602/603) [Course Director F18] F15, W16, F16, W17, F17, W18, S17, S18 Introduction to Biocomputing Bootcamp (BIOSTAT/BIOINF/HUMGEN 606) [2 full days / yr.] Introduction to Bioinformatics & Computational Biology (BIOINF 527) [2 lectures + 3 labs / yr.] F15, F16, F17 Basic Biology for Graduate Students with Quantitative Training (BIOINF 523) [2 lectures / yr.] S15, S16, S17 Lab TA for Isotopes Tech I (MS. State, BCH 4414)

#### **Guest Lectures / Panels**

2018–2019	Lecturer, REU Site: Mathematical and Theoretical Biology Institute (MTBI), Arizona State Univer-
	sity (NSF1757968) [2 days]
2017	Panel member, U. Michigan "New Faculty Orientation to Corporate & Foundation Relations" [70
	attendees]
2016	Experimental Genetics Systems (HUMGEN 632) [1 discussion]
2014	Panel member, BIOINF 527 "Challenges in Biology, Biomedicine, Data & Analysis"
2010	Co-taught Cold Spring Harbor Systems Biology Pre-meeting Workshop
2009	Duke student panelist for "How to prepare for and get into graduate school"
2008	Taught Duke mini-course on Genome Browsers & Databases

#### **Mentorship**

#### **Graduate Students (n=24)**

2024-current	Ingrid Flaspohler (Ph.D. Student, Bioinformatics, University of Michigan) Rackham Graduate Student Research Grant (pre-candidate)
2024-current	Steve Losh (Ph.D. Student, Bioinformatics, University of Michigan)
2024-current	Sowmya Srinivasan (Ph.D. Student, Genetics and Genomics, University of Michigan)
2023–2023	Hawra Aljawad (M.S. Student, Chemical Engineering, University of Michigan) Rackham Graduate Student Research Grant (pre-candidate)
2023–2023	Xinyi Liu (M.S. Student, Bioinformatics, University of Michigan)
2022–2023	Emily Pogson (M.S. Student, Genetics and Genomics, University of Michigan)
2022-current	Katarina Pavlovic (Ph.D. Student, Bioinformatics, University of Michigan) Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate)
2022-current	Rintsen Sherpa (Ph.D. Student, Bioinformatics, University of Michigan) Rackham Graduate Student Research Grant (candidate)
2021-current	Kinsey Van Deynze (Ph.D. Student, Bioinformatics, University of Michigan) NIH Genome Science Training Program (T32) Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate)
2020-current	Andrea Valenzuela (Ph.D. Student, Chemical Biology, University of Michigan)  NIH Cellular Biotechnology Training Program (T32)
2020-current	Breanna McBean (Ph.D. Student, Genetics and Genomics, University of Michigan)

Joint M.S. in Bioinformatics, University of Michigan NIH Genome Science Training Program (T32) Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate) Monica Holmes (M.S. Student, Bioinformatics, University of Michigan) 2019-2020 Camille Mumm (Ph.D. Student, Genetics and Genomics, University of Michigan) 2020-2025 Joint M.S. in Bioinformatics, University of Michigan NIH Genome Science Training Program (T32) Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate) Rackham Pre-doctoral Fellowship 2018-2024 Bradley Crone (Ph.D. Student, Bioinformatics, University of Michigan) Rackham Graduate Student Research Grant (candidate) Melissa Englund (Ph.D. Student, Genetics and Genomics, University of Michigan) 2017-2023 NIH Human Genetics Training Program (T32) Rackham Graduate Student Research Grant (candidate) 2018-2023 Nanxiang (Samuel) Zhao (Ph.D. Student, Bioinformatics, University of Michigan) Nanxiang (Samuel) Zhao (M.S. Student, Bioinformatics, University of Michigan) 2017-2018 Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate) Haley Amemiya (Ph.D. Student, Cellular and Molecular Biology, University of Michigan) 2016-2018 Joint M.S. in Bioinformatics, University of Michigan NIH Cellular & Molecular Biology Training Program (T32) NIH Cellular Biotechnology Training Program (T32) (Declined) PIBS Excellence in Service Award Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate) Maas Professional Development Award Rackham Graduate School Scholar-Activist Award Shriya Sethuraman (Ph.D. Student, Bioinformatics, University of Michigan) 2016-2020 Christopher Castro (Ph.D. Student, Bioinformatics, University of Michigan) 2016-2023 NIH Bioinformatics Training Program (T32) Rackham Merit Fellow Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate) Global Research Engagement Opportunity Fellowship Ningxin Ouyang (Ph.D. Student, Bioinformatics, University of Michigan) 2017-2022 Ningxin Ouyang (M.S. Student, Bioinformatics, University of Michigan) 2015-2017 Rackham Graduate Student Research Grant (candidate) Shengcheng Dong (Ph.D. Student, Bioinformatics, University of Michigan) 2016-2021 Rackham Graduate Student Research Grant (candidate) Torrin McDonald (Ph.D. Student, Genetics and Genomics, University of Michigan) 2015-2021 NIH Human Genetics Training Program (T32) Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate) Greg Farnum (Ph.D. Student, Cellular and Molecular Biology, University of Michigan) 2015-2017 Sierra Nishizaki (Ph.D. Student, Genetics and Genomics, University of Michigan) 2015-2020 Joint M.S. in Bioinformatics, University of Michigan NIH Genome Science Training Program (T32) Rackham Merit Fellow Rackham Summer Award

Rackham Graduate Student Research Grant (candidate)

#### Additional Graduate Rotation Students (n=17)

Karan Smith (Rotation Student, Cellular and Molecular Biology, University of Michigan) 2024 2024 Jun Sik Yun (Rotation Student, Genetics and Genomics, University of Michigan) Jeremy Chen (Rotation Student, Bioinformatics, University of Michigan) 2024 Rosina Carr (Rotation Student, Bioinformatics, University of Michigan) 2023 Connor Ward (Rotation Student, Medical Science Training Progran, University of Michigan) 2023 Brandt Bessell (Rotation Student, Bioinformatics, University of Michigan) 2022 Xiaomeng Du (Rotation Student, Bioinformatics, University of Michigan) 2022 Mahnoor Gondal (Rotation Student, Bioinformatics, University of Michigan) 2022 Xin Li (Rotation Student, Biological Chemistry, University of Michigan) 2022 Bohan Chen (Rotation Student, Cell and Developmental Biology, University of Michigan) 2022 Amelia Lauth (Rotation Student, Cellular and Molecular Biology, University of Michigan) 2021 Margarita Brovkina (Rotation Student, Cellular and Molecular Biology, University of Michigan) 2019 Steve Ho (Rotation Student, Human Genetics, University of Michigan) 2018 2018 Matthew Pun (Rotation Student, Medical Science Training Progran, University of Michigan) Amanda Moccia (Rotation Student, Human Genetics, University of Michigan) 2017 Stephen Carney (Rotation Student, Human Genetics, University of Michigan) 2017 Tingyang Li (Rotation Student, Bioinformatics, University of Michigan)

#### Postdoctoral Fellows (n=3)

2023-current	Melissa Englund (University of Michigan)
2022-2025	Torrin McDonald (University of Michigan)
2021-2022	Shengcheng Dong (University of Michigan)

#### Non-student Lab Volunteers (n=2)

2019-2021	Greg Farnum (University of Michigan)
2018-2019	Monica Holmes (Postbac, University of Michigan)

#### **Undergraduate and High School Students (n=21)**

2025-current	Krrish Thakker (Undergraduate, Biochemistry, University of Michigan)
2023-2024	Kateri Darr (Undergraduate, Computer Science, University of Michigan)
2023-2023	Mason Miller (Undergraduate, Computer Science, University of Michigan)
2022-2024	Summer Ann (Undergraduate, Neuroscience, University of Michigan)
2022-current	Kobe Howcroft (Undergraduate, Computer Science, University of Michigan)
2021-2024	Preston Parana (Undergraduate, UROP Molecular, Cellular, and Developmental Biology, Univer-
	sity of Michigan)
	UROP Blue Ribbon Award
2021-2022	Julia Tweadey (Undergraduate, LSA Honors Program, Life Science Informatics, University of
	Michigan)
2021	Aryn Booker (Undergraduate, UROP Molecular, Cellular, and Developmental Biology, University
	of Michigan)
	UROP Blue Ribbon Award
2020	Marcela Alcaide Aligio (Undergraduate, SROP, Hunter College CUNY)
2019–2020	David Wang (Undergraduate, UROP Computer Science, University of Michigan)
2019–2020	Jack Lu (Undergraduate, UROP Computer Science, University of Michigan)
2019–2020	Diana Davis (Undergraduate, Neuroscience and German, University of Michigan)
2019	Sheila Rasouli (Undergraduate, Neuroscience, University of Toronto)
2019	Vibhasri Davuluri (High School, Girls Who Code Summer Intern)
2016–2019	Cody Morterud (Undergraduate, UROP Computer Science / Honors Capstone, University of Michi-
	gan)
2016–2017	Colten Williams (Undergraduate, UROP Computer Science, University of Michigan)
2016–2017	Courtney Asman (Undergraduate, Neuroscience, University of Michigan)
2014–2017	Maxwell Spadafore (Undergraduate, LS&A Honors Informatics, University of Michigan)
2013–2014	Natalie Ng (High School, Stanford Institutes of Medicine Summer Research)
2013–2014	Dana Wyman (Undergraduate, Biology, Stanford University)

2013 Justin Young (High School, Stanford Institutes of Medicine Summer Research)

Melanie Connick (Undergraduate, Biology, University of New Mexico)
Edward Dai (Undergraduate, Computer Science, Stanford University)

### **Doctoral Thesis Committees (n=49)**

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2024-current	Weizhou Qian (Bioinformatics, University of Michigan, Committee Member)
2024-current	Lingrui Cai (Bioinformatics, University of Michigan, Committee Member)
2024-current	Jinhao Wang (Bioinformatics, University of Michigan, Committee Member)
2024-current	Brandon Klein (Medicinal Chemistry, University of Michigan, Committee Member)
2024-current	Bohan Chen (Bioinformatics, University of Michigan, Committee Member)
2024-current	Sowmya Srinivasan (Genetics and Genomics, University of Michigan, co-Chair)
2024-current	Ingrid Flaspohler (Bioinformatics, University of Michigan, Chair)
2024-current	Steve Losh (Bioinformatics, University of Michigan, Chair)
2024-current	Matthew Hodgman (Bioinformatics, University of Michigan, Committee Member)
2024-current	Lu Lu (Bioinformatics, University of Michigan, Committee Member)
2023-current	Linghua Jiang (Bioinformatics, University of Michigan, Committee Member)
2023-current	Elysia Chou (Bioinformatics, University of Michigan, Committee Member)
2023-current	Rebecca McAvoy (Molecular, Cellular, and Developmental Biology, University of Michigan, Com-
	mittee Member)
2023-current	Chinmay Raut (Bioinformatics, University of Michigan, Committee Member)
2022-current	Katarina Pavlovic (Bioinformatics, University of Michigan, Chair)
2022-current	Rintsen Sherpa (Bioinformatics, University of Michigan, Chair)
2022-current	Emily Peirent (Neuroscience, University of Michigan, Committee Member)
2021-current	Kinsey Van Deynze (Bioinformatics, University of Michigan, Chair)
2021-current	Mashiat Rabbani (Genetics and Genomics, University of Michigan, Committee Member)
2020-current	Andrea Valenzuela (Chemical Biology, University of Michigan, co-Chair)
2020-current	Breanna McBean (Genetics and Genomics, University of Michigan, co-Chair)
2020–2025	Steve Ho (Genetics and Genomics, University of Michigan, Committee Member)
	Applied Machine Learning for Big Data Genomics
2020-2025	Camille Mumm (Genetics and Genomics, University of Michigan, Chair)
	Exploring Repetitive Elements in Neurodegenerative Disease Using Targeted Long-Read Se-
	quencing
2022–2025	Kaiwen Deng (Bioinformatics, University of Michigan, Committee Member)
	Advancing Computational Models and Algorithms for the Analysis of Single-Cell and Neural Data
2022–2024	Franco Tavella (Biophysics, University of Michigan, Committee Member)
	Robustness and Tunability of Biological Oscillations
2018–2024	Bradley Crone (Bioinformatics, University of Michigan, Chair)
	Computational Methods in Functional Prioritization of Polygenic Risk Score Models
2021–2024	Wenjin Gu (Bioinformatics, University of Michigan, Committee Member)
	Development of Viral Integration Analysis Technologies for Virus-Associated Cancer Research
2018–2023	Rucheng Diao (Bioinformatics, University of Michigan, Committee Member)
	Local Chromatin Environments Shape Transcription and Adaptive Immunity in Bacteria
2021–2023	Zijun Gao (Bioinformatics, University of Michigan, Committee Member)
	Advance Machine Learning and Image Analysis Methods for Clinical Decision Support in Cardio-
0010 0000	vascular and Pulmonary Diseases  Nanying (Samuel) Theo (Bioinformation University of Michigan Chair)
2018–2023	Nanxiang (Samuel) Zhao (Bioinformatics, University of Michigan, Chair)
	Decoding Regulatory Variants with Computational Methods in Non-coding Regions of the Human Genome
0000 0000	Ashley Melnick (Cellular and Molecular Biology, University of Michigan, Committee Member)
2020–2023	Cdc73 Protects Notch-Induced Leukemia Cells From DNA Damage and Mitochondrial Stress
2016–2023	Christopher Castro (Bioinformatics, University of Michigan, Chair)
2010-2023	Investigating the Role of Noncoding De Novo Single-Nucleotide Variants in Autism Spectrum Dis-
	order
2017-2023	
2017–2023	Melissa Englund (Genetics and Genomics, University of Michigan, Chair)
2017–2023 2018–2023	

	Epigenetic reprogramming in mutant IDH1 glioma influences radioresistance and neural lineage differentiation
2019–2023	Benjamin Yang (Biomedical Engineering, University of Michigan, Committee Member)
2018–2022	Towards Defining Principles of Cell Fate Plasticity  Marcus Sherman (Bioinformatics, University of Michigan, Committee Member)
2010 2022	Cultivation of enhanced bioinformatic-specific pedagogical manipulatives, interventions, and professional development
2021–2022	Kuan-Han Hank Wu (Bioinformatics, University of Michigan, Committee Member)
	Integrating Electronic Health Records with Genetic Information to Advance Precision Medicine Approaches in Cardiovascular Disease
2017–2022	Amanda Moccia (Genetics and Genomics, University of Michigan, Committee Member) Investigation of Developmental Disorders: Genetic Discovery and Functional Validation
2017–2022	Ningxin Ouyang (Bioinformatics, University of Michigan, Chair)
	Deciphering Transcriptional Regulatory Circuits: Transcription Factor Binding and Regulatory Variants Identification
2015–2021	Torrin McDonald (Genetics and Genomics, University of Michigan, Chair)
	Leveraging New Technologies to Explore Regulatory and Structural Elements of the Human Genome
2018–2021	Heming Yao (Bioinformatics, University of Michigan, Committee Member)
	Machine Learning and Image Processing for Clinical Outcome Prediction: Applications in Medical
0016 0001	Data from Patients with Traumatic Brain Injury, Ulcerative Colitis, and Heart Failure  Mohd Hafiz Bin Mohd Rothi (Molecular, Cellular, and Developmental Biology, University of Michi-
2016–2021	gan, Committee Member)
	Control of Chromatin by RNA-mediated Transcriptional Silencing
2016–2021	Shengcheng Dong (Bioinformatics, University of Michigan, Chair)
	Computational Methods to Identify Regulatory Variants in the Non-coding Regions of the Human Genome
2017–2021	Steven Romanelli (Molecular & Integrative Physiology, University of Michigan, Committee Mem-
	ber)
	Viral CRISPR/Cas9 Gene Transfer for Somatic Knockout in Brown Adipose Tissue
2018–2021	Negar Farzaneh (Bioinformatics, University of Michigan, Committee Member)
0016 0000	Automated Decision Support System for Traumatic Injuries Shriya Sethuraman (Bioinformatics, University of Michigan, co-Chair)
2016–2020	Genome-wide Identification of Non-coding Transcription by RNA Polymerase V and Its Involvement
	in Transcriptional Gene Silencing
2015–2020	Sierra Nishizaki (Genetics and Genomics, University of Michigan, Chair)
	Decoding the Non-coding Genome: Novel Technologies for the Characterization of Non-coding
	Elements and Variation
2017–2020	Christopher Lee (Biostatistics, University of Michigan, Committee Member)
	Improvements and Developments in Gene Regulation and Single-Cell Gene Expression Data
2018–2019	Analysis Christine Ziegler (Biological Chemistry, University of Michigan, Committee Member)
2015–2019	Ari Allyn-Feuer (Bioinformatics, University of Michigan, Committee Member)
2010 2010	The Pharmacoepigenomics Informatics Pipeline and H-GREEN Hi-C Compiler: Discovering Pharmacogenomic Variants and Pathways with the Epigenome and Spatial Genome
2015–2017	Raymond Cavalcante (Bioinformatics, University of Michigan, Committee Member)
	Beyond the Transcriptome: Facilitating Interpretation of Epigenomics and Metabolomics Data
2015–2017	Zhengting Zou (Bioinformatics, University of Michigan, Committee Member)
	Model-based genomic studies of protein sequence evolution: convergence, epistasis, and amino
	acid acceptance rates

### Preliminary Exam Committees (n=44)

2025	Mitchell Witt (Bioinformatics, University of Michigan)
2024	Luke Gohmann (Cellular and Molecular Biology, University of Michigan)
2024	Benjamin Li (Bioinformatics, University of Michigan)
2024	Tiffany Wan (Bioinformatics, University of Michigan)
2024	Zhiyuan Yu (Bioinformatics, University of Michigan)

Rebecca McAvoy (Molecular, Cellular, and Developmental Biology, University of Michigan) 2024 Bonje Obua (Cellular and Molecular Biology, University of Michigan) 2024 Abigail Vallie (Cellular and Molecular Biology, University of Michigan) 2024 2023 Jinhao Wang (Bioinformatics, University of Michigan) Lishi Yin (Bioinformatics, University of Michigan) 2023 2023 Matthew Hodgman (Bioinformatics, University of Michigan) Ilakkiya Venkatachalam (Genetics and Genomics, University of Michigan) 2023 Jianhui Gong (Bioinformatics, University of Michigan) 2023 Mahnoor Gondal (Bioinformatics, University of Michigan) 2023 Elysia Chou (Bioinformatics, University of Michigan) 2023 Sean Moran (Bioinformatics, University of Michigan) 2022 Lu Lu (Bioinformatics, University of Michigan) 2022 Linghua Jiang (Bioinformatics, University of Michigan) 2022 Kaiwen Deng (Bioinformatics, University of Michigan) 2022 Yufeng Zhang (Bioinformatics, University of Michigan) 2022 Anthony Nguyen (Human Genetics, University of Michigan) 2021 Hanbyul Cho (Bioinformatics, University of Michigan) 2021 Charles Ryan (Cellular and Molecular Biology, University of Michigan) 2021 Kuan-Han Wu (Bioinformatics, University of Michigan) 2021 Wenjin Gu (Bioinformatics, University of Michigan) 2021 2020 Jie Cao (Bioinformatics, University of Michigan) Zijun Gao (Bioinformatics, University of Michigan) 2020 Ashley Melnick (Cellular and Molecular Biology, University of Michigan) 2020 Benjamin Yang (Biomedical Engineering, University of Michigan) 2019 Maria Virgilio (Cellular and Molecular Biology, University of Michigan) 2019 Zhi Carrie Li (Bioinformatics, University of Michigan) 2018 2018 Kevin Hu (Bioinformatics, University of Michigan) Siyu Liu (Bioinformatics, University of Michigan) 2018 Alexandra Weber (Bioinformatics, University of Michigan) 2018 Mitch Fernandez (Bioinformatics, University of Michigan) 2018 Tingyang Li (Bioinformatics, University of Michigan) 2017 2017 Marcus Sherman (Bioinformatics, University of Michigan) Adrienne Shami (Human Genetics, University of Michigan) 2017 Trenton Frisbie (Human Genetics, University of Michigan) 2017 Melissa Englund (Human Genetics, University of Michigan) 2017 Peter Orchard (Bioinformatics, University of Michigan) 2017 Li Guan (Bioinformatics, University of Michigan) 2017 2016 Shriya Sethuraman (Bioinformatics, University of Michigan) Jed Carlson (Bioinformatics, University of Michigan) 2016

#### **Industry Experience**

2013–2014 Consultant, Color Genomics
Personalized medicine / genomics startup

#### **Publications**

\* Indicates co-first authorship † Indicates co-senior authorship underscore indicates lab members

- [1] Miller SL, Green KM, Crone B, Switzenberg JA, Tank EH, Krans A, Jansex-West KR, Wieland CM, Ji EW, Petrucelli L, Barmada S, **Boyle AP**, Todd PK. "Cryptic intronic transcriptional initiation generates efficient endogenous mRNA templates for C9orf72-associated RAN translation." *Submitted*.
- [2] McBean B, Abou Zeidane R, Lichtman-Mikol S, Hauk B, Speers J, Tidmore S, Flores CL, Rana PS, Pisano C, Liu M, Santola A, Montero A, Boyle AP, Speers CW. "MELK as a Mediator of Stemness and Metastasis in Aggressive Subtypes of Breast Cancer." *International Journal of Molecular Sciences* 2025, 26(5):2245. PMID:

[3] Lee S, McAfee JC, Lee J, Gomez A, Ledford AT, Clarke D, Min H, Gerstein MB, Boyle AP, Sullivan PF, Beltran AS, Won H. "Massively parallel reporter assay investigates shared genetic variants of eight psychiatric disorders." Cell 2025, S0092-8674(24):01435–1. PMID: 39848247.

- [4] \*Van Deynze K, \*Mumm C, Maltby CJ, Switzenberg JA, Todd P, Boyle AP. "Enhanced detection and genotyping of disease-associated tandem repeats using HMMSTR and targeted long-read sequencing." *Nucleic Acids Research* 2025, 53(2):gkae1202. PMID: 39676678.
- [5] \*Zhou W, \*Mumm C, \*Gan Y, Switzenberg JA, Wang J, De Oliveira P, Kathuria K, Losh SJ, McDonald TL, Bessell B, Van Deynze K, McConnell MJ, Boyle AP, Mills RE. "A personalized multi-platform assessment of somatic mosaicism in the human frontal cortex." bioRxiv 2024.
- [6] Maltby CJ, Krans A, Grudzien SJ, Palacios Y, Muiños J, Suárez A, Asher M, Willey S, Van Deynze K, Mumm C, Boyle AP, Cortese A, Khurana V, Barmada SJ, Dijkstra AA, Todd PK. "AAGGG repeat expansions trigger RFC1-independent synaptic dysregulation in human CANVAS neurons." *Science Advances* 2024, 10(36):eadn2321. PMID: 39231235.
- [7] IGVF Consortium. "Deciphering the impact of genomic variation on function." *Nature* 2024, 633:47–57. PMID: 39232149.
- [8] Yee C, Xiao Y, Chen H, Reddy A, Xu B, Medwig-Kinney T, Zhang W, Boyle AP, Herbst W, Xiang Y, Matus D, Shen K. "An activity-regulated transcriptional program directly drives synaptogenesis." *Nature Neuroscience* 2024, 27(9):1695–1707. PMID: 39103556.
- [9] Parana P, Mumm C, McConnell MJ, **Boyle AP**. "Draft De-Novo Genome Construction of Scytonema sp. PRP1: Isolated from Single-Cell Amplification of Human Neurons." *Submitted* 2024.
- [10] Oh JW, Choi YA, Lim NS, Zhao B, Voshall A, Abyzov A, Antonacci-Fulton L, Aparicio S, Ardlie K, Bell T, Bennett J, Bernstein B, Blanchard T, Boyle AP, Buenrostro J, Burns K, Chen F, Chen R, Choudhury S, vardhan Doddapaneni H, Eichler E, Evrony G, Faith M, Fazzio T, Fulton R, Garber M, Gehlenborg N, Germer S, Getz G, Gibbs R, Hernandez R, Jin F, Korbel J, Landau D, Lawson H, Lennon N, Li H, Li Y, Loh PR, Marth G, McConnell M, Mills R, Montgomery S, Natarajan P, Park P, Satija R, Sedlazeck F, Shao D, Shen H, Stergachis A, Underhill H, Urban A, VonDran M, Walsh C, Wang T, Wu T, Zong C, Lee E, Vaccarino F, Coorens T. "The Somatic Mosaicism across Human Tissues Network." Nature, Submitted.
- [11] <u>Crone B</u>, **Boyle AP**. "Enhancing portability of trans-ancestral polygenic risk scores through tissue-specific functional genomic data integration." *PLoS Genetics* 2024, 20(8):e1011356. PMID: 39110742.
- [12] The Critical Assessment of Genome Interpretation Consortium. "CAGI, the Critical Assessment of Genome Interpretation, establishes progress and prospects for computational genetic variant interpretation methods." *Genome Biology* 2024, 25:53. PMID: 38389099.
- [13] Zhao N, Dong S, Boyle AP. "Organ-specific prioritization and annotation of non-coding regulatory variants in the human genome." *bioRxiv* 2023.
- [14] Zhao N, Wang S, Huang Q, Dong S, **Boyle AP**. "Explain-seq: an end-to-end pipeline from training to interpretation of sequence-based deep learning models." *bioRxiv* 2023.
- [15] Holmes MJ, Mahjour B, Castro CP, Farnum GA, Diehl AG, Boyle AP. "HaplotagLR: an efficient and configurable utility for haplotagging long reads." *PLoS ONE* 2024, 19(3):1–15. PMID: 38478504.
- [16] McAfee JC, Lee S, Lee J, Bell JL, Krupa O, Davis J, Insigne K, Bond ML, Zhao N, Boyle AP, Phanstiel DH, Love MI, Stein JL, Ruzicka WB, Davila-Velderrain J, Kosuri S, Won H. "Systematic investigation of allelic regulatory activity of schizophrenia-associated common variants." *Cell Genomics* 2023, 3:100404. PMID: 37868037.
- [17] Moritz L, Schon SB, Rabbani M, Sheng Y, Agrawal R, Glass-Klaiber J, Sultan C, M CJ, Clements J, Baldwin MR, <u>Diehl AG</u>, **Boyle AP**, O'Brien PJ, Ragunathan K, Hu YC, Kelleher NL, Nandakumar J, Li JZ, Orwig KE, Redding S, Hammoud SS. "Sperm chromatin structure and reproductive fitness are altered by substitution of a single amino acid in mouse protamine 1." *Nature Structural & Molecular Biology* 2023. PMID: 37460896.
- [18] <u>Castro CP</u>, <u>Diehl AG</u>, **Boyle AP**. "Challenges in screening for de novo noncoding variants contributing to genetically complex phenotypes." *Human Genetics and Genomics Advances* 2023, 4(3):100210. PMID: 37305558.

[19] \*Mumm C, \*Drexel ML, McDonald TL, Diehl AG, Switzenberg JA, Boyle AP. "OnRamp: rapid nanopore plasmid validation." *Genome Research* 2023, 33(5):741–749. PMID: 37156622.

- [20] \*Dong S, \*Zhao N, Spragins E, Kagda MS, Li M, Assis PR, Jolanki O, Luo Y, Cherry JM, †Boyle AP, †Hitz BC. "Annotating and prioritizing human non-coding variants with RegulomeDB v.2." *Nature Genetics* 2023, 55(5):724–726. PMID: 37173523.
- [21] Ouyang N, **Boyle AP**. "Quantitative assessment of association between noncoding variants and transcription factor binding." *bioRxiv* 2022.
- [22] Nishizaki SS, Boyle AP. "SEMplMe: A tool for integrating DNA methylation effects in transcription factor binding affinity predictions." *BMC Bioinformatics* 2022, 23:317. PMID: 35927613.
- [23] Qin T, Lee C, Li S, Cavalcante RG, Orchard P, Yao H, Zhang H, Wang S, Patil S, **Boyle AP**, Sartor MA. "Comprehensive enhancer-target gene assignments improve gene set level interpretation of genome-wide regulatory data." *Genome Biology* 2022, 23:105. PMID: 35473573.
- [24] Bao Y, Wadden J, Erb-Downward JR, Ranjan P, Zhou W, McDonald TL, Mills RE, Boyle AP, Dickson RP, Blaauw D, Welch JD. "SquiggleNet: real-time, direct classification of nanopore signals." Genome Biology 2021, 22:298. PMID: 34706748.
- [25] Dong S, **Boyle AP**. "Prioritization of regulatory variants with tissue-specific function in the non-coding regions of human genome." *Nucleic Acids Research* 2021, 50:e6–e6. PMID: 34648033.
- [26] \*McDonald TL, \*Zhou W, Castro CP, Mumm C, Switzenberg JA, †Mills RE, †Boyle AP. "Cas9 targeted enrichment of mobile elements using nanopore sequencing." *Nature Communications* 2021, 12:3586. PMID: 34117247.
- [27] \*Nishizaki SS, \*McDonald TL, Farnum GA, Holmes MJ, Drexel ML, Switzenberg JA, Boyle AP. "The inducible lac operator-repressor system is functional in zebrafish cells." *Frontiers in Genetics* 2021, 12. PMID: 34220959.
- [28] Zhao N, Boyle AP. "F-Seq2: improving the feature density based peak caller with dynamic statistics." *NAR Genomics and Bioinformatics* 2021, 3. PMID: 33655209.
- [29] \*Tsuzuki M, \*Sethuraman S, Coke AN, Rothi MH, Boyle AP, Wierzbicki AT. "Broad noncoding transcription suggests genome surveillance by RNA polymerase V." Proceedings of the National Academy of Sciences 2020, 117(48):30799–30804. PMID: 33199612.
- [30] <u>Diehl AG</u>, **Boyle AP**. "MapGL: Inferring evolutionary gain and loss of short genomic sequence features by phylogenetic maximum parsimony." **BMC Bioinformatics** 2020, 21:416. PMID: 32962625.
- [31] Rothi MH, <u>Sethuraman S</u>, Dolata J, **Boyle AP**, Wierzbicki AT. "DNA methylation directs nucleosome positioning in RNA-mediated transcriptional silencing." *bioRxiv* 2020.
- [32] The ENCODE Project Consortium. "Perspectives on ENCODE." *Nature* 2020, 583(7818):693–698. PMID: 32728248.
- [33] The ENCODE Project Consortium. "Expanded encyclopaedias of DNA elements in the human and mouse genomes." *Nature* 2020, 583(7818):699–710. PMID: 32728249.
- [34] Ouyang N, **Boyle AP**. "TRACE: transcription factor footprinting using chromatin accessibility data and DNA sequence." *Genome Research* 2020, 30(7):1040–1046. PMID: 32660981.
- [35] <u>Diehl AG</u>, <u>Ouyang N</u>, **Boyle AP**. "Transposable elements contribute to cell and species-specific chromatin looping and gene regulation in mammalian genomes." *Nature Communications* 2020, 11:1796. PMID: 32286261.
- [36] Lee CT, Cavalcante RG, Lee C, Qin T, Patil S, Wang S, Tsai Z, Boyle AP, Sartor MA. "Poly-Enrich: count-based methods for gene set enrichment testing with genomic regions." NAR Genomics and Bioinformatics 2020, 2. PMID: 32051932.
- [37] Nishizaki SS, Ng N, Dong S, Porter RS, Morterud C, Williams C, Asman C, Switzenberg JA, Boyle AP. "Predicting the effects of SNPs on transcription factor binding affinity." *Bioinformatics* 2019, 50:2434. PMID: 31373606.

[38] <u>Diehl AG</u>, **Boyle AP**. "CGIMP: Real-time exploration and covariate projection for self-organizing map datasets." *Journal of Open Source Software* 2019, 4(39):1520.

- [39] Amemiya HM, †Kundaje A, †**Boyle AP**. "The ENCODE Blacklist: Identification of Problematic Regions of the Genome." *Scientific Reports* 2019, 9:9354. PMID: 31249361.
- [40] Dong S, **Boyle AP**. "Predicting functional variants in enhancer and promoter elements using RegulomeDB." **Human Mutation** 2019, 33(8):831. PMID: 31228310.
- [41] Shigaki D, Adato O, Adhikar AN, <u>Dong S</u>, Hawkins-Hooker A, Inoue F, Juven-Gershon T, Kenlay H, Martin B, Patra A, Penzar DP, Schubach M, Xiong C, Yan Z, **Boyle AP**, Kreimer A, Kulakovskiy IV, Reid J, Unger R, Yosef N, Shendure J, Ahituv N, Kircher M, Beer MA. "Integration of Multiple Epigenomic Marks Improves Prediction of Variant Impact in Saturation Mutagenesis Reporter Assay." *Human mutation* 2019, 33(8):831. PMID: 31106481.
- [42] Varshney A, VanRenterghem H, Orchard P, †Boyle AP, †Stitzel ML, †Ucar D, Parker SC. "Cell specificity of regulatory annotations and their genetic effects on gene expression." *Genetics* 2019, 211(2):549–562. PMID: 30593493.
- [43] <u>Diehl AG</u>, **Boyle AP**. "Conserved and species-specific transcription factor co-binding patterns drive divergent gene regulation in human and mouse." *Nucleic Acids Research* 2018, 46(4):1878–1894. PMID: 29361190.
- [44] Nielsen JB, Fritsche LG, Zhou W, Teslovich TM, Holmen OL, Gustafsson S, Gabrielsen ME, Schmidt EM, Beaumont R, Wolford BN, Lin M, Brummett CM, Preuss MH, Refsgaard L, Bottinger EP, Graham SE, Surakka I, Chu Y, Skogholt AH, Dalen H, Boyle AP, Oral H, Herron TJ, Kitzman J, Jalife J, Svendsen JH, Olesen MS, Njølstad I, Løchen ML, Baras A, Gottesman O, Marcketta A, O'Dushlaine C, Ritchie MD, Wilsgaard T, Loos RJF, Frayling TM, Boehnke M, Ingelsson E, Carey DJ, Dewey FE, Kang HM, Abecasis GR, Hveem K, Willer CJ. "Genome-wide Study of Atrial Fibrillation Identifies Seven Risk Loci and Highlights Biological Pathways and Regulatory Elements Involved in Cardiac Development." American Journal of Human Genetics 2017, 102:103–115. PMID: 29290336.
- [45] Spadafore M, Najarian K, **Boyle AP**. "A proximity-based graph clustering method for the identification and application of transcription factor clusters." *BMC Bioinformatics* 2017, 18:530. PMID: 29187152.
- [46] \*Yang B, \*Zhou W, \*Jiao J, Nielsen JB, Mathis MR, Heydarpour M, Lettre G, Folkersen L, Prakash S, Schurmann C, Fritsche L, <u>Farnum GA</u>, Lin M, Othman M, Hornsby W, Driscoll A, Levasseur A, Thomas M, Farhat L, Dubé MP, Isselbacher EM, Franco-Cereceda A, Guo Dc, Bottinger EP, Deeb GM, Booher A, Kheterpal S, Chen YE, Kang HM, Kitzman J, Cordell HJ, Keavney BD, Goodship JA, Ganesh SK, Abecasis G, Eagle KA, **Boyle AP**, Loos RJF, †Eriksson P, †Tardif JC, †Brummett CM, †Milewicz DM, †Body SC, †Willer CJ. "Protein-altering and regulatory genetic variants near GATA4 implicated in bicuspid aortic valve." *Nature Communications* 2017, 8:15481. PMID: 28541271.
- [47] Nishizaki SS, Boyle AP. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.
- [48] Diehl AG, Boyle AP. "Deciphering ENCODE." Trends in Genetics 2016, 32(4):238-249. PMID: 26962025.
- [49] Phanstiel DH, Boyle AP, Heidari N, Snyder MP. "Mango: A bias correcting ChIA-PET analysis pipeline." Bioinformatics 2015. PMID: 26034063.
- [50] \*Cheng Y, \*Ma Z, Kim BH, Wu W, Cayting P, Boyle AP, Sundaram V, Xing X, Dogan N, Li J, Euskirchen G, Lin S, Lin Y, Visel A, Kawli T, Yang X, Patacsil D, Keller CA, Giardine B, Mouse ENCODE Consortium, Kundaje A, Wang T, Pennacchio LA, Weng Z, †Hardison RC, †Snyder MP. "Principles of regulatory information conservation between mouse and human." *Nature* 2014, 515(7527):371–375. PMID: 25409826.
- [51] \*Yue F, \*Cheng Y, \*Breschi A, \*Vierstra J, \*Wu W, \*Ryba T, \*Sandstrom R, \*Ma Z, \*Davis C, \*Pope BD, \*Shen Y, Pervouchine DD, Djebali S, Thurman RE, Kaul R, Rynes E, Kirilusha A, Marinov GK, Williams BA, Trout D, Amrhein H, Fisher-Aylor K, Antoshechkin I, DeSalvo G, See LH, Fastuca M, Drenkow J, Zaleski C, Dobin A, Prieto P, Lagarde J, Bussotti G, Tanzer A, Denas O, Li K, Bender MA, Zhang M, Byron R, Groudine MT, McCleary D, Pham L, Ye Z, Kuan S, Edsall L, Wu YC, Rasmussen MD, Bansal MS, Kellis M, Keller CA, Morrissey CS, Mishra T, Jain D, Dogan N, Harris RS, Cayting P, Kawli T, Boyle AP, Euskirchen G, Kundaje A, Lin S, Lin Y, Jansen C, Malladi VS, Cline MS, Erickson DT, Kirkup VM, Learned K, Sloan CA, Rosenbloom KR, Lacerda de Sousa

B, Beal K, Pignatelli M, Flicek P, Lian J, Kahveci T, Lee D, Kent WJ, Ramalho Santos M, Herrero J, Notredame C, Johnson A, Vong S, Lee K, Bates D, Neri F, Diegel M, Canfield T, Sabo PJ, Wilken MS, Reh TA, Giste E, Shafer A, Kutyavin T, Haugen E, Dunn D, Reynolds AP, Neph S, Humbert R, Hansen RS, De Bruijn M, Selleri L, Rudensky A, Josefowicz S, Samstein R, Eichler EE, Orkin SH, Levasseur D, Papayannopoulou T, Chang KH, Skoultchi A, Gosh S, Disteche C, Treuting P, Wang Y, Weiss MJ, Blobel GA, Cao X, Zhong S, Wang T, Good PJ, Lowdon RF, Adams LB, Zhou XQ, Pazin MJ, Feingold EA, Wold B, Taylor J, Mortazavi A, Weissman SM, Stamatoyannopoulos JA, Snyder MP, Guigo R, Gingeras TR, Gilbert DM, Hardison RC, Beer MA, Ren B, Mouse ENCODE Consortium. "A comparative encyclopedia of DNA elements in the mouse genome." *Nature* 2014, 515(7527):355–364. PMID: 25409824.

- [52] \*Boyle AP, \*Araya CL, Brdlik C, Cayting P, Cheng C, Cheng Y, Gardner K, Hillier LW, Janette J, Jiang L, Kasper D, Kawli T, Kheradpour P, Kundaje A, Li JJ, Ma L, Niu W, Rehm EJ, Rozowsky J, Slattery M, Spokony R, Terrell R, Vafeados D, Wang D, Weisdepp P, Wu YC, Xie D, Yan KK, Feingold EA, Good PJ, Pazin MJ, Huang H, Bickel PJ, Brenner SE, Reinke V, Waterston RH, Gerstein M, †White KP, †Kellis M, †Snyder M. "Comparative analysis of regulatory information and circuits across distant species." Nature 2014, 512(7515):453–456. PMID: 25164757.
- [53] Araya CL, Kawli T, Kundaje A, Jiang L, Wu B, Vafeados D, Terrell R, Weissdepp P, Gevirtzman L, Mace D, Niu W, Boyle AP, Xie D, Ma L, Murray JI, Reinke V, Waterston RH, Snyder M. "Regulatory analysis of the C. elegans genome with spatiotemporal resolution." *Nature* 2014, 512(7515):400–405. PMID: 25164749.
- [54] Phanstiel DH, **Boyle AP**, Araya CL, Snyder MP. "Sushi.R: flexible, quantitative and integrative genomic visualizations for publication-quality multi-panel figures." *Bioinformatics* 2014. PMID: 24903420.
- [55] \*Xie D, \*Boyle AP, \*Wu L, Kawli T, Zhai J, Snyder M. "Dynamic trans-acting factor colocalization in human cells." *Cell* 2013, 155(3):713–724. PMID: 24243024.
- [56] \*Kasowski M, \*Kyriazopoulou-Panagiotopoulou S, \*Grubert F, \*Zaugg JB, \*Kundaje A, Liu Y, Boyle AP, Zhang QC, Zakharia F, Spacek DV, Li J, Xie D, Steinmetz LM, Hogenesch JB, Kellis M, Batzoglou S, Snyder M. "Extensive variation in chromatin states across humans." *Science* 2013, 342(6159):750–752. PMID: 24136358.
- [57] **Boyle AP**, Hong EL, Hariharan M, Cheng Y, Schaub MA, Kasowski M, Karczewski KJ, Park J, Hitz BC, Weng S, Cherry JM, Snyder M. "Annotation of functional variation in personal genomes using RegulomeDB." *Genome Research* 2012, 22(9):1790–1797. PMID: 22955989.
- [58] Schaub MA, **Boyle AP**, Kundaje A, †Batzoglou S, †Snyder M. "Linking disease associations with regulatory information in the human genome." *Genome Research* 2012, 22(9):1748–1759. PMID: 22955986.
- [59] The ENCODE Project Consortium. "An integrated encyclopedia of DNA elements in the human genome." *Nature* 2012, 489(7414):57–74. PMID: 22955616.
- [60] \*Gerstein MB, \*Kundaje A, \*Hariharan M, \*Landt SG, \*Yan KK, \*Cheng C, \*Mu XJ, \*Khurana E, \*Rozowsky J, \*Alexander R, \*Min R, \*Alves P, Abyzov A, Addleman N, Bhardwaj N, Boyle AP, Cayting P, Charos A, Chen DZ, Cheng Y, Clarke D, Eastman C, Euskirchen G, Frietze S, Fu Y, Gertz J, Grubert F, Harmanci A, Jain P, Kasowski M, Lacroute P, Leng J, Lian J, Monahan H, O'Geen H, Ouyang Z, Partridge EC, Patacsil D, Pauli F, Raha D, Ramirez L, Reddy TE, Reed B, Shi M, Slifer T, Wang J, Wu L, Yang X, Yip KY, Zilberman-Schapira G, Batzoglou S, Sidow A, Farnham PJ, Myers RM, Weissman SM, Snyder M. "Architecture of the human regulatory network derived from ENCODE data." Nature 2012, 489(7414):91–100. PMID: 22955619.
- [61] \*Chen R, \*Mias GI, \*Li-Pook-Than J, \*Jiang L, Lam HYK, Chen R, Miriami E, Karczewski KJ, Hariharan M, Dewey FE, Cheng Y, Clark MJ, Im H, Habegger L, Balasubramanian S, O'Huallachain M, Dudley JT, Hillenmeyer S, Haraksingh R, Sharon D, Euskirchen G, Lacroute P, Bettinger K, **Boyle AP**, Kasowski M, Grubert F, Seki S, Garcia M, Whirl-Carrillo M, Gallardo M, Blasco MA, Greenberg PL, Snyder P, Klein TE, Altman RB, Butte AJ, Ashley EA, Gerstein M, Nadeau KC, Tang H, Snyder M. "Personal omics profiling reveals dynamic molecular and medical phenotypes." *Cell* 2012, 148(6):1293–1307. PMID: 22424236.
- [62] \*Song L, \*Zhang Z, \*Grasfeder LL, \*Boyle AP, \*Giresi PG, \*Lee B, \*Sheffield NC, Graff S, Huss M, Keefe D, Liu Z, London D, McDaniell RM, Shibata Y, Showers KA, Simon JM, Vales T, Wang T, Winter D, Zhang Z, Clarke ND, †Birney E, †Iyer VR, †Crawford GE, †Lieb JD, †Furey TS. "Open chromatin defined by DNasel and FAIRE identifies regulatory elements that shape cell-type identity." *Genome Research* 2011, 21(10):1757–1767. PMID: 21750106.

[63] The ENCODE Project Consortium. "A user's guide to the encyclopedia of DNA elements (ENCODE)." *PLoS Biology* 2011, 9(4):e1001046. PMID: 21526222.

- [64] **Boyle AP**, Song L, Lee B, London D, Keefe D, Birney E, Iyer VR, †Crawford GE, †Furey TS. "High-resolution genome-wide in vivo footprinting of diverse transcription factors in human cells." *Genome Research* 2011, 21:456–464. PMID: 21106903.
- [65] \*Stitzel ML, \*Sethupathy P, Pearson DS, Chines PS, Song L, Erdos MR, Welch R, Parker SCJ, Boyle AP, Scott LJ, Margulies EH, Boehnke M, Furey TS, Crawford GE, Collins FS. "Global epigenomic analysis of primary human pancreatic islets provides insights into type 2 diabetes susceptibility loci." *Cell Metabolism* 2010, 12(5):443–455. PMID: 21035756.
- [66] McDaniell R, Lee B, Song L, Liu Z, Boyle AP, Erdos MR, Scott LJ, Morken MA, Kucera KS, Battenhouse A, Keefe D, Collins FS, Willard HF, Lieb JD, Furey TS, †Crawford GE, †lyer VR, †Birney E. "Heritable individual-specific and allele-specific chromatin signatures in humans." *Science* 2010, 328(5975):235–239. PMID: 20299549.
- [67] Georgiev S, Boyle AP, Jayasurya K, Mukherjee S, Ohler U. "Evidence-ranked motif identification." Genome Biology 2010, 11(2):R19. PMID: 20156354.
- [68] Babbitt CC, Fedrigo O, Pfefferle AD, Boyle AP, Horvath JE, Furey TS, Wray GA. "Both noncoding and protein-coding RNAs contribute to gene expression evolution in the primate brain." *Genome Biology and Evolution* 2010, 2:67–79. PMID: 20333225.
- [69] Xu X, Tsumagari K, Sowden J, Tawil R, Boyle AP, Song L, Furey TS, Crawford GE, Ehrlich M. "DNasel hypersensitivity at gene-poor, FSH dystrophy-linked 4q35.2." *Nucleic Acids Research* 2009, 37(22):7381–7393. PMID: 19820107.
- [70] **Boyle AP**, Furey TS. "High-resolution mapping studies of chromatin and gene regulatory elements." *Epigenomics* 2009, 1(2):319–329. PMID: 20514362.
- [71] **Boyle AP**, Guinney J, Crawford GE, Furey TS. "F-Seq: a feature density estimator for high-throughput sequence tags." *Bioinformatics* 2008, 24(21):2537–2538. PMID: 18784119.
- [72] Boyle AP, Davis S, Shulha HP, Meltzer P, Margulies EH, Weng Z, †Furey TS, †Crawford GE. "High-resolution mapping and characterization of open chromatin across the genome." Cell 2008, 132(2):311–322. PMID: 18243105.
- [73] **Boyle AP**, Boyle JA, Bridges SM. "Identification of regulatory elements in archaea using self-organizing maps." In *Proc RECOMB* 2004.
- [74] **Boyle AP**, Boyle JA. "Global analysis of microbial translation initiation regions." In *Journal of the Mississippi Academy of Sciences*, *Volume 48* 2003:138–150.
- [75] **Boyle AP**, Bridges S. "Clustering of archael gene regulatory regions." In *FASEB Journal*, *Volume 17* 2003:A985–A985.
- [76] **Boyle AP**, Boyle JA. "Visualization of aligned genomic open reading frame data." *Biochemistry and Molecular Biology Education* 2003, 31:64–68.
- [77] Wan X, Boyle JA, Bridges SM, Boyle AP. "Interactive clustering for exploration of genomic data." In *Proceedings of the Artificial Neural Networks in Engineering Conference*, Volume 12, St. Louis, MO 2002:753–758.

#### **Patents**

[1] Karczewski K, Snyder M, Butte AJ, Dudley JT, Hong E, Boyle A, Cherry MJ, Park J: **Method and System for the Use of Biomarkers for Regulatory Dysfunction in Disease** 2018.