

Alan P. Boyle

Education

2005–2009	Doctor of Philosophy , Computational Biology and Bioinformatics Duke University, Durham, NC
2001–2005	Bachelor of Science , <i>summa cum laude</i> , Biochemistry and Molecular Biology Bachelor of Science , <i>summa cum laude</i> , Computer Science Mississippi State University, Starkville, MS

Academic Appointments

2025–present	Professor with tenure , Department of Computational Medicine & Bioinformatics Professor , Department of Human Genetics
2023–present	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	Assistant Professor , Department of Human Genetics
2014–2020	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

2017–2025	U24 HG009293 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.	(Multi-PI: Boyle, Cherry)
2021–2026	U01 HG011952 NIH/NHGRI 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.	(PI: Boyle)
2022–2026	R01 GM144484 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.	(PI: Boyle)
2023–2028	UG3 NS132084 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.	(Multi-PI: Mills, Boyle, McConnell)
2022–2025	Taubman Institute Innovation Projects 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.	(co-PI: Todd, Boyle, Mills)
2021–2026	K08 HL153799 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.	(PI: Denstaedt; Consultant)
2021–2026	R01 NS122165 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.	(PI: Castro; Co-I with Effort)
2022–2026	R01 CA260677 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.	(PI: Malek; Co-I with Effort)
2023–2028	R01 NS099280 NIH/NINDS 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.	(PI: Todd; Consultant)
2024–2029	R01 DE032699 NIH/NIDCR 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.	(PI: Brenner, Mills, Spector; Co-I with Effort)

Completed

2013–2017	R00 HG007356 Pathway to Independence Award (K99/R00) 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.	(PI: Boyle)
2015–2017	FG-2015-65465 Alfred P. Sloan Foundation Fellowship in Computational & Evolutionary Molecular Biology	(PI: Boyle)
2016–2020	R01 HL130705 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 whole-genome screening.	(PI: Willer; Co-I with Effort)
2017–2018	Eleanor and Larry Jackier U-M/Technion and Weizmann Collaborative Research Grant Michigan - Israel Partnership for Research & Education Identifying novel disease related mutations in the genomic environments around Transcription 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.	(co-PI: Boyle, Mandel-Gutfreund)
2017–2024	R35 HL135824 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.	(PI: Willer; Co-I with Effort)
2019	NVIDIA GPU Grant NVIDIA Corporation	(PI: Boyle)
2017–2022	DBI-1651614 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.	(PI: Boyle)
2022–2022	R21 HG011493 S1 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.	(Multi-PI: Boyle, Mills)
2019–2022	Precision Health Investigators Award 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.	(co-PI: Todd, Boyle, Mills)
2022	NVIDIA GPU Grant NVIDIA Corporation	(PI: Boyle)
2021–2022	Cancer Center Discovery 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.	(PI: Boyle)

2020–2023	W81XWH2010336 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.	(PI: Aguilar; Co-I with Effort)
2020–2023	R21 HG011493 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.	(Multi-PI: Boyle, Mills)
2018–2024	R01 HD093570 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.	(PI: Bielas; Co-I with Effort)
2022–2024	Michigan Alzheimer's Disease Center Developmental Project 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.	(PI: Zhou; Consultant)
2022–2025	R21 CA2578964 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.	(PI: Boyle)
2021–2025	R01 HD104680 NIH/NICHD Sperm Chromatin: Implications on organismal development and fertility This project seeks to explore protamine chromatin structure in mouse sperm.	(PI: Hammoud; Co-I with Effort)

Professional Service

Service

2024–current	DHG Faculty Development Committee
2023–current	R01 Bootcamp Medical School Cohort Coach
2023–current	Somatic Mosaicism across Human Tissues (SMaHT) Consortium Steering Committee
2021–current	Impact of Genomic Variation on Function (IGVF) Consortium Steering Committee
2022–current	University of Michigan Biomedical Research Council (BMRC) (Standing Member)
2020–2022	DHG M.S. Admissions Committee
2018–current	Lab Safety Liaison for DCM&B
2017–2024	DCM&B Preliminary Exam Abstract Review Committee (PARC) [Chair 2018–2022]
2019–2020	DHG Ph.D. Admissions Committee
2017–2020	DHG Faculty Recruitment and Promotions Committee
2016–2020	DCM&B Seminar Series Committee [Chair]
2018–2019	Cellular and Molecular Biology Admissions Committee
2017–2019	EBS Faculty IT Committee
2016–2019	DCM&B Faculty Recruitment Committee
2015–2018	DCM&B Admissions Committee
2015–2017	DHG Computational Support Committee
2015, 2016, 2025	DCM&B Retreat Planning Committee Chair (including 1st annual)
2014	<i>Ad hoc</i> admissions reviewer, University of Michigan DCM&B
2008–2009	Duke Computational Biology & Bioinformatics student committee

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	<i>Ad hoc</i> reviewer (>100 verified reviews) for the journals: <i>Science</i> , <i>Nature Biotechnology</i> , <i>Nature Genetics</i> , <i>Genome Research</i> , <i>Genome Biology</i> , <i>Nature Neuroscience</i> , <i>Nature Communications</i> , <i>Nature Protocols</i> , <i>Bioinformatics</i> , <i>Nucleic Acids Research</i> , <i>BMC Biology</i> , <i>BMC Bioinformatics</i> , <i>PLOS Computational Biology</i> , <i>Oncotarget</i> , <i>Scientific Reports</i> , <i>Atherosclerosis</i> , <i>BioEssays</i> , <i>Gene</i>
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 Conference (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 <i>Bioinformatics</i>

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 Technologies 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, W22, W23, W24, W25	Bioinformatics Concepts and Algorithms (BIOINF 529) [Course Director]
F15, F16, F17, F18, F19, F20, F21	Gene Structure and Regulation (HUMGEN 541) [3 lectures + 2 discussions / yr.]
F19, F22	Research Responsibility and Ethics (PIBS 503) [1 discussion / yr.]
F21, W22, F24, W25	Genetics Student Seminar (HUMGEN 821/822) [Mentor]
F17, F18	Experimental Genetics Systems (HUMGEN 632) [Course Director]
F15, W16, F16, W17, F17, W18, F18	Bioinformatics Journal Club (BIOINF 602/603) [Course Director F18]
S17, S18	Introduction to Biocomputing Bootcamp (BIOSAT/BIOINF/HUMGEN 606) [2 full days / yr.]
F15, F16, F17	Introduction to Bioinformatics & Computational Biology (BIOINF 527) [2 lectures + 3 labs / yr.]
S15, S16, S17	Basic Biology for Graduate Students with Quantitative Training (BIOINF 523) [2 lectures / yr.]
F03	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 University (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) <i>Rackham Graduate Student Research Grant (pre-candidate)</i>
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) <i>Rackham Graduate Student Research Grant (pre-candidate)</i>
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) <i>Rackham Graduate Student Research Grant (pre-candidate)</i> <i>Rackham Graduate Student Research Grant (candidate)</i>
2022–current	Rintsen Sherpa (Ph.D. Student, Bioinformatics, University of Michigan) <i>Rackham Graduate Student Research Grant (candidate)</i>
2021–current	Kinsey Van Deynze (Ph.D. Student, Bioinformatics, University of Michigan) <i>NIH Genome Science Training Program (T32)</i> <i>Rackham Graduate Student Research Grant (pre-candidate)</i> <i>Rackham Graduate Student Research Grant (candidate)</i>
2020–current	Andrea Valenzuela (Ph.D. Student, Chemical Biology, University of Michigan) <i>NIH Cellular Biotechnology Training Program (T32)</i>
2020–current	Breanna McBean (Ph.D. Student, Genetics and Genomics, University of Michigan)

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

Additional Graduate Rotation Students (n=17)

2024	Karan Smith (Rotation Student, Cellular and Molecular Biology, University of Michigan)
2024	Jun Sik Yun (Rotation Student, Genetics and Genomics, University of Michigan)
2024	Jeremy Chen (Rotation Student, Bioinformatics, University of Michigan)
2023	Rosina Carr (Rotation Student, Bioinformatics, University of Michigan)
2023	Connor Ward (Rotation Student, Medical Science Training Program, University of Michigan)
2022	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)
2021	Amelia Lauth (Rotation Student, Cellular and Molecular Biology, University of Michigan)
2019	Margarita Brovkina (Rotation Student, Cellular and Molecular Biology, University of Michigan)
2018	Steve Ho (Rotation Student, Human Genetics, University of Michigan)
2018	Matthew Pun (Rotation Student, Medical Science Training Program, University of Michigan)
2017	Amanda Moccia (Rotation Student, Human Genetics, University of Michigan)
2017	Stephen Carney (Rotation Student, Human Genetics, University of Michigan)
2016	Tingyang Li (Rotation Student, Bioinformatics, University of Michigan)

Postdoctoral Fellows (n=3)

2023–2025	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, University of Michigan)
	<i>UROP Blue Ribbon Award</i>
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)
	<i>UROP Blue Ribbon Award</i>
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 Michigan)
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)
2012	Melanie Connick (Undergraduate, Biology, University of New Mexico)
2012	Edward Dai (Undergraduate, Computer Science, Stanford University)

Doctoral Thesis Committees (n=50)

2025–current	Brandt Bessell (Bioinformatics, University of Michigan, Committee Member)
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, Committee 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)
2021–2025	Zhenhao Zhang (Bioinformatics, University of Michigan, Committee Member) <i>Decoding Genome Regulation through Deep Learning Models</i>
2020–2025	Steve Ho (Genetics and Genomics, University of Michigan, Committee Member) <i>Applied Machine Learning for Big Data Genomics</i>
2020–2025	Camille Mumm (Genetics and Genomics, University of Michigan, Chair) <i>Exploring Repetitive Elements in Neurodegenerative Disease Using Targeted Long-Read Sequencing</i>
2022–2025	Kaiwen Deng (Bioinformatics, University of Michigan, Committee Member) <i>Advancing Computational Models and Algorithms for the Analysis of Single-Cell and Neural Data</i>
2022–2024	Franco Tavella (Biophysics, University of Michigan, Committee Member) <i>Robustness and Tunability of Biological Oscillations</i>
2018–2024	Bradley Crone (Bioinformatics, University of Michigan, Chair) <i>Computational Methods in Functional Prioritization of Polygenic Risk Score Models</i>
2021–2024	Wenjin Gu (Bioinformatics, University of Michigan, Committee Member) <i>Development of Viral Integration Analysis Technologies for Virus-Associated Cancer Research</i>
2018–2023	Rucheng Diao (Bioinformatics, University of Michigan, Committee Member) <i>Local Chromatin Environments Shape Transcription and Adaptive Immunity in Bacteria</i>
2021–2023	Zijun Gao (Bioinformatics, University of Michigan, Committee Member) <i>Advance Machine Learning and Image Analysis Methods for Clinical Decision Support in Cardiovascular and Pulmonary Diseases</i>
2018–2023	Nanxiang (Samuel) Zhao (Bioinformatics, University of Michigan, Chair) <i>Decoding Regulatory Variants with Computational Methods in Non-coding Regions of the Human Genome</i>
2020–2023	Ashley Melnick (Cellular and Molecular Biology, University of Michigan, Committee Member) <i>Cdc73 Protects Notch-Induced Leukemia Cells From DNA Damage and Mitochondrial Stress</i>
2016–2023	Christopher Castro (Bioinformatics, University of Michigan, Chair) <i>Investigating the Role of Noncoding De Novo Single-Nucleotide Variants in Autism Spectrum Disorder</i>
2017–2023	Melissa Englund (Genetics and Genomics, University of Michigan, Chair)

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

Preliminary Exam Committees (n=45)

2025	Xinyi Deng (Bioinformatics, University of Michigan)
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)
2024	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)
2023	Jinhao Wang (Bioinformatics, University of Michigan)
2023	Lishi Yin (Bioinformatics, University of Michigan)
2023	Matthew Hodgman (Bioinformatics, University of Michigan)
2023	Ilakkiya Venkatachalam (Genetics and Genomics, University of Michigan)
2023	Jianhui Gong (Bioinformatics, University of Michigan)
2023	Mahnor Gondal (Bioinformatics, University of Michigan)
2023	Elysia Chou (Bioinformatics, University of Michigan)
2022	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)
2021	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)
2020	Jie Cao (Bioinformatics, University of Michigan)
2020	Zijun Gao (Bioinformatics, University of Michigan)
2020	Ashley Melnick (Cellular and Molecular Biology, University of Michigan)
2019	Benjamin Yang (Biomedical Engineering, University of Michigan)
2019	Maria Virgilio (Cellular and Molecular Biology, University of Michigan)
2018	Zhi Carrie Li (Bioinformatics, University of Michigan)
2018	Kevin Hu (Bioinformatics, University of Michigan)
2018	Siyu Liu (Bioinformatics, University of Michigan)
2018	Alexandra Weber (Bioinformatics, University of Michigan)
2018	Mitch Fernandez (Bioinformatics, University of Michigan)
2017	Tingyang Li (Bioinformatics, University of Michigan)
2017	Marcus Sherman (Bioinformatics, University of Michigan)
2017	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)
2016	Shriya Sethuraman (Bioinformatics, University of Michigan)
2016	Jed Carlson (Bioinformatics, University of Michigan)

Industry Experience

2013–2014	Consultant, Color Genomics Personalized medicine / genomics startup
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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, Petrucci L, Barmada S, **Boyle AP**, Todd PK. "Cryptic intronic transcriptional initiation generates efficient endogenous mRNA templates for C9orf72-associated RAN translation." **Accepted, PNAS**.
- [2] Coorens THH, Oh JW, Choi YA, Lim NS, Zhao B, Voshall A, Abyzov A, Antonacci-Fulton L, Aparicio S, Ardlie KG, Bell TJ, Bennett JT, Bernstein BE, Blanchard TG, **Boyle AP**, Buenrostro JD, Burns KH, Chen F, Chen R, Choudhury S, Doddapaneni HV, Eichler EE, Evrony GD, Faith MA, Fazzio TG, Fulton RS, Garber M, Gehlenborg

- N, Germer S, Getz G, Gibbs RA, Hernandez RG, Jin F, Korbel JO, Landau DA, Lawson HA, Lennon NJ, Li H, Li Y, Loh PR, Marth G, McConnell MJ, Mills RE, Montgomery SB, Natarajan P, Park PJ, Satija R, Sedlazeck FJ, Shao DD, Shen H, Stergachis AB, Underhill HR, Urban AE, VonDran MW, Walsh CA, Wang T, Wu TP, Zong C, Lee EA, Vaccarino FM, Somatic Mosaicism across Human Tissues Network. "The Somatic Mosaicism across Human Tissues Network." **Nature** 2025, 643(8070):47–59. PMID: 40604182.
- [3] 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: 40076867.
- [4] 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.
- [5] *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.
- [6] *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.
- [7] 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.
- [8] IGVF Consortium. "Deciphering the impact of genomic variation on function." **Nature** 2024, 633:47–57. PMID: 39232149.
- [9] 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.
- [10] 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.
- [11] Crone B, **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, Diehl AG, **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] Castro CP, Diehl AG, **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] Diehl AG, **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, Sethuraman S, 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] Diehl AG, Ouyang N, **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, 35:2434. PMID: 31373606.
- [38] Diehl AG, **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, Dong S, 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] Diehl AG, **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, Wilsaard 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, Farnum GA, 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, Kuttyavin 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, Skoultschi A, Gosh S, Distech 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, Fietze 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, McDaniel 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 DNaseI 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] McDaniel 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, †Iyer 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. "DNaseI 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.