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

Associate Professor, Department of Human Genetics Affiliate, Michigan Neuroscience Institute Affiliate Member, Rogel Cancer Center Member, Cellular and Molecular Biology Program Member, Center for RNA Biomedicine Member, Genome Science Training Program (GSTP) Member, Michigan Predoctoral Training Program in Genetics (GTP)	2020-present	Associate Professor with tenure, Department of Computational Medicine & Bioinformatics
Affiliate Member, Rogel Cancer Center 2017-present 2016-present 2015-present 2015-present Member, Genome Science Training Program (GSTP) Member, Michigan Predoctoral Training Program in Genetics (GTP)		Associate Professor, Department of Human Genetics
2017-present 2016-present 2015-present 2015-present Member, Cellular and Molecular Biology Program Member, Center for RNA Biomedicine Member, Genome Science Training Program (GSTP) Member, Michigan Predoctoral Training Program in Genetics (GTP)	2021-present	Affiliate, Michigan Neuroscience Institute
Member, Center for RNA Biomedicine Member, Genome Science Training Program (GSTP) Member, Michigan Predoctoral Training Program in Genetics (GTP)	2020-present	Affiliate Member, Rogel Cancer Center
Member, Genome Science Training Program (GSTP) Member, Michigan Predoctoral Training Program in Genetics (GTP)	2017-present	Member, Cellular and Molecular Biology Program
Member, Michigan Predoctoral Training Program in Genetics (GTP)	2016-present	Member, Center for RNA Biomedicine
, g	2015-present	Member, Genome Science Training Program (GSTP)
		Member, Michigan Predoctoral Training Program in Genetics (GTP)
2014-present Member, Program in Biomedical Sciences	2014-present	Member, Program in Biomedical Sciences
Member, Bioinformatics Training Program		Member, Bioinformatics Training Program
2015–2020 Assistant Professor, Department of Human Genetics	2015–2020	Assistant Professor, Department of Human Genetics
Assistant Professor, Department of Computational Medicine & Bioinformatics	2014–2020	Assistant Professor, Department of Computational Medicine & Bioinformatics
University of Michigan, Ann Arbor, MI		University of Michigan, Ann Arbor, MI
2010–2014 Postdoctoral Scholar, Genetics	2010–2014	Postdoctoral Scholar, Genetics
Stanford University, Stanford, CA; Advisor: Dr. Michael Snyder		Stanford University, Stanford, CA; Advisor: Dr. Michael Snyder
Spring 2010 Postdoctoral Associate, Computational Biology	Spring 2010	Postdoctoral Associate. Computational Biology
Duke University, Durham, NC; Advisor: Dr. Terrence S. Furey	, 3	, ,

Scholarships, Fellowships, and Honors

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 February 1, 2023

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.

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. R21 CA2578964

(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 devel-

opment.

2022-2025

U01 HG011952 (PI: Boyle) 2021-2026

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.

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.

Precision Health Investigators Award 2019-2024

(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.

Cancer Center Discovery 2021-2022

(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.

NVIDIA GPU Grant (PI: Boyle) 2022 **NVIDIA** Corporation

2018-2024

(PI: Bielas; Co-I with Effort)

NIH/NICHD

R01 HD093570

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.

W81XWH2010336 2020-2023

(PI: Aguilar; Co-I with Effort)

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.

F32 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.

2021-2026 R01 HD104680

(PI: Hammoud; Co-I with Effort)

NIH/NICHD

Sperm Chromatin: Implications on organismal development and fertility

This project seeks to explore protamine chromatin structure in mouse sperm.

2021-2026 R01 NS122165

(PI: Castro; Co-I with Effort)

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.

2022-2026 R01 CA260677

(PI: Malek; Co-I with Effort)

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.

2022–2024 Michigan Alzheimer's Disease Center Developmental Project

(PI: Zhou; Consultant)

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.

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

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 (Supple-

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

Professional Service

Service

DHG M.S. Admissions Committee DCM&B Diversity, Equity, & Inclusion Committee [Ally/Chair 2018–2020] Lab Safety Liaison for DCM&B DCM&B Preliminary Exam Abstract Review Committee (PARC) [Chair 2018–2022] DHG Ph.D. Admissions Committee DHG Faculty Recruitment and Promotions Committee DCM&B Seminar Series Committee [Chair] Cellular and Molecular Biology Admissions Committee DCM&B Faculty IT Committee DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Faculty Recruitment Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B Duke Computational Biology & Bioinformatics student committee	2022-current	University of Michigan Biomedical Research Council (BMRC) (Standing Member)
Lab Safety Liaison for DCM&B DCM&B Preliminary Exam Abstract Review Committee (PARC) [Chair 2018–2022] DHG Ph.D. Admissions Committee DHG Faculty Recruitment and Promotions Committee DCM&B Seminar Series Committee [Chair] Cellular and Molecular Biology Admissions Committee EBS Faculty IT Committee DCM&B Faculty Recruitment Committee DCM&B Faculty Recruitment Committee DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2020-2022	DHG M.S. Admissions Committee
DCM&B Preliminary Exam Abstract Review Committee (PARC) [Chair 2018–2022] DHG Ph.D. Admissions Committee DHG Faculty Recruitment and Promotions Committee DCM&B Seminar Series Committee [Chair] Cellular and Molecular Biology Admissions Committee EBS Faculty IT Committee DCM&B Faculty Recruitment Committee DCM&B Faculty Recruitment Committee DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Retreat Planing Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2018-current	DCM&B Diversity, Equity, & Inclusion Committee [Ally/Chair 2018–2020]
DHG Ph.D. Admissions Committee DHG Faculty Recruitment and Promotions Committee DCM&B Seminar Series Committee [Chair] Cellular and Molecular Biology Admissions Committee EBS Faculty IT Committee DCM&B Faculty Recruitment Committee DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2018-current	Lab Safety Liaison for DCM&B
DHG Faculty Recruitment and Promotions Committee DCM&B Seminar Series Committee [Chair] Cellular and Molecular Biology Admissions Committee EBS Faculty IT Committee DCM&B Faculty Recruitment Committee DCM&B Faculty Recruitment Committee DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DCM&B Admissions Committee DHG Computational Support Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2017-current	DCM&B Preliminary Exam Abstract Review Committee (PARC) [Chair 2018–2022]
DCM&B Seminar Series Committee [Chair] Cellular and Molecular Biology Admissions Committee EBS Faculty IT Committee DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2019-2020	DHG Ph.D. Admissions Committee
Cellular and Molecular Biology Admissions Committee EBS Faculty IT Committee DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Admissions Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2017-2020	DHG Faculty Recruitment and Promotions Committee
EBS Faculty IT Committee DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DHG Computational Support Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2016-2020	DCM&B Seminar Series Committee [Chair]
2016–2019 DCM&B Faculty Recruitment Committee DCM&B Admissions Committee DHG Computational Support Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2018-2019	Cellular and Molecular Biology Admissions Committee
2015–2018 DCM&B Admissions Committee DHG Computational Support Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2017–2019	EBS Faculty IT Committee
DHG Computational Support Committee DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2016–2019	DCM&B Faculty Recruitment Committee
DCM&B Retreat Planing Committee Chair (including 1st annual) Ad hoc admissions reviewer, University of Michigan DCM&B	2015–2018	DCM&B Admissions Committee
Ad hoc admissions reviewer, University of Michigan DCM&B	2015–2017	DHG Computational Support Committee
	2015–2016	DCM&B Retreat Planing Committee Chair (including 1st annual)
2008–2009 Duke Computational Biology & Bioinformatics student committee	2014	Ad hoc 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	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
2019-current	Editorial Board, PLoS ONE
2019-current	Review Editor, Bioinformatics and Computational Biology for Frontiers in Genetics
2019-current	Review Editor, Bioinformatics and Computational Biology for Frontiers in Plant Science
2019-current	Review Editor, Bioinformatics and Computational Biology for Frontiers in Bioengineering and
	Biotechnology
2019	Program Committee, Studies of Phenotypes and Clinical Applications, 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-current	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

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

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

W19, W20, W21, W22, W23	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	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 (BIOSTAT/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

St Lectures / Failers		
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

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)
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, Human Genetics, University of Michigan) Joint M.S. in Bioinformatics, University of Michigan NIH Genome Science Training Program (T32) Rackham Graduate Student Research Grant (pre-candidate)
2019–2020	Monica Holmes (M.S. Student, Bioinformatics, University of Michigan)

Camille Mumm (Ph.D. Student, Human Genetics, University of Michigan) 2020-current Joint M.S. in Bioinformatics, University of Michigan NIH Genome Science Training Program (T32) Rackham Graduate Student Research Grant (pre-candidate) Bradley Crone (Ph.D. Student, Bioinformatics, University of Michigan) 2018-current Rackham Graduate Student Research Grant (candidate) Melissa Englund (Ph.D. Student, Human Genetics, University of Michigan) 2017-current NIH Human Genetics Training Program (T32) Rackham Graduate Student Research Grant (candidate) Samuel Zhao (Ph.D. Student, Bioinformatics, University of Michigan) 2018-current 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 2016-current Christopher Castro (Ph.D. Student, Bioinformatics, University of Michigan) 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, Human Genetics, 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, Human Genetics, 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

Emily Pogson (Rotation Student, Human Genetics, University of Michigan)
Katarina Pavlovic (Rotation Student, Bioinformatics, University of Michigan)
Brandt Bessell (Rotation Student, Bioinformatics, University of Michigan)
Rintsen Sherpa (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 Progran, 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

2022-current	Torrin McDonald (University of Michigan)
2021–2022	Shengcheng Dong (University of Michigan)

Non-student Lab Volunteers

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

Undergraduate and High School Students

2022-current	Summer Ann (Undergraduate, Neuroscience, University of Michigan)
2022-current	Kobe Howcroft (Undergraduate, Computer Science, University of Michigan)
2021-current	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)
2012	Melanie Connick (Undergraduate, Biology, University of New Mexico)
2012	Edward Dai (Undergraduate, Computer Science, Stanford University)

Doctoral Thesis Committees

2022-current	Kaiwen Deng (Bioinformatics, University of Michigan, Committee Memeber)
2022-current	Emily Peirent (Neuroscience, University of Michigan, Committee Memeber)
2022-current	Franco Tavella (Biophysics, University of Michigan, Committee Member)
2021-current	Zijun Gao (Bioinformatics, University of Michigan, Committee Member)
2021-current	Wenjin Gu (Bioinformatics, University of Michigan, Committee Member)
2021-current	Kinsey Van Deynze (Bioinformatics, University of Michigan, Chair)

2021-current	Mashiat Rabbani (Human Genetics, University of Michigan, Committee Member)
2020-current	Andrea Valenzuela (Chemical Biology, University of Michigan, co-Chair)
2020-current	Breanna McBean (Human Genetics, University of Michigan, co-Chair)
2020-current	Ashley Melnick (Cellular and Molecular Biology, University of Michigan, Committee Member)
2020-current	Camille Mumm (Human Genetics, University of Michigan, Chair)
2018-current	Stephen Carney (Cancer Biology, University of Michigan, Committee Member)
2018-current	Rucheng Diao (Bioinformatics, University of Michigan, Committee Member)
2018-current	Samuel Zhao (Bioinformatics, University of Michigan, Chair)
2018-current	Bradley Crone (Bioinformatics, University of Michigan, Chair)
2017-current	Melissa Englund (Human Genetics, University of Michigan, Chair)
2016-current	Christopher Castro (Bioinformatics, University of Michigan, Chair)
2019–2023	Benjamin Yang (Biomedical Engineering, University of Michigan, Committee Member)
	Towards Defining Principles of Cell Fate Plasticity
2018–2022	Marcus Sherman (Bioinformatics, University of Michigan, Committee Member)
	Cultivation of enhanced bioinformatic-specific pedagogical manipulatives, interventions, and pro-
	fessional 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 (Human Genetics, 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 Vari-
	ants Identification
2015–2021	Torrin McDonald (Human Genetics, 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
	Data from Patients with Traumatic Brain Injury, Ulcerative Colitis, and Heart Failure
2016–2021	Mohd Hafiz Bin Mohd Rothi (Molecular, Cellular, and Developmental Biology, University of Michi-
	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)
	Automated Decision Support System for Traumatic Injuries
2016–2020	Shriya Sethuraman (Bioinformatics, University of Michigan, co-Chair)
	Genome-wide Identification of Non-coding Transcription by RNA Polymerase V and Its Involvement
	in Transcriptional Gene Silencing
2015–2020	Sierra Nishizaki (Human Genetics, 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
0010 0010	Analysis Christing Ziggler (Biglegieal Chemistry, University of Mighigan, Committee Member)
2018–2019	Christine Ziegler (Biological Chemistry, University of Michigan, Committee Member)
2015–2018	Ari Allyn-Feuer (Bioinformatics, University of Michigan, Committee Member) The Pharmacoepigenomics Informatics Pipeline and H-GREEN Hi-C Compiler: Discovering Phar-
2015 2017	macogenomic Variants and Pathways with the Epigenome and Spatial Genome Raymond Cavalcante (Bioinformatics, University of Michigan, Committee Member)
2015–2017	Beyond the Transcriptome: Facilitating Interpretation of Epigenomics and Metabolomics Data
2015 2017	Zhengting Zou (Bioinformatics, University of Michigan, Committee Member)
2015–2017	Zirongung Zou (Diolinormatics, Oniversity of Milonigan, Committee Member)

Model-based genomic studies of protein sequence evolution: convergence, epistasis, and amino acid acceptance rates

Preliminary Exam Committees

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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] 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.
- [2] Holmes MJ, Mahjour B, Castro CP, Farnum GA, Diehl AG, Boyle AP. "LRphase: an efficient method for assigning haplotype identity to long reads." bioRxiv 2023.
- [3] Ouyang N, **Boyle AP**. "Quantitative assessment of association between noncoding variants and transcription factor binding." *bioRxiv* 2022.
- [4] <u>Castro CP</u>, <u>Diehl AG</u>, **Boyle AP**. "Challenges in screening for de novo noncoding variants contributing to genetically complex phenotypes." *bioRxiv* 2022.
- [5] Mumm C, Drexel ML, McDonald TL, Diehl AG, Switzenberg JA, Boyle AP. "OnRamp: rapid nanopore plasmid validation." bioRxiv 2022.
- [6] Moritz L, Schon SB, Rabbani M, Sheng Y, Pendlebury DF, Agrawal R, Sultan C, Jorgensen K, Zheng X, Diehl AG, Ragunathan K, Hu YC, Nandakumar J, Li JZ, **Boyle AP**, Orwig KE, Redding S, Hammoud SS. "Single

- residue substitution in protamine 1 disrupts sperm genome packaging and embryonic development in mice." **bioRxiv** 2021.
- [7] Rothi MH, <u>Sethuraman S</u>, Dolata J, **Boyle AP**, Wierzbicki AT. "DNA methylation directs nucleosome positioning in RNA-mediated transcriptional silencing." *bioRxiv* 2020.
- [8] *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." *In Press, Nature Genetics* 2022.
- [9] <u>Nishizaki SS</u>, **Boyle AP**. "SEMplMe: A tool for integrating DNA methylation effects in transcription factor binding affinity predictions." *BMC Bioinformatics* 2022, 23:317.
- [10] 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.
- [11] 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.
- [12] 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.
- [13] *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.
- [14] *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.
- [15] Zhao N, Boyle AP. "F-Seq2: improving the feature density based peak caller with dynamic statistics." *NAR Genomics and Bioinformatics* 2021, 3. PMID: 33655209.
- [16] *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.
- [17] <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.
- [18] The ENCODE Project Consortium. "Perspectives on ENCODE." *Nature* 2020, 583(7818):693–698. PMID: 32728248.
- [19] The ENCODE Project Consortium. "Expanded encyclopaedias of DNA elements in the human and mouse genomes." *Nature* 2020, 583(7818):699–710. PMID: 32728249.
- [20] Ouyang N, Boyle AP. "TRACE: transcription factor footprinting using chromatin accessibility data and DNA sequence." *Genome Research* 2020, 30:1040–1046. PMID: 32660981.
- [21] <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.
- [22] 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.
- [23] 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.
- [24] <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.

[25] Amemiya HM, †Kundaje A, †**Boyle AP**. "The ENCODE Blacklist: Identification of Problematic Regions of the Genome." *Scientific Reports* 2019, 9:9354. PMID: 31249361.

- [26] Dong S, **Boyle AP**. "Predicting functional variants in enhancer and promoter elements using RegulomeDB." *Human Mutation* 2019, 33(8):831. PMID: 31228310.
- [27] 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.
- [28] 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.
- [29] <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.
- [30] 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.
- [31] 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.
- [32] *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.
- [33] Nishizaki SS, Boyle AP. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.
- [34] Diehl AG, Boyle AP. "Deciphering ENCODE." Trends in Genetics 2016, 32(4):238–249. PMID: 26962025.
- [35] Phanstiel DH, Boyle AP, Heidari N, Snyder MP. "Mango: A bias correcting ChIA-PET analysis pipeline." Bioinformatics 2015. PMID: 26034063.
- [36] *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.
- [37] *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.

- [38] *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.
- [39] 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.
- [40] 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.
- [41] *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.
- [42] *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.
- [43] **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.
- [44] 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.
- [45] The ENCODE Project Consortium. "An integrated encyclopedia of DNA elements in the human genome." *Nature* 2012, 489(7414):57–74. PMID: 22955616.
- [46] *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.
- [47] *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.
- [48] *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.
- [49] The ENCODE Project Consortium. "A user's guide to the encyclopedia of DNA elements (ENCODE)." *PLoS Biology* 2011, 9(4):e1001046. PMID: 21526222.

[50] **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.

- [51] *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.
- [52] 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.
- [53] Georgiev S, Boyle AP, Jayasurya K, Mukherjee S, Ohler U. "Evidence-ranked motif identification." Genome Biology 2010, 11(2):R19. PMID: 20156354.
- [54] 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.
- [55] 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.
- [56] Boyle AP, Furey TS. "High-resolution mapping studies of chromatin and gene regulatory elements." *Epigenomics* 2009, 1(2):319–329. PMID: 20514362.
- [57] **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.
- [58] 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.
- [59] Boyle AP, Boyle JA, Bridges SM. "Identification of regulatory elements in archaea using self-organizing maps." In Proc RECOMB 2004.
- [60] **Boyle AP**, Boyle JA. "Global analysis of microbial translation initiation regions." In *Journal of the Mississippi Academy of Sciences*, *Volume 48* 2003:138–150.
- [61] **Boyle AP**, Bridges S. "Clustering of archael gene regulatory regions." In *FASEB Journal*, *Volume 17* 2003:A985–A985.
- [62] Boyle AP, Boyle JA. "Visualization of aligned genomic open reading frame data." Biochemistry and Molecular Biology Education 2003, 31:64–68.
- [63] 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

[64] 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, (Granted United States patent 9,946,835).