# 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**

2020-present	Associate Professor with tenure, Department of Computational Medicine & Bioinformatics
	Associate Professor, Department of Human Genetics
2020-present	Affiliate Member, Rogel Cancer Center
2017-present	Member, Cellular and Molecular Biology Program
2016-present	Member, Center for RNA Biomedicine
2015-present	Member, Genome Science Training Program (GSTP)
	Member, Michigan Predoctoral Training Program in Genetics (GTP)
2014-present	Member, Program in Biomedical Sciences
	Member, Bioinformatics Training Program
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
. 0	Duke University, Durham, NC; Advisor: Dr. Terrence S. Furey

# 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

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

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.

2017–2022 DBI-1651614 (PI: Boyle)

NSF/BIO/DBI

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

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

2020–2022 R21 HG011493 (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.

2019–2021 Precision Health Investigators Award (co-PI: Todd, Boyle, Mills) University of Michigan

Short Tandem repeats in precision health and human disease

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

2021–2022 Cancer Center Discovery (PI: Boyle)

University of Michigan

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

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

2018–2023 | R01 HD093570 (PI: Bielas; Co-I with Effort)

NIH/NICHD

Genetic Diagnosis of Neurodevelopmental Disorders in India

This study will establish whole-exome sequencing to study mendelian genetic disorders at the All India Institute of Medical Sciences.

2020–2023 W81XWH2010336 (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.

2021–2026 F32 HL153799 (PI: Denstaedt; Consultant)

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.

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.

Alan P. Boyle July 14, 2021

FG-2015-65465 (PI: Boyle) 2015-2017

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

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

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

(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.

R35 HL135824 2017-2024 (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.

**NVIDIA GPU Grant** (PI: Boyle) 2019

**NVIDIA** Corporation

#### **Professional Service**

#### **Service**

DCM&B Diversity, Equity, & Inclusion Committee [Ally/Chair 2018–2020] 2018-current Lab Safety Liaison for DCM&B 2018-current DCM&B Preliminary Exam Abstract Review Committee (PARC) [Chair 2018–current] 2017-current DHG M.S. Admissions Committee 2020-2021 DHG Ph.D. Admissions Committee 2019-2020 DHG Faculty Recruitment and Promotions Committee 2017-2020 DCM&B Seminar Series Committee [Chair] 2016-2020 2018-2019 Cellular and Molecular Biology Admissions Committee EBS Faculty IT Committee 2017-2019 DCM&B Faculty Recruitment Committee 2016-2019

DCM&B Admissions Committee 2015-2018

**DHG Computational Support Committee** 2015-2017

DCM&B Retreat Planing Committee Chair (including 1st annual) 2015-2016 2014 Ad hoc admissions reviewer, University of Michigan DCM&B Duke Computational Biology & Bioinformatics student committee 2008-2009

#### Memberships

Member, American Society of Human Genetics (ASHG) 2018-current Member, International Society for Computational Biology (ISCB) 2013-current Member, American Association for the Advancement of Science (AAAS) 2012-2014 Member, Gamma Sigma Delta Agricultural Honor Society 2005-current

#### Manuscript Reviewing Activity

Ad hoc reviewer (100 verified reviews) for the journals: Nature Genetics, Genome Research, Since 2009

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

Editorial Board, PLoS ONE 2019-current

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 <i>Bioinformatics</i>

# **Grant Reviewing Activity**

2021	University of Michigan internal review for Falk Trust Catalyst Award (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 (Ad Hoc)
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 (Ad Hoc)
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)

<b>J</b> (	
W19, W20, W21	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.]
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		
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**

2021-current	Kinsey Van Deynze (Ph.D. Student, Bioinformatics, University of Michigan) NIH Genome Science Training Program (T32)
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)

NIH Genome Science Training Program (T32) Monica Holmes (M.S. Student, Bioinformatics, University of Michigan) 2019-2020 Camille Mumm (Ph.D. Student, Human Genetics, University of Michigan) 2020-current NIH Genome Science Training Program (T32) Rackham Graduate Student Research Grant (pre-candidate) Bradley Crone (Ph.D. Student, Bioinformatics, University of Michigan) 2018-current 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) 2017-current 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 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 2016-2020 Shriya Sethuraman (Ph.D. Student, Bioinformatics, University of Michigan) Christopher Castro (Ph.D. Student, Bioinformatics, University of Michigan) 2016-current NIH Bioinformatics Training Program (T32) Rackham Merit Fellow Rackham Graduate Student Research Grant (pre-candidate) Global Research Engagement Opportunity Fellowship Ningxin Ouyang (Ph.D. Student, Bioinformatics, University of Michigan) 2015-current 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-current 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 NIH Genome Science Training Program (T32) Rackham Merit Fellow Rackham Summer Award Rackham Graduate Student Research Grant (candidate)

#### **Additional Graduate Rotation Students**

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	Tingvang Li (Rotation Student, Bioinformatics, University of Michigan)

## **Postdoctoral Fellows**

2021-current | Shengcheng Dong (University of Michigan)

## **Non-student Lab Volunteers**

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

## **Undergraduate and High School Students**

2021	Aryn Booker (Undergraduate, UROP Molecular, Cellular, and Developmental Biology, University
	of Michigan)
	UROP Blue Ribbon Award
2021	Preston Parana (Undergraduate, UROP Molecular, Cellular, and Developmental Biology, Univer-
	sity 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**

2021-current	Wenjin Gu (Bioinformatics, University of Michigan)
2021-current	Mashiat Rabbani (Human Genetics, University of Michigan)
2020-current	Ashley Melnick (Cellular and Molecular Biology, University of Michigan)
2019-current	Benjamin Yang (Biomedical Engineering, University of Michigan)
2018-current	Christine Ziegler (Biological Chemistry, University of Michigan)
2018-current	Heming Yao (Bioinformatics, University of Michigan)
2018-current	Stephen Carney (Cancer Biology, University of Michigan)
2018-current	Marcus Sherman (Bioinformatics, University of Michigan)
2018–2021	Negar Farzaneh (Bioinformatics, University of Michigan)
2018-current	Rucheng Diao (Bioinformatics, University of Michigan)
2017–2021	Steven Romanelli (Molecular & Integrative Physiology, University of Michigan)
2017-current	Amanda Moccia (Human Genetics, University of Michigan)
2017–2020	Christopher Lee (Biostatistics, University of Michigan)
2016-current	Mohd Hafiz Bin Mohd Rothi (Molecular, Cellular, and Developmental Biology, University of Michi-
	gan)
2015–2018	Ari Allyn-Feuer (Bioinformatics, University of Michigan)
2015–2017	Raymond Cavalcante (Bioinformatics, University of Michigan)
2015–2017	Zhengting Zou (Bioinformatics, University of Michigan)

## **Preliminary Exam Committees**

2021	Charles Ryan (Cellular and Molecular Biology, University of Michigan)
	Kuan Han Wu (Diginformatica Haiyaraity of Michigan)

2021 Kuan-Han Wu (Bioinformatics, University of Michigan)

Wenjin Gu (Bioinformatics, University of Michigan) 2021 Jie Cao (Bioinformatics, University of Michigan) 2020 Zijun Gao (Bioinformatics, University of Michigan) 2020 2020 Ashley Melnick (Cellular and Molecular Biology, University of Michigan) Benjamin Yang (Biomedical Engineering, University of Michigan) 2019 2019 Maria Virgilio (Cellular and Molecular Biology, University of Michigan) 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) 2018 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) 2017 Shriya Sethuraman (Bioinformatics, University of Michigan) 2016 Jed Carlson (Bioinformatics, University of Michigan) 2016

# **Industry Experience**

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

#### **Publications**

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

- [1] Dong S, **Boyle AP**. "Prioritization of regulatory variants with tissue-specific function in the non-coding regions of human genome." *bioRxiv* 2021.
- [2] Rothi MH, <u>Sethuraman S</u>, Dolata J, **Boyle AP**, Wierzbicki AT. "DNA methylation directs nucleosome positioning in RNA-mediated transcriptional silencing." *bioRxiv* 2020.
- [3] Qin T, Lee C, Cavalcante R, 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." bioRxiv 2020.
- [4] Nishizaki SS, Boyle AP. "SEMplMe: A tool for integrating DNA methylation effects in transcription factor binding affinity predictions." bioRxiv 2020.
- [5] 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.
- [6] \*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.
- [7] Zhao N, Boyle AP. "F-Seq2: improving the feature density based peak caller with dynamic statistics." NAR Genomics and Bioinformatics 2021, 3. PMID: 33655209.
- [8] \*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. PMID: 33199612.
- [9] <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.
- [10] The ENCODE Project Consortium. "Perspectives on ENCODE." Nature 2020, 583(7818):693–698. PMID: 32728248.

[11] The ENCODE Project Consortium. "Expanded encyclopaedias of DNA elements in the human and mouse genomes." *Nature* 2020, 583(7818):699–710. PMID: 32728249.

- [12] Ouyang N, **Boyle AP**. "TRACE: transcription factor footprinting using chromatin accessibility data and DNA sequence." *Genome Research* 2020, 30:1040–1046. PMID: 32660981.
- [13] <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.
- [14] 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.
- [15] 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.
- [16] <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.
- [17] Amemiya HM, Kundaje A, **Boyle AP**. "The ENCODE Blacklist: Identification of Problematic Regions of the Genome." *Scientific Reports* 2019, 9:9354. PMID: 31249361.
- [18] Dong S, **Boyle AP**. "Predicting functional variants in enhancer and promoter elements using RegulomeDB." *Human Mutation* 2019, 33(8):831. PMID: 31228310.
- [19] 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.
- [20] 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.
- [21] <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.
- [22] 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.
- [23] 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.
- [24] \*Yang B, \*Zhou W, \*Jiao J, Nielsen JB, Mathis MR, Heydarpour M, Lettre G, Folkersen L, Prakash S, Schurmann C, Fritsche L, <u>Farnum GA</u>, Lin M, Othman M, Hornsby W, Driscoll A, Levasseur A, Thomas M, Farhat L, Dubé MP, Isselbacher EM, Franco-Cereceda A, Guo Dc, Bottinger EP, Deeb GM, Booher A, Kheterpal S, Chen YE, Kang HM, Kitzman J, Cordell HJ, Keavney BD, Goodship JA, Ganesh SK, Abecasis G, Eagle KA, **Boyle AP**, Loos RJF, †Eriksson P, †Tardif JC, †Brummett CM, †Milewicz DM, †Body SC, †Willer CJ. "Protein-altering and regulatory genetic variants near GATA4 implicated in bicuspid aortic valve." *Nature Communications* 2017, 8:15481. PMID: 28541271.
- [25] Nishizaki SS, Boyle AP. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.

- [26] Diehl AG, Boyle AP. "Deciphering ENCODE." Trends in Genetics 2016, 32(4):238-249. PMID: 26962025.
- [27] Phanstiel DH, Boyle AP, Heidari N, Snyder MP. "Mango: A bias correcting ChIA-PET analysis pipeline." Bioinformatics 2015. PMID: 26034063.
- [28] \*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.
- [29] \*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.
- [30] \*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.
- [31] 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.
- [32] 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.
- [33] \*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.
- [34] \*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.
- [35] 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.
- [36] 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.
- [37] The ENCODE Project Consortium. "An integrated encyclopedia of DNA elements in the human genome." *Nature* 2012, 489(7414):57–74. PMID: 22955616.
- [38] \*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.

- [39] \*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.
- [40] \*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.
- [41] The ENCODE Project Consortium. "A user's guide to the encyclopedia of DNA elements (ENCODE)." PLoS Biology 2011, 9(4):e1001046. PMID: 21526222.
- [42] **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.
- [43] \*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.
- [44] 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.
- [45] Georgiev S, Boyle AP, Jayasurya K, Mukherjee S, Ohler U. "Evidence-ranked motif identification." Genome Biology 2010, 11(2):R19. PMID: 20156354.
- [46] 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.
- [47] 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.
- [48] Boyle AP, Furey TS. "High-resolution mapping studies of chromatin and gene regulatory elements." *Epigenomics* 2009, 1(2):319–329. PMID: 20514362.
- [49] **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.
- [50] 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.
- [51] Boyle AP, Boyle JA. "Global analysis of microbial translation initiation regions." In *Journal of the Mississippi Academy of Sciences*, Volume 48 2003:138–150.
- [52] Boyle AP, Boyle JA. "Visualization of aligned genomic open reading frame data." Biochemistry and Molecular Biology Education 2003, 31:64–68.
- [53] 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**

[54] 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).

[55] 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 (continuation of u.s. patent application no. 13/592,292)." 2018, (Pending United States patent Appl. 15/954,354).