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.

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.

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

(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

Professional Service

Service

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

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

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 NIH/NIMH Study Section ZMH1 ERB-C (08) - Fine-Mapping Genome-Wide Associated Loci to 2020 Identify Proximate Causal Mechanisms of Serious Mental Illness (Ad Hoc) NIH/NIMH Study Section ZMH1 ERB-C (01) - PsychENCODE: Non-Coding Functional Elements 2019 in the Human Brain and Their Role in the Development of Psychiatric Disorders (Ad Hoc) 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 2019-current Biotechnology Program Committee, Studies of Phenotypes and Clinical Applications, ISMB/ECCB 2019 Reviewer for Internal Michigan Searle applications 2018-2019 Program Committee, Great Lakes Bioinformatics and Canadian Computational Biology Confer-2015-2018 ence (GLBIO/CCBC) Program Committee, Algorithms for Computational Biology (ALCOB) 2015-2016 Reviewer for UK Medical Research Council (RCUK MRC) 2015 Reviewer for UK Biotechnology and Biological Sciences Research Council (RCUK BBSRC) 2015

2015	Reviewer for Michigan Institute for Clinical & Health Research (MICHR) Postdoctoral Translational
	Scholars Program
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

Teaching and Mentorship

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reaching	(F = Fall Term.	, W = Winter Term	, S = Summer Term)

W19, W20, W21	Bioinformatics Concepts and Algorithms (BIOINF 529) [Course Director]
F15, F16, F17, F18, F19, F20	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 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

2019-currrent	Monica Holmes (M.S. Student, Bioinformatics, University of Michigan)
2020-current	Camille Mumm (Ph.D. Student, Human Genetics, University of Michigan)
2018-current	Bradley Crone (Ph.D. Student, Bioinformatics, University of Michigan)
2017–current	Melissa Englund (Ph.D. Student, Human Genetics, University of Michigan) NIH Human Genetics Training Program (T32) Rackham Graduate Student Research Grant (candidate)
2017–current	Samuel Zhao (Ph.D. Student, Bioinformatics, University of Michigan) Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate)
2016–2018	Haley Amemiya (Ph.D. Student, Cellular and Molecular Biology, 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
2016–2020	Shriya Sethuraman (Ph.D. Student, Bioinformatics, University of Michigan)
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) 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-current 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

2021	Kinsey Van Deynze (Rotation Student, Bioinformatics, University of Michigan)
2020	Andrea Valenzuela (Rotation Student, Chemical Biology, University of Michigan)
2020	Breanna McBean (Rotation Student, Human Genetics, 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	Tingvang Li (Rotation Student, Bioinformatics, 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-current	Aryn Booker (Undergraduate, UROP Molecular, Cellular, and Developmental Biology, University
	of Michigan)
2021-current	Preston Parana (Undergraduate, UROP Molecular, Cellular, and Developmental Biology, Univer-
	sity of Michigan)
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

Ashley Melnick (Cellular and Molecular Biology, University of Michigan) 2020-current Benjamin Yang (Biomedical Engineering, University of Michigan) 2019-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-current Negar Farzaneh (Bioinformatics, University of Michigan) 2018-current Rucheng Diao (Bioinformatics, University of Michigan) 2018-current Steven Romanelli (Molecular & Integrative Physiology, University of Michigan) 2017-current Amanda Moccia (Human Genetics, University of Michigan) 2017-current Christopher Lee (Biostatistics, University of Michigan) 2017-2020 Mohd Hafiz Bin Mohd Rothi (Molecular, Cellular, and Developmental Biology, University of Michi-2016-current gan) Ari Allyn-Feuer (Bioinformatics, University of Michigan) 2015-2018 Raymond Cavalcante (Bioinformatics, University of Michigan) 2015-2017 Zhengting Zou (Bioinformatics, University of Michigan) 2015-2017

Preliminary Exam Committees

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

Publications

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

- [1] McDonald TL, Zhou W, Castro CP, Mumm C, Switzenberg JA, Mills RE, Boyle AP. "Cas9 targeted enrichment of mobile elements using nanopore sequencing." 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] *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." *bioRxiv* 2020.
- [6] Zhao N, Boyle AP. "F-Seq2: improving the feature density based peak caller with dynamic statistics." NAR Genomics and Bioinformatics, In Press 2021.
- [7] *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.
- [8] <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.
- [9] The ENCODE Project Consortium. "Perspectives on ENCODE." Nature 2020, 583(7818):693–698. PMID: 32728248.
- [10] The ENCODE Project Consortium. "Expanded encyclopaedias of DNA elements in the human and mouse genomes." *Nature* 2020, 583(7818):699–710. PMID: 32728249.
- [11] Ouyang N, Boyle AP. "TRACE: transcription factor footprinting using chromatin accessibility data and DNA sequence." *Genome Research* 2020, 30:1040–1046. PMID: 32660981.
- [12] <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.
- [13] 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.
- [14] 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.
- [15] <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.
- [16] Amemiya HM, Kundaje A, **Boyle AP**. "The ENCODE Blacklist: Identification of Problematic Regions of the Genome." *Scientific Reports* 2019, 9:9354. PMID: 31249361.
- [17] Dong S, **Boyle AP**. "Predicting functional variants in enhancer and promoter elements using RegulomeDB." *Human Mutation* 2019, 33(8):831. PMID: 31228310.
- [18] 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.
- [19] 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.
- [20] <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.

- [21] 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.
- [22] 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.
- [23] *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.
- [24] Nishizaki SS, Boyle AP. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.
- [25] Diehl AG, Boyle AP. "Deciphering ENCODE." Trends in Genetics 2016, 32(4):238–249. PMID: 26962025.
- [26] Phanstiel DH, Boyle AP, Heidari N, Snyder MP. "Mango: A bias correcting ChIA-PET analysis pipeline." Bioinformatics 2015. PMID: 26034063.
- [27] *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.
- [28] *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.
- [29] *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.
- [30] 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.

- [31] 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.
- [32] *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.
- [33] *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.
- [34] 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.
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