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 F

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.

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

This project seeks to provide new insights into atrial fibrillation mechanisms through whole-

genome 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.

NVIDIA GPU Grant (PI: Boyle)

NVIDIA Corporation

Professional Service

Service

2020–current DHG M.S. Admissions Committee DHG Ph.D. Admissions Committee

2018-current DCM&B Diversity, Equity, & Inclusion Ally [Chair 2018–2020]

2018-current Lab Safety Liaison for DCM&B

2018–2019 Cellular and Molecular Biology Admissions Committee

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

2017–2020 DHG Faculty Recruitment and Promotions Committee

2017–2019 EBS Faculty IT Committee

2016–2019 DCM&B Faculty Recruitment Committee
2016–2020 DCM&B Seminar Series Committee [Chair]

2015–2018 DCM&B Admissions Committee

2015–2017 DHG Computational Support Committee

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

2012–2014 Member, American Association for the Advancement of Science (AAAS)

2005-current | Member, Gamma Sigma Delta Agricultural Honor Society

Reviewing Activity

Since 2009

Ad hoc reviewer (100 verified reviews) for the journals: 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

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)
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
2018–2019	Reviewer for Internal Michigan Searle applications
2015–2018	Program Committee, Great Lakes Bioinformatics and Canadian Computational Biology Confer-
	ence (GLBIO/CCBC)
2015–2016	Program Committee, Algorithms for Computational Biology (ALCOB)
2015	Reviewer for UK Medical Research Council (RCUK MRC)
2015	Reviewer for UK Biotechnology and Biological Sciences Research Council (RCUK BBSRC)
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

Teaching (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

2019–2020	Monica Holmes (M.S. Student, Bioinformatics, University of Michigan)
2020-current	Camille Mumm (Ph.D. Student, Human Genetics, University of Michigan) Rackham Graduate Student Research Grant (pre-candidate)
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

2015-current Ningxin Ouyang (Ph.D. Student, Bioinformatics, University of Michigan)

Rackham Graduate Student Research Grant (candidate)

2016-current | Shengcheng Dong (Ph.D. Student, Bioinformatics, University of Michigan)

Rackham Graduate Student Research Grant (candidate)

2015-current Torrin McDonald (Ph.D. Student, Human Genetics, University of Michigan)

NIH Human Genetics Training Program (T32)

Rackham Graduate Student Research Grant (pre-candidate) Rackham Graduate Student Research Grant (candidate)

2015–2017 Greg Farnum (Ph.D. Student, Cellular and Molecular Biology, University of Michigan)

2015–2020 | Sierra Nishizaki (Ph.D. Student, Human Genetics, 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

Kinsey Van Deynze (Rotation Student, Bioinformatics, University of Michigan)
Andrea Valenzuela (Rotation Student, Chemical Biology, University of Michigan)
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)

Amanda Moccia (Rotation Student, Human Genetics, University of Michigan)
Stephen Carney (Rotation Student, Human Genetics, University of Michigan)

2016 Tingyang 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	Aryn Booker (Undergraduate, UROP Molecular, Cellular, and Developmental Biology, University
	of Michigan)
2021	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

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-current	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)
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)

Adrienne Shami (Human Genetics, University of Michigan)
Trenton Frisbie (Human Genetics, University of Michigan)
Melissa Englund (Human Genetics, University of Michigan)
Peter Orchard (Bioinformatics, University of Michigan)
Li Guan (Bioinformatics, University of Michigan)
Shriya Sethuraman (Bioinformatics, University of Michigan)
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] Dong S, **Boyle AP**. "Prioritization of regulatory variants with tissue-specific function in the non-coding regions of human genome." *bioRxiv* 2021.
- [2] 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.
- [3] Rothi MH, <u>Sethuraman S</u>, Dolata J, **Boyle AP**, Wierzbicki AT. "DNA methylation directs nucleosome positioning in RNA-mediated transcriptional silencing." *bioRxiv* 2020.
- [4] 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.
- [5] Nishizaki SS, Boyle AP. "SEMplMe: A tool for integrating DNA methylation effects in transcription factor binding affinity predictions." *bioRxiv* 2020.
- [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." *bioRxiv* 2020.
- [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, <u>Dong S</u>, Hawkins-Hooker A, Inoue F, Juven-Gershon T, Kenlay H, Martin B, Patra A, Penzar DP, Schubach M, Xiong C, Yan Z, **Boyle AP**, Kreimer A, Kulakovskiy IV, Reid J, Unger R, Yosef N, Shendure J, Ahituv N, Kircher M, Beer MA. "Integration of Multiple Epigenomic Marks Improves Prediction of Variant Impact in Saturation Mutagenesis Reporter Assay." *Human mutation* 2019, 33(8):831. PMID: 31106481.
- [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, Diebali 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.
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