2017 Palmer Commons
100 Washtenaw Ave.
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www.BoyleLab.org

Alan P. Boyle

Education

2005–2009 **Doctor of Philosophy,** Computational Biology and Bioinformatics
Duke University, Durham, NC

2001–2005 **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
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.

2021–2026 U01 HG011952 (PI: Boyle)
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.

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 traits
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.

2019 | NVIDIA GPU Grant (PI: Boyle)
NVIDIA Corporation

Professional Service

Service

2018–current | DCM&B Diversity, Equity, & Inclusion Committee [Ally/Chair 2018–2020]
2018–current | Lab Safety Liaison for DCM&B
2017–current | DCM&B Preliminary Exam Abstract Review Committee (PARC) [Chair 2018–current]
2020–2021 | DHG M.S. Admissions Committee
2019–2020 | DHG Ph.D. Admissions Committee
2017–2020 | DHG Faculty Recruitment and Promotions Committee
2016–2020 | DCM&B Seminar Series Committee [Chair]
2018–2019 | Cellular and Molecular Biology Admissions Committee
2017–2019 | EBS Faculty IT Committee
2016–2019 | DCM&B Faculty Recruitment Committee
2015–2018 | DCM&B Admissions Committee
2015–2017 | DHG Computational Support Committee
2015–2016 | 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

Manuscript 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*
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 Conference (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

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)

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)
Joint M.S. in Bioinformatics, University of Michigan
NIH Genome Science Training Program (T32)

2019–2020 | Monica Holmes (M.S. Student, Bioinformatics, University of Michigan)

2020–current | Camille Mumm (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)

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)

2018–current | Samuel Zhao (Ph.D. Student, Bioinformatics, University of Michigan)
2017–2018 | Samuel Zhao (M.S. 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)
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

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

2017–current | Ningxin Ouyang (Ph.D. Student, Bioinformatics, University of Michigan)
2015–2017 | Ningxin Ouyang (M.S. Student, Bioinformatics, University of Michigan)
Rackham Graduate Student Research Grant (candidate)

2016–2021 | 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)
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

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

2021–current 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

2021–current Julia Tweadey (Undergraduate, LSA Honors Program, Life Science Informatics, University of Michigan)
2021–current Preston Parana (Undergraduate, UROP Molecular, Cellular, and Developmental Biology, University of Michigan)
UROP Blue Ribbon Award
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 Michigan)
2016–2017 Colten Williams (Undergraduate, UROP Computer Science, University of Michigan)
2016–2017 Courtney Asman (Undergraduate, Neuroscience, University of Michigan)
2014–2017 Maxwell Spadafore (Undergraduate, LS&A Honors Informatics, University of Michigan)
2013–2014 Natalie Ng (High School, Stanford Institutes of Medicine Summer Research)
2013–2014 Dana Wyman (Undergraduate, Biology, Stanford University)
2013 Justin Young (High School, Stanford Institutes of Medicine Summer Research)
2012 Melanie Connick (Undergraduate, Biology, University of New Mexico)
2012 Edward Dai (Undergraduate, Computer Science, Stanford University)

Doctoral Thesis Committees

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–current	Rucheng Diao (Bioinformatics, University of Michigan)
2017–current	Amanda Moccia (Human Genetics, University of Michigan)
2016–2021	Mohd Hafiz Bin Mohd Rothi (Molecular, Cellular, and Developmental Biology, University of Michigan)
2017–2021	Steven Romanelli (Molecular & Integrative Physiology, University of Michigan)
2018–2021	Negar Farzaneh (Bioinformatics, University of Michigan)
2017–2020	Christopher Lee (Biostatistics, University of Michigan)
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	Anthony Nguyen (Human Genetics, University of Michigan)
2021	Hanbyul Cho (Bioinformatics, University of Michigan)
2021	Charles Ryan (Cellular and Molecular Biology, University of Michigan)
2021	Kuan-Han Wu (Bioinformatics, University of Michigan)
2021	Wenjin Gu (Bioinformatics, University of Michigan)
2020	Jie Cao (Bioinformatics, University of Michigan)
2020	Zijun Gao (Bioinformatics, University of Michigan)
2020	Ashley Melnick (Cellular and Molecular Biology, University of Michigan)
2019	Benjamin Yang (Biomedical Engineering, University of Michigan)
2019	Maria Virgilio (Cellular and Molecular Biology, University of Michigan)
2018	Zhi Carrie Li (Bioinformatics, University of Michigan)
2018	Kevin Hu (Bioinformatics, University of Michigan)
2018	Siyu Liu (Bioinformatics, University of Michigan)
2018	Alexandra Weber (Bioinformatics, University of Michigan)
2018	Mitch Fernandez (Bioinformatics, University of Michigan)
2017	Tingyang Li (Bioinformatics, University of Michigan)
2017	Marcus Sherman (Bioinformatics, University of Michigan)
2017	Adrienne Shami (Human Genetics, University of Michigan)
2017	Trenton Frisbie (Human Genetics, University of Michigan)
2017	Melissa Englund (Human Genetics, University of Michigan)
2017	Peter Orchard (Bioinformatics, University of Michigan)
2017	Li Guan (Bioinformatics, University of Michigan)
2016	Shriya Sethuraman (Bioinformatics, University of Michigan)
2016	Jed Carlson (Bioinformatics, University of Michigan)

Industry Experience

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

Publications

* Indicates co-first authorship † Indicates co-senior authorship

underscore indicates lab members

[1] 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.

[2] Bao Y, Wadden J, Erb-Downward JR, Ranjan P, Zhou W, McDonald TL, Mills RE, **Boyle AP**, Dickson RP, Blaauw D, Welch JD. "Real-time, direct classification of nanopore signals with squigglenet." ***Genome Biology, Accepted*** 2021.

[3] Dong S, **Boyle AP**. "Prioritization of regulatory variants with tissue-specific function in the non-coding regions of human genome." ***Nucleic Acids Research, Accepted*** 2021.

[4] Rothi MH, Sethuraman S, Dolata J, **Boyle AP**, Wierzbicki AT. "DNA methylation directs nucleosome positioning in RNA-mediated transcriptional silencing." ***bioRxiv*** 2020.

[5] 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.

[6] Nishizaki SS, **Boyle AP**. "SEMplMe: A tool for integrating DNA methylation effects in transcription factor binding affinity predictions." ***bioRxiv*** 2020.

[7] *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.

[8] *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.

[9] Zhao N, **Boyle AP**. "F-Seq2: improving the feature density based peak caller with dynamic statistics." ***NAR Genomics and Bioinformatics*** 2021, 3. PMID: 33655209.

[10] *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.

[11] Diehl AG, **Boyle AP**. "MapGL: Inferring evolutionary gain and loss of short genomic sequence features by phylogenetic maximum parsimony." ***BMC Bioinformatics*** 2020, 21:416. PMID: 32962625.

[12] The ENCODE Project Consortium. "Perspectives on ENCODE." ***Nature*** 2020, 583(7818):693–698. PMID: 32728248.

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

[14] Ouyang N, **Boyle AP**. "TRACE: transcription factor footprinting using chromatin accessibility data and DNA sequence." ***Genome Research*** 2020, 30:1040–1046. PMID: 32660981.

[15] Diehl AG, Ouyang N, **Boyle AP**. "Transposable elements contribute to cell and species-specific chromatin looping and gene regulation in mammalian genomes." ***Nature Communications*** 2020, 11:1796. PMID: 32286261.

[16] 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.

[17] 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.

[18] Diehl AG, **Boyle AP**. "CGIMP: Real-time exploration and covariate projection for self-organizing map datasets." ***Journal of Open Source Software*** 2019, 4(39):1520.

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

[20] Dong S, **Boyle AP**. "Predicting functional variants in enhancer and promoter elements using RegulomeDB." ***Human Mutation*** 2019, 33(8):831. PMID: 31228310.

[21] 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.

[22] 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.

[23] Diehl AG, **Boyle AP**. "Conserved and species-specific transcription factor co-binding patterns drive divergent gene regulation in human and mouse." ***Nucleic Acids Research*** 2018, 46(4):1878–1894. PMID: 29361190.

[24] 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.

[25] 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.

[26] *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.

[27] Nishizaki SS, **Boyle AP**. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." ***Trends in Genetics*** 2017, 33:34–45. PMID: 27939749.

[28] Diehl AG, **Boyle AP**. "Deciphering ENCODE." ***Trends in Genetics*** 2016, 32(4):238–249. PMID: 26962025.

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