

# Alan P. Boyle

2017 Palmer Commons  
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[www.BoyleLab.org](http://www.BoyleLab.org)

## Education

2005–2009	<b>Doctor of Philosophy</b> , Computational Biology and Bioinformatics Duke University, Durham, NC
2001–2005	<b>Bachelor of Science</b> , <i>summa cum laude</i> , Biochemistry and Molecular Biology <b>Bachelor of Science</b> , <i>summa cum laude</i> , Computer Science Mississippi State University, Starkville, MS

## Academic Appointments

2017–present	<b>Member</b> , Cellular and Molecular Biology Program
2016–present	<b>Member</b> , Center for RNA Biomedicine
2015–present	<b>Member</b> , Genome Science Training Program (GSTP) <b>Member</b> , Michigan Predoctoral Training Program in Genetics (GTP)
2014–present	<b>Member</b> , Program in Biomedical Sciences <b>Member</b> , Bioinformatics Training Program
2015–present	<b>Assistant Professor</b> , Department of Human Genetics
2014–present	<b>Assistant Professor</b> , Department of Computational Medicine & Bioinformatics University of Michigan, Ann Arbor, MI
2010–2014	<b>Postdoctoral Scholar</b> , Genetics Stanford University, Stanford, CA; Advisor: Dr. Michael Snyder
Spring 2010	<b>Postdoctoral Associate</b> , 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

2017–2020	U41 HG009293 NIH/NHGRI RegulomeDB: A Resource for the Human Regulome	(Multi PI: Boyle, Cherry) Total Costs: \$2,171,753
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	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 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.	(PI: Boyle) Total Costs: \$979,984
2018–2023	R01 HD093570 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.	(PI: Bielas; Co-I with Effort) Total Costs: \$2,304,265
2019–2021	Precision Health Investigators Award 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.	(co-PI: Todd, Boyle, Mills) Total Costs: \$300,000
2020–2023	W81XWH2010336 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.	(PI: Aguilar; Co-I with Effort) Total Costs: \$1,081,608

## Completed

2013–2017	R00 HG007356 Pathway to Independence Award (K99/R00) 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.	(PI: Boyle) Total Costs: \$987,771
2015–2017	FG-2015-65465 Alfred P. Sloan Foundation Fellowship in Computational & Evolutionary Molecular Biology	(PI: Boyle) Total Costs: \$50,000
2016–2020	R01 HL130705 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.	(PI: Willer; Co-I with Effort) Total Costs: \$2,784,005
2017–2018	Eleanor and Larry Jackier U-M/Technion and Weizmann Collaborative Research Grant Michigan - Israel Partnership for Research & Education Identifying novel disease related mutations in the genomic environments around Transcription 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.	(PI: Boyle, Mandel-Gutfreund) Total Costs: \$50,000
2017–2024	R35 HL135824 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.	(PI: Willer; Co-I with Effort) Total Costs: \$4,650,000
2019	NVIDIA GPU Grant NVIDIA Corporation	(PI: Boyle) In-Kind Value: \$3,600

## Professional Service

### Service

2019–current	DHG Admissions Committee
2018–current	DCM&B Diversity, Equity, & Inclusion Ally [Chair]
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]
2017–2019	EBS Faculty IT Committee
2017–current	DHG Faculty Recruitment and Promotions Committee
2016–current	DCM&B Faculty Recruitment Committee
2016–current	DCM&B Seminar Series Committee [Chair 2016–current]
2015–2018	DCM&B Admissions Committee
2015–2017	DHG Computational Support Committee
2015–2016	DCM&B Retreat Planning Committee Chair (including 1st annual)
2014	<i>Ad hoc</i> 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	<i>Ad hoc</i> reviewer ( <a href="#">96 verified reviews</a> ) for the journals: <i>Nature Genetics</i> , <i>Genome Research</i> , <i>Genome Biology</i> , <i>Nature Neuroscience</i> , <i>Nature Communications</i> , <i>Nature Protocols</i> , <i>Bioinformatics</i> , <i>Nucleic Acids Research</i> , <i>BMC Biology</i> , <i>BMC Bioinformatics</i> , <i>Oncotarget</i> , <i>Scientific Reports</i> , <i>Atherosclerosis</i> , <i>BioEssays</i> , <i>Gene</i>
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–current	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 Conference (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 <i>Bioinformatics</i>

## Teaching and Mentorship

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

W19, W20	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–current	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

2018–current	Bradley Crone (Ph.D. Student, Bioinformatics, University of Michigan)
2017–current	Melissa Englund (Ph.D. Student, Human Genetics, University of Michigan) <i>NIH Human Genetics Training Program (T32)</i> <i>Rackham Graduate Student Research Grant (candidate)</i>
2017–current	Samuel Zhao (Ph.D. Student, Bioinformatics, University of Michigan) <i>Rackham Graduate Student Research Grant (pre-candidate)</i> <i>Rackham Graduate Student Research Grant (candidate)</i>
2016–2018	Haley Amemiya (Ph.D. Student, Cellular and Molecular Biology, University of Michigan) <i>NIH Cellular &amp; Molecular Biology Training Program (T32)</i> <i>NIH Cellular Biotechnology Training Program (T32) (Declined)</i> <i>PIBS Excellence in Service Award</i> <i>Rackham Graduate Student Research Grant (pre-candidate)</i> <i>Rackham Graduate Student Research Grant (candidate)</i> <i>Maas Professional Development Award</i> <i>Rackham Graduate School Scholar-Activist Award</i>
2016–current	Shriya Sethuraman (Ph.D. Student, Bioinformatics, University of Michigan)
2016–current	Shengcheng Dong (Ph.D. Student, Bioinformatics, University of Michigan) <i>Rackham Graduate Student Research Grant (candidate)</i>
2016–current	Christopher Castro (Ph.D. Student, Bioinformatics, University of Michigan) <i>NIH Bioinformatics Training Program (T32)</i> <i>Rackham Merit Fellow</i> <i>Rackham Graduate Student Research Grant (pre-candidate)</i> <i>Global Research Engagement Opportunity Fellowship</i>
2015–current	Ningxin Ouyang (Ph.D. Student, Bioinformatics, University of Michigan)
2015–current	Torin McDonald (Ph.D. Student, Human Genetics, University of Michigan) <i>NIH Human Genetics Training Program (T32)</i> <i>Rackham Graduate Student Research Grant (pre-candidate)</i> <i>Rackham Graduate Student Research Grant (candidate)</i>
2015–2017	Greg Farnum (Ph.D. Student, Cellular and Molecular Biology, University of Michigan)
2015–current	Sierra Nishizaki (Ph.D. Student, Human Genetics, University of Michigan) <i>NIH Genome Science Training Program (T32)</i>

*Rackham Merit Fellow*  
*Rackham Summer Award*  
*Rackham Graduate Student Research Grant (candidate)*

### Additional Graduate Rotation Students

2020	Camille Mumm (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)
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)

### Non-student Lab Volunteers

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

### Undergraduate and High School Students

2020–current	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)
2019–2019	Sheila Rasouli (Undergraduate, Neuroscience, University of Toronto)
2019–2019	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

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	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 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

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
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## Publications

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

- [1] Abascal F, Acosta R, Addleman NJ, Adrian J, Afzal V, Aken B, Akiyama JA, Jammal OA, Amrhein H, Anderson SM, Andrews GR, Antoshechkin I, Ardlie KG, Armstrong J, Astley M, Banerjee B, Barkal AA, Barnes IHA, Barozzi I, Barrell D, Barson G, Bates D, Baymuradov UK, Bazile C, Beer MA, Beik S, Bender MA, Bennett R, Bouvrette LPB, Bernstein BE, Berry A, Bhaskar A, Bignell A, Blue SM, Bodine DM, Boix C, Boley N, Borrmann T, Borsari B, **Boyle AP**, Brandsmeier LA, Breschi A, Bresnick EH, Brooks JA, Buckley M, Burge CB, Byron R, Cahill E, Cai L, Cao L, Carty M, Castanon RG, Castillo A, Chaib H, Chan ET, Chee DR, Chee S, Chen H, Chen H, Chen JY, Chen S, Cherry JM, Chhetri SB, Choudhary JS, Chrast J, Chung D, Clarke D, Cody NAL, Coppola CJ, Coursen J, D'Ippolito AM, Dalton S, Danyko C, Davidson C, Davila-Velderrain J, Davis CA, Dekker J, Deran A, DeSalvo G, Despacio-Reyes G, Dewey CN, Dickel DE, Diegel M, Diekhans M, Dileep V, Ding B, Djebali S, Dobin A, Dominguez D, Donaldson S, Drenkow J, Dreszer TR, Drier Y, Duff MO, Dunn D, Eastman C, Ecker JR, Edwards MD, El-Ali N, Elhajjajy SI, Elkins K, Emili A, Epstein CB, Evans RC, Ezkurdia I, Fan K, Farnham PJ, Farrell NP, Feingold EA, Ferreira AM, Fisher-Aylor K, Fitzgerald S, Flicek P, Foo CS, Fortier K, Frankish A, Freese P, Fu S, Fu XD, Fu Y, Fukuda-Yuzawa Y, Fulciniti M, Funnell APW, Gabdank I, Galeev T, Gao M, Giron CG, Garvin TH, Gelboin-Burkhart CA, Georgolopoulos G, Gerstein MB, Giardine BM, Gifford DK, Gilbert DM, Gilchrist DA, Gillespie S, Gingeras TR, Gong P, Gonzalez A, Gonzalez JM, Good P, Goren A, Gorkin DU, Graveley BR, Gray M, Greenblatt JF, Griffiths E, Groudine MT, Grubert F, Gu M, Guigó R, Guo H, Guo Y, Guo Y, Gursoy G, Gutierrez-Arcelus M, Halow J, Hardison RC, Hardy M, Hariharan M, Harmanci A, Harrington A, Harrow JL, Hashimoto TB, Hasz RD, Hatan M, Haugen E, Hayes JE, He P, He Y, Heidari N, Hendrickson D, Heuston EF, Hilton JA, Hitz BC, Hochman A, Holgren C, Hou L, Hou S, Hsiao YHE, Hsu S, Huang H, Hubbard TJ, Huey J, Hughes TR, Hunt T, Ibarrientos S, Issner R, Iwata M, Izuogu O, Jaakkola T, Jameel N, Jansen C, Jiang L, Jiang P, Johnson A, Johnson R, Jungreis I, Kadaba M, Kasowski M, Kasparian M, Kato M, Kaul R, Kawli T, Kay M, Keen JC, Keles S, Keller CA, Kelley D, Kellis M, Kheradpour P, Kim DS, Kirilusha A, Klein RJ, Knoechel B, Kuan S, Kulik MJ, Kumar S, Kundaje A, Kutayavin T, Lagarde J, Lajoie BR, Lambert NJ, Lazar J, Lee AY, Lee D, Lee E, Lee JW, Lee K, Leslie CS, Levy S, Li B, Li H, Li N, Li X, Li YI, Li Y, Li Y, Li Y, Lian J, Libbrecht MW, Lin S, Lin Y, Liu D, Liu J, Liu P, Liu T, Liu XS, Liu Y, Liu Y, Long M, Lou S, Loveland J, Lu A, Lu Y, Lécuyer E, Ma L, Mackiewicz M, Mannion BJ, Mannstadt M, Manthavadi D, Marinov GK, Martin FJ, Mattei E, McCue K, McEown M, McVicker G, Meadows SK, Meissner A, Mendenhall EM, Messer CL, Meuleman W, Meyer C, Miller S, Milton MG, Mishra T, Moore DE, Moore HM, Moore JE, Moore SH, Moran J, Mortazavi A, Mudge JM, Munshi N, Murad R, Myers RM, Nandakumar V, Nandi P, Narasimha AM, Narayanan AK, Naughton H, Navarro

- FCP, Navas P, Nazarovs J, Nelson J, Neph S, Neri FJ, Nery JR, Nesmith AR, Consortium TEP. "Expanded encyclopaedias of dna elements in the human and mouse genomes." *Nature* 2020, 583(7818):699–710.
- [2] Ouyang N, **Boyle AP**. "TRACE: transcription factor footprinting using DNase I hypersensitivity data and DNA sequence." *Genome Research* 2020.
- [3] 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.
- [4] 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.
- [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] Diehl AG, **Boyle AP**. "MapGL: Inferring evolutionary gain and loss of short genomic sequence features by phylogenetic maximum parsimony." *bioRxiv* 2019.
- [7] Nishizaki SS, Ng N, Dong S, Porter RS, Mortrud C, Williams C, Asman C, Switzenberg JA, **Boyle AP**. "Predicting the effects of SNPs on transcription factor binding affinity." *Bioinformatics* 2019, 35:2434. PMID: 31373606.
- [8] 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.
- [9] Amemiya HM, Kundaje A, **Boyle AP**. "The ENCODE Blacklist: Identification of Problematic Regions of the Genome." *Scientific Reports* 2019, 9:9354. PMID: 31249361.
- [10] Dong S, **Boyle AP**. "Predicting functional variants in enhancer and promoter elements using RegulomeDB." *Human Mutation* 2019, 33(8):831. PMID: 31228310.
- [11] 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.
- [12] 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.
- [13] 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.
- [14] Nielsen JB, Fritsche LG, Zhou W, Teslovich TM, Holmen OL, Gustafsson S, Gabrielsen ME, Schmidt EM, Beaumont R, Wolfford 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, Wilsaard T, Loos RJJ, 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.
- [15] 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.
- [16] \*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.
- [17] Nishizaki SS, **Boyle AP**. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.
- [18] Diehl AG, **Boyle AP**. "Deciphering ENCODE." *Trends in Genetics* 2016, 32(4):238–249. PMID: 26962025.
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## Patents

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