(Multi PI: Boyle, Cherry)

Total Costs: \$2,171,753

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

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

Scholarships, Fellowships, and Honors

Endowment for the Basic Sciences Teaching Award 2019 First Place in CAGI5 Regulation Saturation Challenge 2018 **NSF CAREER Award** 2017 Institutional nominee for W.M. Keck Foundation Medical Science Research Program 2016 Institutional nominee for Searle Scholar Award 2016 Alfred P. Sloan Foundation Fellowship in Computational & Evolutionary Molecular Biology 2015-2017 NIH Pathway to Independence Award (K99/R00) [1K99HG007356-01] 2013-2014 AAAS/Science Program for Excellence in Science 2012 NSF Graduate Research Fellowship 2005-2008 James B. Duke Fellowship 2005-2009 Mayo Clinic Summer Undergraduate Research Fellow Summer 2004 Barry M. Goldwater Memorial Scholarship 2003 The Institute for Genomic Research (TIGR) Summer Fellow Summer 2003 Robert C. Byrd Honors Scholarship 2001 Mississippi State University Presidential Scholarship 2001 National Merit Scholarship 2001

Grant Support

Active

2017–2020 U41 HG009293

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.

DBI-1651614 2017-2022

(PI: Boyle)

NSF/BIO/DBI

Total Costs: \$979,984

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.

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

NIH/NICHD

Total Costs: \$2,304,265

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.

NVIDIA GPU Grant

NIH/NHGRI

(PI: Boyle)

NVIDIA Corporation In-Kind Value: \$3,600

Completed

R00 HG007356 Pathway to Independence Award (K99/R00) 2013-2017

(PI: Boyle)

Total Costs: \$987,771

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.

FG-2015-65465 2015-2017

(PI: Boyle)

Alfred P. Sloan Foundation

Total Costs: \$50,000

Fellowship in Computational & Evolutionary Molecular Biology

2016-2020 R01 HL130705 (PI: Willer; Co-I with Effort)

NIH/NHLBI

Total Costs: \$2,784,005

Large-scale human genetics to understand molecular mechanisms of atrial fibrillation and related

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

2017-2018

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

(PI: Boyle, Mandel-Gutfreund)

Michigan - Israel Partnership for Research & Education

Total Costs: \$50,000

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

Total Costs: \$4,650,000

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.

Professional Service

Service

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

Lab Safety Liaison for DCM&B 2018-current

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

EBS Faculty IT Committee 2017-2019

DHG Faculty Recruitment and Promotions Committee 2017-current

DCM&B Faculty Recruitment Committee 2016-current

DCM&B Seminar Series Committee [Chair 2016–current] 2016-current

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

curity Activity		
Ad hoc reviewer (90 verified reviews) for the journals: Nature Genetics, Genome Research, Genome Biology, Nature Neuroscience, Nature Communications, Nature Protocols, Bioinformat-		
ics, Nucleic Acids Research, BMC Bioinformatics, Oncotarget, Scientific Reports, Atherosclerosis,		
BioEssays, Gene		
Editorial Board, PLoS ONE		
Review Editor, Bioinformatics and Computational Biology for Frontiers in Genetics		
Review Editor, Bioinformatics and Computational Biology for Frontiers in Plant Science		
Review Editor, Bioinformatics and Computational Biology for Frontiers in Bioengineering and		
Biotechnology		
Program Committee, Studies of Phenotypes and Clinical Applications, ISMB/ECCB		
Reviewer for Internal Michigan Searle applications		
Program Committee, Great Lakes Bioinformatics and Canadian Computational Biology Confer-		
ence (GLBIO/CCBC)		
Program Committee, Algorithms for Computational Biology (ALCOB)		
Reviewer for UK Medical Research Council (RCUK MRC)		
Reviewer for UK Biotechnology and Biological Sciences Research Council (RCUK BBSRC)		
Reviewer for Michigan Institute for Clinical & Health Research (MICHR) Postdoctoral Translational		
Scholars Program		
Program Committee, Gene Regulation and Transcriptomics, ISMB/ECCB		
DNA Day Essay Contest Detailed Review Judge for ASHG		
Distinguished contributor as a leading reviewer for the journal <i>Bioinformatics</i>		

Teaching and Mentorship

Teaching	(F = Fall Term	, W = Winter Term	, S = Summer Ter	m)
----------	----------------	-------------------	------------------	----

W19	Bioinformatics Concepts and Algorithms (BIOINF 529) [Course Director]
S17, S18	Introduction to Biocomputing Bootcamp (BIOSTAT/BIOINF/HUMGEN 606) [2 full days / yr.]
F17, F18	Experimental Genetics Systems (HUMGEN 632) [Course Director]
F15, F16, F17, F18	Gene Structure and Regulation (HUMGEN 541) [3 lectures + 2 discussions / yr.]
F15, W16, F16, W17, F17, W18, F18	Bioinformatics Journal Club (BIOINF 602/603) [Course Director F18]
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

2	2018–current	Lecturer, REU Site: Mathematical and Theoretical Biology Institute (MTBI), Arizona State Univer-
		sity (NSF1757968) [2 lectures]
	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

Duke student panelist for "How to prepare for and get into graduate school"
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) 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-current	Shriya Sethuraman (Ph.D. Student, Bioinformatics, University of Michigan)
2016-current	Shengcheng Dong (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)
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)
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) NIH Genome Science Training Program (T32) Rackham Merit Fellow Rackham Summer Award Rackham Graduate Student Research Grant (candidate)

Additional Graduate Rotation Students

	- Graduato Hotation Graduito		
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

Diana Davis (Undergraduate, Neuroscience and German, University of Michigan) 2019-current Sheila Rasouli (Undergraduate, Neuroscience, University of Toronto) 2019-current Vibhasri Davuluri (High School, Girls Who Code Summer Intern) 2019-current Cody Morterud (Undergraduate, UROP Computer Science / Honors Capstone, University of Michi-2016-2019 gan) 2016-2017 Colten Williams (Undergraduate, UROP Computer Science, University of Michigan) Courtney Asman (Undergraduate, Neuroscience, University of Michigan) 2016-2017 2014-2017 Maxwell Spadafore (Undergraduate, LS&A Honors Informatics, University of Michigan) Natalie Ng (High School, Stanford Institutes of Medicine Summer Research) 2013-2014 Dana Wyman (Undergraduate, Biology, Stanford University) 2013-2014 Justin Young (High School, Stanford Institutes of Medicine Summer Research) 2013 2012 Melanie Connick (Undergraduate, Biology, University of New Mexico) Edward Dai (Undergraduate, Computer Science, Stanford University) 2012

Doctoral Thesis Committees

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 2018-current Stephen Carney (Cancer Biology, University of Michigan) 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 2017-current Christopher Lee (Biostatistics, University of Michigan) 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 2015-2017 Zhengting Zou (Bioinformatics, University of Michigan)

Preliminary Exam Committees

2019

Benjamin Yang (Biomedical Engineering, University of Michigan) Maria Virgilio (Cellular and Molecular Biology, University of Michigan) 2019 Zhi Carrie Li (Bioinformatics, University of Michigan) 2018 Kevin Hu (Bioinformatics, University of Michigan) 2018 Siyu Liu (Bioinformatics, University of Michigan) 2018 Alexandra Weber (Bioinformatics, University of Michigan) 2018 Mitch Fernandez (Bioinformatics, University of Michigan) 2018 Tingyang Li (Bioinformatics, University of Michigan) 2017 Marcus Sherman (Bioinformatics, University of Michigan) 2017 2017 Adrienne Shami (Human Genetics, University of Michigan) Trenton Frisbie (Human Genetics, University of Michigan) 2017 Melissa Englund (Human Genetics, University of Michigan) 2017 Peter Orchard (Bioinformatics, University of Michigan) 2017 Li Guan (Bioinformatics, University of Michigan) 2017 Shriya Sethuraman (Bioinformatics, University of Michigan) 2016 2016 Jed Carlson (Bioinformatics, University of Michigan)

Industry Experience

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

Publications

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

- [1] <u>Diehl AG</u>, **Boyle AP**. "Mapgl: Inferring evolutionary gain and loss of short genomic sequence features by phylogenetic maximum parsimony." *bioRxiv* 2019.
- [2] Ouyang N, **Boyle AP**. "Trace: transcription factor footprinting using dnase i hypersensitivity data and dna sequence." **bioRxiv** 2019.
- [3] <u>Diehl AG</u>, <u>Ouyang N</u>, **Boyle AP**. "Transposable elements strongly contribute to cell-specific and species-specific looping diversity in mammalian genomes." *bioRxiv* 2019.
- [4] Lee CT, Cavalcante RG, Lee C, Qin T, Patil S, Wang S, Tsai ZT, Boyle AP, Sartor MA. "Poly-Enrich: Count-based Methods for Gene Set Enrichment Testing with Genomic Regions and Updates to ChIP-Enrich." bioRxiv 2018.
- [5] <u>Nishizaki SS</u>, Ng N, <u>Dong S</u>, Porter RS, <u>Morterud C</u>, <u>Williams C</u>, <u>Asman C</u>, <u>Switzenberg JA</u>, **Boyle AP**. "Predicting the effects of SNPs on transcription factor binding affinity." *Bioinformatics* 2019, 50:2434. PMID: 31373606.
- [6] <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.
- [7] Amemiya HM, Kundaje A, **Boyle AP**. "The ENCODE Blacklist: Identification of Problematic Regions of the Genome." *Scientific Reports* 2019, 9:9354. PMID: 31249361.
- [8] Dong S, Boyle AP. "Predicting functional variants in enhancer and promoter elements using RegulomeDB." *Human Mutation* 2019, 33(8):831. PMID: 31228310.
- [9] 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.
- [10] 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.
- [11] <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.
- [12] 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.
- [13] 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.
- [14] *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.

- [15] Nishizaki SS, Boyle AP. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.
- [16] Diehl AG, Boyle AP. "Deciphering ENCODE." Trends in Genetics 2016, 32(4):238–249. PMID: 26962025.
- [17] Phanstiel DH, Boyle AP, Heidari N, Snyder MP. "Mango: A bias correcting ChIA-PET analysis pipeline." Bioinformatics 2015. PMID: 26034063.
- [18] *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.
- [19] *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.
- [20] *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.
- [21] 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.
- [22] 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.
- [23] *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.
- [24] *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.
- [25] 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.
- [26] Schaub MA, Boyle AP, Kundaje A, †Batzoglou S, †Snyder M. "Linking disease associations with regulatory information in the human genome." *Genome Research* 2012, 22(9):1748–1759. PMID: 22955986.
- [27] The ENCODE Project Consortium. "An integrated encyclopedia of DNA elements in the human genome." *Nature* 2012, 489(7414):57–74. PMID: 22955616.

- [28] *Gerstein MB, *Kundaje A, *Hariharan M, *Landt SG, *Yan KK, *Cheng C, *Mu XJ, *Khurana E, *Rozowsky J, *Alexander R, *Min R, *Alves P, Abyzov A, Addleman N, Bhardwaj N, **Boyle AP**, Cayting P, Charos A, Chen DZ, Cheng Y, Clarke D, Eastman C, Euskirchen G, Frietze S, Fu Y, Gertz J, Grubert F, Harmanci A, Jain P, Kasowski M, Lacroute P, Leng J, Lian J, Monahan H, O'Geen H, Ouyang Z, Partridge EC, Patacsil D, Pauli F, Raha D, Ramirez L, Reddy TE, Reed B, Shi M, Slifer T, Wang J, Wu L, Yang X, Yip KY, Zilberman-Schapira G, Batzoglou S, Sidow A, Farnham PJ, Myers RM, Weissman SM, Snyder M. "Architecture of the human regulatory network derived from ENCODE data." *Nature* 2012, 489(7414):91–100. PMID: 22955619.
- [29] *Chen R, *Mias GI, *Li-Pook-Than J, *Jiang L, Lam HYK, Chen R, Miriami E, Karczewski KJ, Hariharan M, Dewey FE, Cheng Y, Clark MJ, Im H, Habegger L, Balasubramanian S, O'Huallachain M, Dudley JT, Hillenmeyer S, Haraksingh R, Sharon D, Euskirchen G, Lacroute P, Bettinger K, **Boyle AP**, Kasowski M, Grubert F, Seki S, Garcia M, Whirl-Carrillo M, Gallardo M, Blasco MA, Greenberg PL, Snyder P, Klein TE, Altman RB, Butte AJ, Ashley EA, Gerstein M, Nadeau KC, Tang H, Snyder M. "Personal omics profiling reveals dynamic molecular and medical phenotypes." *Cell* 2012, 148(6):1293–1307. PMID: 22424236.
- [30] *Song L, *Zhang Z, *Grasfeder LL, *Boyle AP, *Giresi PG, *Lee B, *Sheffield NC, Graff S, Huss M, Keefe D, Liu Z, London D, McDaniell RM, Shibata Y, Showers KA, Simon JM, Vales T, Wang T, Winter D, Zhang Z, Clarke ND, †Birney E, †Iyer VR, †Crawford GE, †Lieb JD, †Furey TS. "Open chromatin defined by DNasel and FAIRE identifies regulatory elements that shape cell-type identity." *Genome Research* 2011, 21(10):1757–1767. PMID: 21750106.
- [31] The ENCODE Project Consortium. "A user's guide to the encyclopedia of DNA elements (ENCODE)." **PLoS Biology** 2011, 9(4):e1001046. PMID: 21526222.
- [32] **Boyle AP**, Song L, Lee B, London D, Keefe D, Birney E, Iyer VR, †Crawford GE, †Furey TS. "High-resolution genome-wide in vivo footprinting of diverse transcription factors in human cells." *Genome Research* 2011, 21:456–464. PMID: 21106903.
- [33] *Stitzel ML, *Sethupathy P, Pearson DS, Chines PS, Song L, Erdos MR, Welch R, Parker SCJ, Boyle AP, Scott LJ, Margulies EH, Boehnke M, Furey TS, Crawford GE, Collins FS. "Global epigenomic analysis of primary human pancreatic islets provides insights into type 2 diabetes susceptibility loci." *Cell Metabolism* 2010, 12(5):443–455. PMID: 21035756.
- [34] McDaniell R, Lee B, Song L, Liu Z, Boyle AP, Erdos MR, Scott LJ, Morken MA, Kucera KS, Battenhouse A, Keefe D, Collins FS, Willard HF, Lieb JD, Furey TS, †Crawford GE, †lyer VR, †Birney E. "Heritable individual-specific and allele-specific chromatin signatures in humans." Science 2010, 328(5975):235–239. PMID: 20299549.
- [35] Georgiev S, **Boyle AP**, Jayasurya K, Mukherjee S, Ohler U. "Evidence-ranked motif identification." *Genome Biology* 2010, 11(2):R19. PMID: 20156354.
- [36] Babbitt CC, Fedrigo O, Pfefferle AD, Boyle AP, Horvath JE, Furey TS, Wray GA. "Both noncoding and protein-coding RNAs contribute to gene expression evolution in the primate brain." *Genome Biology and Evolution* 2010, 2:67–79. PMID: 20333225.
- [37] Xu X, Tsumagari K, Sowden J, Tawil R, Boyle AP, Song L, Furey TS, Crawford GE, Ehrlich M. "DNasel hypersensitivity at gene-poor, FSH dystrophy-linked 4q35.2." *Nucleic Acids Research* 2009, 37(22):7381–7393. PMID: 19820107.
- [38] **Boyle AP**, Furey TS. "High-resolution mapping studies of chromatin and gene regulatory elements." *Epigenomics* 2009, 1(2):319–329. PMID: 20514362.
- [39] **Boyle AP**, Guinney J, Crawford GE, Furey TS. "F-Seq: a feature density estimator for high-throughput sequence tags." *Bioinformatics* 2008, 24(21):2537–2538. PMID: 18784119.
- [40] **Boyle AP**, Davis S, Shulha HP, Meltzer P, Margulies EH, Weng Z, †Furey TS, †Crawford GE. "High-resolution mapping and characterization of open chromatin across the genome." *Cell* 2008, 132(2):311–322. PMID: 18243105.
- [41] **Boyle AP**, Boyle JA. "Global analysis of microbial translation initiation regions." In *Journal of the Mississippi Academy of Sciences*, *Volume 48* 2003:138–150.

- [42] **Boyle AP**, Boyle JA. "Visualization of aligned genomic open reading frame data." *Biochemistry and Molecular Biology Education* 2003, 31:64–68.
- [43] Wan X, Boyle JA, Bridges SM, Boyle AP. "Interactive clustering for exploration of genomic data." In *Proceedings of the Artificial Neural Networks in Engineering Conference*, Volume 12, St. Louis, MO 2002:753–758.

Patents

- [44] Karczewski K, Snyder M, Butte AJ, Dudley JT, Hong E, Boyle A, Cherry MJ, Park J. "Method and system for the use of biomarkers for regulatory dysfunction in disease." 2018, (Granted United States patent 9,946,835).
- [45] Karczewski K, Snyder M, Butte AJ, Dudley JT, Hong E, Boyle A, Cherry MJ, Park J. "Method and system for the use of biomarkers for regulatory dysfunction in disease (continuation of u.s. patent application no. 13/592,292)." 2018, (Pending United States patent Appl. 15/954,354).