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

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 (Multi PI: Cherry, Boyle; Genomic Research Project PI: Boyle)
NIH/NHGRI Total Costs: \$2,171,753

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

The goal of this project is the study of the turnover of cohesin binding sites in the human and mouse genomes.

Eleanor and Larry Jackier U-M/Technion and Weizmann Collaborative Research Grant 2017-2018

(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.

R35 HL135824 2017-2024

(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 under-

lying association with devastating cardiovascular diseases.

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

NIH/NHGRI

(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

traits

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

genome screening.

Professional Service

Service

DCM&B Diversity, Equity, & Inclusion representative 2018-current

Lab Safety Liason for DCM&B 2018-current

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

EBS Faculty IT Committee 2017-current

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

DCM&B Admissions Committee 2015-current

2015-2017 DHG Computational Support Committee

DCM&B Retreat Planing Committee Chair (including 1st annual) 2015-2016 Ad hoc admissions reviewer, University of Michigan DCM&B 2014 Duke Computational Biology & Bioinformatics student committee 2008-2009

Memberships

Member, American Society of Human Genetics (ASHG) 2018-current

Member, International Society for Computational Biology (ISCB) 2013-current

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

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

Reviewing Activity

Ad hoc reviewer for the journals: Nature Genetics, Genome Research, Genome Biology, Na-Since 2009 ture Neuroscience, Nature Communications, Nature Protocols, Bioinformatics, Nucleic Acids Research, BMC Bioinformatics, Oncotarget, Scientific Reports, Atherosclerosis, BioEssays, Gene Program Committee, Great Lakes Bioinformatics and Canadian Computational Biology Confer-2015-curent ence (GLBIO/CCBC) Program Committee, Algorithms for Computational Biology (ALCOB) 2015-2016 Reviewer for UK Medical Research Council (RCUK MRC) 2015 Reviewer for UK Biotechnology and Biological Sciences Research Council (RCUK BBSRC) 2015 Reviewer for Michigan Institute for Clinical & Health Research (MICHR) Postdoctoral Translational 2015 Scholars Program 2013-current Program Committee, Gene Regulation and Transcriptomics, ISMB/ECCB DNA Day Essay Contest Detailed Review Judge for ASHG 2012-2015 Distinguished contributor as a leading reviewer for the journal *Bioinformatics*

Teaching and Mentorship

Teaching

2019	-current	Bioinformatics Concepts and Algorithms (BIOINF 529) [Course Director]
2018	3-current	Lecturer, Mathematical and Theoretical Biology Institute, Arizona State University [2 lectures]
	2017	Panel member, U. Michigan "New Faculty Orientation to Corporate & Foundation Relations" [70]
		attendees]
	2017	Experimental Genetics Systems (HUMGEN 632) [Course Director]
	2016	Experimental Genetics Systems (HUMGEN 632) [1 discussion]
2015	-current	Gene Structure and Regulation (HUMGEN 541) [3 lectures + 2 discussions / yr.]
2015	-current	Bioinformatics Journal Club (BIOINF 602/603) [Course Director Fall 2018]
20	15–2017	Introduction to Bioinformatics & Computational Biology (BIOINF 527) [2 lectures + 3 labs / yr.]
20	15–2017	Basic Biology for Graduate Students with Quantitative Training (BIOINF 523) [2 lectures / yr.]
	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
F	Fall 2003	Lab TA for Isotopes Tech I (MS. State, BCH 4414)

Mentorship

Graduate Students

2017-current	Melissa Englund (Ph.D. Student, Human Genetics, University of Michigan)
2017–current	NIH Human Genetics Training Program (T32) Samuel Zhao (Ph.D. Student, Bioinformatics, University of Michigan)
	Rackham Graduate Student Research Grant (pre-candidate)
2016-current	Haley Amemiya (Ph.D. Student, Cellular and Molecular Biology, University of Michigan)
	NIH Cellular & Molecular Biology Training Program (T32)
	PIBS Excellence in Service Award
	Rackham Graduate Student Research Grant (pre-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)
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 2015–current Greg Farnum (Ph.D. Student, Cellular and Molecular Biology, University of Michigan)

Sierra Nishizaki (Ph.D. Student, Human Genetics, University of Michigan)

NIH Genome Science Training Program (T32)

Rackham Merit Fellow Rackham Summer Award

Additional Graduate Rotation Students

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

Undergraduate and High School Students

2016-2018	Cody Morterud (Undergraduate, UROP Computer Science, 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

2018-current	Marcus Sherman (Bioinformatics, University of Michigan)
2018-current	Christopher Lee (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-current	Christopher Lee (Biostatistics, University of Michigan)
2016-current	Mohd Hafiz Bin Mohd Rothi (Molecular, Cellular, and Developmental Biology, University of Michi-
	gan)
2015-current	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

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] <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.
- [2] 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.
- [3] 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.
- [4] *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.
- [5] Nishizaki SS, Boyle AP. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.
- [6] Diehl AG, Boyle AP. "Deciphering ENCODE." Trends in Genetics 2016, 32(4):238-249. PMID: 26962025.
- [7] Phanstiel DH, Boyle AP, Heidari N, Snyder MP. "Mango: A bias correcting ChIA-PET analysis pipeline." Bioinformatics 2015. PMID: 26034063.
- [8] *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.
- [9] *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.

- [10] *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.
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- [12] 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.
- [13] *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.
- [14] *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.
- [15] Boyle AP, Hong EL, Hariharan M, Cheng Y, Schaub MA, Kasowski M, Karczewski KJ, Park J, Hitz BC, Weng S, Cherry JM, Snyder M. "Annotation of functional variation in personal genomes using RegulomeDB." *Genome Research* 2012, 22(9):1790–1797. PMID: 22955989.
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- [20] *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.
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- [26] 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.
- [27] 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.
- [28] Boyle AP, Furey TS. "High-resolution mapping studies of chromatin and gene regulatory elements." *Epigenomics* 2009, 1(2):319–329. PMID: 20514362.
- [29] **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.
- [30] **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.
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- [32] **Boyle AP**, Boyle JA. "Visualization of aligned genomic open reading frame data." *Biochemistry and Molecular Biology Education* 2003, 31:64–68.
- [33] 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

[34] 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, (United States patent US 9,946,835 B2).