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: 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.

2017–2022 DBI-1651614

(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.

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 under-

lying association with devastating cardiovascular diseases.

2018-2023

R01 HD093570

(PI: Bielas; Co-I with Effort) Total Costs: \$2,304,265

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.

Completed

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

(PI: Boyle)

NIH/NHGRI 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.

2015-2017 | FG-2015-65465

(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

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

2018–current Lab Safety Liason for DCM&B

2017-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

2016-current DCM&B Faculty Recruitment Committee

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

2015-current 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

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 Reviewer for Internal Michigan Searle grants 2018 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 2015 Reviewer for Michigan Institute for Clinical & Health Research (MICHR) Postdoctoral Translational Scholars Program Program Committee, Gene Regulation and Transcriptomics, ISMB/ECCB 2013-current DNA Day Essay Contest Detailed Review Judge for ASHG 2012-2015 Distinguished contributor as a leading reviewer for the journal *Bioinformatics* 2012

Teaching and Mentorship

Teaching

2019-current	Bioinformatics Concepts and Algorithms (BIOINF 529) [Course Director]
2018-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-current	Introduction to Biocomputing Bootcamp (BIOSTAT/BIOINF/HUMGEN 606) [2 days / yr.]
2017-current	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]
2015–2017	Introduction to Bioinformatics & Computational Biology (BIOINF 527) [2 lectures + 3 labs / yr.]
2015–2017	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
Fall 2003	Lab TA for Isotopes Tech I (MS. State, BCH 4414)

Mentorship

Graduate Students

Melissa Englund (Ph.D. Student, Human Genetics, University of Michigan)
NIH Human Genetics Training Program (T32)

2017-current Samuel Zhao (Ph.D. Student, Bioinformatics, University of Michigan)
Rackham Graduate Student Research Grant (pre-candidate)
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

Shriya Sethuraman (Ph.D. Student, Bioinformatics, University of Michigan)
Shengcheng Dong (Ph.D. Student, Bioinformatics, University of Michigan)
Christopher Castro (Ph.D. Student, Bioinformatics, University of Michigan)
NIH Bioinformatics Training Program (T32)

Rackham Merit Fellow

Rackham Graduate Student Research Grant (pre-candidate)

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

Steve Ho (Rotation Student, Human Genetics, University of Michigan)
Bradley Crone (Rotation Student, Bioinformatics, University of Michigan)
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-current	Cody Morterud (Undergraduate, UROP Computer Science / Honors Capstone, University of Michi-
	gan)
2016-2017	Colten Williams (Undergraduate, UROP Computer Science, University of Michigan)
2016-2017	Courtney Asman (Undergraduate, Neuroscience, University of Michigan)
2014-2017	Maxwell Spadafore (Undergraduate, LS&A Honors Informatics, University of Michigan)
2013-2014	Natalie Ng (High School, Stanford Institutes of Medicine Summer Research)
2013-2014	Dana Wyman (Undergraduate, Biology, Stanford University)
2013	Justin Young (High School, Stanford Institutes of Medicine Summer Research)
2012	Melanie Connick (Undergraduate, Biology, University of New Mexico)
2012	Edward Dai (Undergraduate, Computer Science, Stanford University)

Doctoral Thesis Committees

2018-current	Stephen Carney (Cancer Biology, University of Michigan)
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)
2015–2018	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

2018	Zhi Carrie Li (Bioinformatics, University of Michigan)
2018	Kevin Hu (Bioinformatics, University of Michigan)

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 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) 2017 Shriya Sethuraman (Bioinformatics, University of Michigan) 2016 Jed Carlson (Bioinformatics, University of Michigan) 2016

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, <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.
- [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.

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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.
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- [22] **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.
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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.
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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.
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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).