(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.

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.

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.

2019 NVIDIA GPU Grant

(PI: Boyle)

NVIDIA Corporation

In-Kind Value: \$3,600

2019–2021 Precision Health Investigators Award

(co-PI: Todd, Boyle, Mills)

University of Michigan

Total Costs: \$300,000

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.

2019-2022 BA180048

(PI: Aguilar; Co-I with Effort)

DoD/USAMRAA

Total Costs: \$1,081,608

Understanding & Enhancing the Regenerative Capacity of Skeletal Muscle to Trauma by Targeting Muscle-Nerve Synergy

The goal of this project is to study the regenerative capacity of satellite cells in muscle by performing single cell analysis of expression and chromatin accessibility across differentiation and to explore the molecular mechanisms driving these changes.

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 F

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 NIH/NHLBI (PI: Willer; Co-I with Effort) 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

DHG Admissions Committee 2019-current DCM&B Diversity, Equity, & Inclusion Ally [Chair] 2018-current Lab Safety Liaison for DCM&B 2018-current Cellular and Molecular Biology Admissions Committee 2018-2019 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 DCM&B Admissions Committee 2015-2018 **DHG Computational Support Committee** 2015-2017 DCM&B Retreat Planing Committee Chair (including 1st annual) 2015-2016 Ad hoc admissions reviewer, University of Michigan DCM&B

Memberships

2008-2009

2013-current 2013-current 2012-2014 Member, American Society of Human Genetics (ASHG)
Member, International Society for Computational Biology (ISCB)
Member, American Association for the Advancement of Science (AAAS)
Member, Gamma Sigma Delta Agricultural Honor Society

Duke Computational Biology & Bioinformatics student committee

Reviewing Activity

Ad hoc reviewer (90 verified reviews) for the journals: Nature Genetics, Genome Research, Since 2009 Genome Biology, Nature Neuroscience, Nature Communications, Nature Protocols, Bioinformatics, Nucleic Acids Research, BMC Bioinformatics, Oncotarget, Scientific Reports, Atherosclerosis, BioEssays, Gene NIH/NIMH Study Section ZMH1 ERB-C (01) - PsychENCODE: Non-Coding Functional Elements 2019 in the Human Brain and Their Role in the Development of Psychiatric Disorders (Ad Hoc) 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 2019-current Biotechnology Program Committee, Studies of Phenotypes and Clinical Applications, ISMB/ECCB 2019-current Reviewer for Internal Michigan Searle applications 2018-2019 Program Committee, Great Lakes Bioinformatics and Canadian Computational Biology Confer-2015-2018 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 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 (F = Fall Term, W = Winter Term, S = Summer Term)

w₁₉, w₂₀ Bioinformatics Concepts and Algorithms (BIOINF 529) [Course Director]

F15, F16, F17, F18, F19	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 (NCF4757000) In Lecture 1	
	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	
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) 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) Rackham Graduate Student Research Grant (candidate)
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) Rackham Graduate Student Research Grant (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

Margarita Brovkina (Rotation Student, Cellular and Molecular Biology, University of Michigan)
 Steve Ho (Rotation Student, Human Genetics, 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)

Non-student Lab Volunteers

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

Undergraduate and High School Students

2019-current	David Wang (Undergraduate, UROP Computer Science, University of Michigan)
2019-current	Jack Lu (Undergraduate, UROP Computer Science, University of Michigan)
2019-current	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 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

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-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

Benjamin Yang (Biomedical Engineering, University of Michigan)
Maria Virgilio (Cellular and Molecular Biology, University of Michigan)

Zhi Carrie Li (Bioinformatics, University of Michigan) 2018 Kevin Hu (Bioinformatics, University of Michigan) 2018 Siyu Liu (Bioinformatics, University of Michigan) 2018 2018 Alexandra Weber (Bioinformatics, University of Michigan) 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**. "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] 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.
- [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, 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.
- [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.
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