

Alan P. Boyle

2017 Palmer Commons
100 Washtenaw Ave.
Ann Arbor, MI 48109-2218
apboyle@umich.edu
www.BoyleLab.org

Education

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

Academic Appointments

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

| | | |
|-----------|---|---|
| | 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 |
| 2016–2019 | DCM&B Faculty Recruitment Committee |
| 2016–2020 | DCM&B Seminar Series Committee [Chair] |
| 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 (96 verified reviews) 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 | 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

| | |
|-----------|---|
| 2018–2019 | 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

| | |
|--------------|--|
| 2020–current | Camille Mumm (Ph.D. Student, Human Genetics, University of Michigan) |
| 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 & 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 | Torrin 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) |

NIH Genome Science Training Program (T32)
Rackham Merit Fellow
Rackham Summer Award
Rackham Graduate Student Research Grant (candidate)

Additional Graduate Rotation Students

| | |
|------|---|
| 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 | 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 | Sheila Rasouli (Undergraduate, Neuroscience, University of Toronto) |
| 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 |
|-----------|--|

Publications

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

- [1] Qin T, Lee C, Cavalcante RG, Orchard P, Yao H, Zhang H, Wang S, Patil S, **Boyle AP**, Sartor MA. “Comprehensive enhancer-target gene assignments improve gene set level interpretation of genome-wide regulatory data.” **Submitted** 2020.
- [2] Zhao N, **Boyle AP**. “F-Seq2: improving the feature density based peak caller with dynamic statistics.” **bioRxiv** 2020.
- [3] Nishizaki SS, **Boyle AP**. “SEMplMe: A tool for integrating DNA methylation effects in transcription factor binding affinity predictions.” **bioRxiv** 2020.
- [4] *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.
- [5] *Tsunami M, *Sethuraman S, Coke AN, Rothu MH, **Boyle AP**, Wierzbicki AT. “Broad non-coding transcription suggests genome surveillance by RNA Polymerase V.” **Submitted** 2020.
- [6] Diehl AG, **Boyle AP**. “MapGL: Inferring evolutionary gain and loss of short genomic sequence features by phylogenetic maximum parsimony.” **BMC Bioinformatics** 2020, 21:416. PMID: 32962625.
- [7] The ENCODE Project Consortium. “Perspectives on ENCODE.” **Nature** 2020, 583(7818):693–698.
- [8] The ENCODE Project Consortium. “Expanded encyclopaedias of DNA elements in the human and mouse genomes.” **Nature** 2020, 583(7818):699–710.
- [9] Ouyang N, **Boyle AP**. “TRACE: transcription factor footprinting using chromatin accessibility data and DNA sequence.” **Genome Research** 2020, 30:1040–1046. PMID: 32660981.
- [10] 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.
- [11] 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.

- [12] 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.
- [13] 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.
- [14] Amemiya HM, Kundaje A, **Boyle AP**. "The ENCODE Blacklist: Identification of Problematic Regions of the Genome." *Scientific Reports* 2019, 9:9354. PMID: 31249361.
- [15] Dong S, **Boyle AP**. "Predicting functional variants in enhancer and promoter elements using RegulomeDB." *Human Mutation* 2019, 33(8):831. PMID: 31228310.
- [16] 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.
- [17] 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.
- [18] 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.
- [19] Nielsen JB, Fritsche LG, Zhou W, Teslovich TM, Holmen OL, Gustafsson S, Gabrielsen ME, Schmidt EM, Beaumont R, Woford 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.
- [20] 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.
- [21] *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.
- [22] Nishizaki SS, **Boyle AP**. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.
- [23] Diehl AG, **Boyle AP**. "Deciphering ENCODE." *Trends in Genetics* 2016, 32(4):238–249. PMID: 26962025.
- [24] Phanstiel DH, **Boyle AP**, Heidari N, Snyder MP. "Mango: A bias correcting ChIA-PET analysis pipeline." *Bioinformatics* 2015. PMID: 26034063.
- [25] *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.

- [26] *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, Kawi 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, Skoultschi A, Gosh S, Distech 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.
- [27] ***Boyle AP**, *Araya CL, Brdlik C, Cayting P, Cheng C, Cheng Y, Gardner K, Hillier LW, Janette J, Jiang L, Kasper D, Kawi 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, Weissdepp 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.
- [28] Araya CL, Kawi 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.
- [29] 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.
- [30] *Xie D, ***Boyle AP**, *Wu L, Kawi T, Zhai J, Snyder M. "Dynamic trans-acting factor colocalization in human cells." *Cell* 2013, 155(3):713–724. PMID: 24243024.
- [31] *Kasowski M, *Kyriazopoulou-Panagiotopoulou S, *Grubert F, *Zaug 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.
- [32] **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.
- [33] 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.
- [34] The ENCODE Project Consortium. "An integrated encyclopedia of DNA elements in the human genome." *Nature* 2012, 489(7414):57–74. PMID: 22955616.
- [35] *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, Fietze S, Fu Y, Gertz J, Grubert F, Harman A, Jain P, Kasowski M, Lacroute P, Leng J, Lian J, Monahan H, O'Geen H, Ouyang Z, Partridge EC, Patocsil 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.
- [36] *Chen R, *Mias GI, *Li-Pook-Than J, *Jiang L, Lam HYK, Chen R, Mirami 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.

- [37] *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 DNaseI and FAIRE identifies regulatory elements that shape cell-type identity." *Genome Research* 2011, 21(10):1757–1767. PMID: 21750106.
- [38] The ENCODE Project Consortium. "A user's guide to the encyclopedia of DNA elements (ENCODE)." *PLoS Biology* 2011, 9(4):e1001046. PMID: 21526222.
- [39] **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.
- [40] *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.
- [41] 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, †Iyer VR, †Birney E. "Heritable individual-specific and allele-specific chromatin signatures in humans." *Science* 2010, 328(5975):235–239. PMID: 20299549.
- [42] Georgiev S, **Boyle AP**, Jayasurya K, Mukherjee S, Ohler U. "Evidence-ranked motif identification." *Genome Biology* 2010, 11(2):R19. PMID: 20156354.
- [43] 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.
- [44] Xu X, Tsumagari K, Sowden J, Tawil R, **Boyle AP**, Song L, Furey TS, Crawford GE, Ehrlich M. "DNaseI hypersensitivity at gene-poor, FSH dystrophy-linked 4q35.2." *Nucleic Acids Research* 2009, 37(22):7381–7393. PMID: 19820107.
- [45] **Boyle AP**, Furey TS. "High-resolution mapping studies of chromatin and gene regulatory elements." *Epigenomics* 2009, 1(2):319–329. PMID: 20514362.
- [46] **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.
- [47] **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.
- [48] **Boyle AP**, Boyle JA. "Global analysis of microbial translation initiation regions." In *Journal of the Mississippi Academy of Sciences, Volume 48* 2003:138–150.
- [49] **Boyle AP**, Boyle JA. "Visualization of aligned genomic open reading frame data." *Biochemistry and Molecular Biology Education* 2003, 31:64–68.
- [50] 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

- [51] 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).
- [52] 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).