# Alan P. Boyle

# **Education**

Doctor of Philosophy, Computational Biology and Bioinformatics Duke University, Durham, NC
 Bachelor of Science, Biochemistry and Molecular Biology Bachelor of Science, Computer Science
 Mississippi State University, Starkville, MS

# **Academic Appointments**

Assistant Professor, Department of Computational Medicine & Bioinformatics 2014-present 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

| 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**

| C         |  |                           |
|-----------|--|---------------------------|
| 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 o | f genomic regions into an |
|           | equivalent genome-wide assay.  |                           |
| 2015–2017 | Fellowship in Computational & Evolutionary Molecular Biology         | (PI: Boyle)               |

Alfred P. Sloan Foundation

U41 HG009293 2017-2020

Total Costs: \$2,171,753 NIH/NHGRI

RegulomeDB: A Resource for the Human Regulome

This project seeks to expand and support a RegulomeDB, a database for prioritizing and predicting

functional variants in the human genome.

DBI-1651614 2017-2022

(PI: Bovle)

Total Costs: \$50,000

(Multi PI: Cherry, Boyle)

Total Costs: \$979,984 NSF/BIO/DBI 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.

#### Completed

R01 HL130705 2016-2020

(PI: Willer; Co-I with Effort) Total Costs: \$2,784,005

NIH/NHLBI

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 whole-

genome screening.

# **Professional Service**

#### Service

DHG Faculty Recruitment and Promotions Committee 2017-current DCM&B Faculty Recruitment Committee 2016-current

DCM&B Seminar Series Committee Chair 2016-current DCM&B Admissions Committee 2015-current

**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 2014 Member, International Society for Computational Biology (ISCB) 2013-current Member, American Association for the Advancement of Science (AAAS) 2012-current Duke Computational Biology & Bioinformatics student committee 2008-2009

Member, Gamma Sigma Delta Agricultural Honor Society 2005-current

### **Reviewing Activity**

Since 2009 Ad hoc reviewer for the journals: Nature Genetics, Genome Research, Genome Biology, Nature Neuroscience, Nature Communications, Nature Protocols, Bioinformatics, BMC Bioinformatics,

Oncotarget, Scientific Reports, Atherosclerosis, BioEssays, Gene

Program Committee, Great Lakes Bioinformatics and Canadian Computational Biology Conference 2015-curent

(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

2012–2015 DNA Day Essay Contest Detailed Review Judge for ASHG
2012 Distinguished contributor as a leading reviewer for the journal *Bioinformatics* 

# **Teaching and Mentorship**

### **Teaching**

Bioinformatics Seminar Course (BIOINF 601) [Course Director] 2017-current Experimental Genetics Systems (HUMGEN 632) [1 discussion] 2016 Gene Structure and Regulation (HUMGEN 541) [3 lectures + 2 discussions / yr.] 2015-current Introduction to Bioinformatics & Computational Biology (BIOINF 527) [2 lectures + lab / yr.] 2015-current Bioinformatics Journal Club (BIOINF 602/603) 2015-current Basic Biology for Graduate Students with Quantitative Training (BIOINF 523) [2 lectures / yr.] 2015-current BIOINF 527 Panel on "Challenges in Biology, Biomedicine, Data & Analysis" 2014 Co-taught Cold Spring Harbor Systems Biology Pre-meeting Workshop 2010 Duke student panelist for "How to prepare for and get into graduate school" 2009 Taught Duke mini-course on Genome Browsers & Databases 2008 Lab TA for Isotopes Tech I (MS. State, BCH 4414) Fall 2003

# Mentorship

### **Graduate Students**

| 2017-current | Melissa Englund (Ph.D. Student, Human Genetics, University of Michigan)               |
|--------------|---|
|              | NIH Human Genetics Training Program (T32)   |
| 2017-current | Samuel Zhao (M.S. Student, Bioinformatics, University of Michigan)                    |
| 2016-current | Haley Amemiya (Ph.D. Student, Cellular and Molecular Biology, University of Michigan) |
|              | NIH Cellular & Molecular Biology Training Program (T32)                               |
| 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  |
| 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)   |
| 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  |

# **Additional Graduate Rotation Students**

| 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)    |

# **Undergraduate and High School Students**

| 2016-2017 | 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**

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 2015–2017 Ari Allyn-Feuer (Bioinformatics, University of Michigan)
Raymond Cavalcante (Bioinformatics, University of Michigan)

2015–2017 Zhengting Zou (Bioinformatics, University of Michigan)

# **Industry Experience**

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

**Publications** \* Indicates co-first authorship

- [1] 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.
- [2] Nishizaki SS, **Boyle AP**. "Mining the Unknown: Assigning Function to Noncoding Single Nucleotide Polymorphisms." *Trends in Genetics* 2017, 33:34–45. PMID: 27939749.
- [3] Diehl AG, Boyle AP. "Deciphering ENCODE." Trends in Genetics 2016, 32(4):238-249. PMID: 26962025.
- [4] Phanstiel DH, **Boyle AP**, Heidari N, Snyder MP. "Mango: A bias correcting ChIA-PET analysis pipeline." *Bioinformatics* 2015. PMID: 26034063.
- [5] 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.
- [6] 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.
- [7] **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.
- [8] Araya CL, Kawli T, Kundaje A, Jiang L, Wu B, Vafeados D, Terrell R, Weissdepp P, Gevirtzman L, Mace D, Niu W, **Boyle AP**, Xie D, Ma L, Murray JI, Reinke V, Waterston RH, Snyder M. "Regulatory analysis of the C. elegans genome with spatiotemporal resolution." *Nature* 2014, 512(7515):400–405. PMID: 25164749.

- [9] 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.
- [10] \*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.
- [11] \*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.
- [12] **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.
- [13] 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.
- [14] The ENCODE Project Consortium. "An integrated encyclopedia of DNA elements in the human genome." *Nature* 2012, 489(7414):57–74. PMID: 22955616.
- [15] \*Gerstein MB, \*Kundaje A, \*Hariharan M, \*Landt SG, \*Yan KK, \*Cheng C, \*Mu XJ, \*Khurana E, \*Rozowsky J, \*Alexander R, \*Min R, \*Alves P, Abyzov A, Addleman N, Bhardwaj N, **Boyle AP**, Cayting P, Charos A, Chen DZ, Cheng Y, Clarke D, Eastman C, Euskirchen G, Frietze S, Fu Y, Gertz J, Grubert F, Harmanci A, Jain P, Kasowski M, Lacroute P, Leng J, Lian J, Monahan H, O'Geen H, Ouyang Z, Partridge EC, Patacsil D, Pauli F, Raha D, Ramirez L, Reddy TE, Reed B, Shi M, Slifer T, Wang J, Wu L, Yang X, Yip KY, Zilberman-Schapira G, Batzoglou S, Sidow A, Farnham PJ, Myers RM, Weissman SM, Snyder M. "Architecture of the human regulatory network derived from ENCODE data." *Nature* 2012, 489(7414):91–100. PMID: 22955619.
- [16] \*Chen R, \*Mias GI, \*Li-Pook-Than J, \*Jiang L, Lam HYK, Chen R, Miriami E, Karczewski KJ, Hariharan M, Dewey FE, Cheng Y, Clark MJ, Im H, Habegger L, Balasubramanian S, O'Huallachain M, Dudley JT, Hillenmeyer S, Haraksingh R, Sharon D, Euskirchen G, Lacroute P, Bettinger K, **Boyle AP**, Kasowski M, Grubert F, Seki S, Garcia M, Whirl-Carrillo M, Gallardo M, Blasco MA, Greenberg PL, Snyder P, Klein TE, Altman RB, Butte AJ, Ashley EA, Gerstein M, Nadeau KC, Tang H, Snyder M. "Personal omics profiling reveals dynamic molecular and medical phenotypes." *Cell* 2012, 148(6):1293–1307. PMID: 22424236.
- [17] \*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.
- [18] The ENCODE Project Consortium. "A user's guide to the encyclopedia of DNA elements (ENCODE)." *PLoS Biology* 2011, 9(4):e1001046. PMID: 21526222.
- [19] **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.
- [20] \*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.
- [21] 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.
- [22] Georgiev S, **Boyle AP**, Jayasurya K, Mukherjee S, Ohler U. "Evidence-ranked motif identification." *Genome Biology* 2010, 11(2):R19. PMID: 20156354.
- [23] 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.

- [24] 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.
- [25] **Boyle AP**, Furey TS. "High-resolution mapping studies of chromatin and gene regulatory elements." *Epigenomics* 2009, 1(2):319–329. PMID: 20514362.
- [26] **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.
- [27] **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.
- [28] **Boyle AP**, Boyle JA. "Global analysis of microbial translation initiation regions." In *Journal of the Mississippi Academy of Sciences*, *Volume 48* 2003:138–150.
- [29] **Boyle AP**, Boyle JA. "Visualization of aligned genomic open reading frame data." *Biochemistry and Molecular Biology Education* 2003, 31:64–68.
- [30] 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**

[31] Karczewski K, Snyder M, Butte AJ, Dudley JT, Hong E, Boyle A, Cherry MJ. "Method and system for the use of biomarkers for regulatory dysfunction in disease." 2013, (US Patent Application 20130116931).