CURRICULUM VITAE

Name James P. Noonan, Ph.D.

School: Yale University School of Medicine and the Graduate School

Education

B.S.	Biology and English Literature, State University of New York at Binghamton, 1997; The University of Edinburgh, 1995-1996 (3rd year exchange)
Ph.D.	Genetics, Stanford University School of Medicine, 2004 Thesis: *The evolution of protocadherin gene cluster diversity* Advisor: Richard M. Myers, Ph.D.

Career/Academic Appointments

2004-2007	Postdoctoral Fellow, Genomics Division, Lawrence Berkeley National Laboratory, Berkeley, CA Advisor: Edward M. Rubin, M.D., Ph.D.
2007-2013	Assistant Professor, Department of Genetics, Yale School of Medicine, New Haven, CT
2013-2014	Associate Professor, Department of Genetics, Yale School of Medicine, New Haven, CT
2014-2016	Associate Professor, Department of Genetics and Department of Ecology and Evolutionary Biology, Yale University
2016-2018	Associate Professor (tenure), Department of Genetics and Department of Ecology and Evolutionary Biology, Yale University
2018-present	Associate Professor (tenure), Department of Genetics, Department of Ecology and Evolutionary Biology, and Department of Neuroscience, Yale University

Administrative Positions

2016-present	Executive Director for Genome Sciences, Yale Center for Genome Analysis
2017-present	Director, Training Program in the Genetics and Genomics of Human Disease

Professional Honors and Recognition

<u>International/National/Regional</u>

2008	Career Award, Edward Mallinckrodt Jr. Foundation
2004	NIH NRSA Postdoctoral Fellowship
1997	Phi Beta Kappa, State University of New York at Binghamton

Grant History

<u>Active</u>

Agency:	Nomis Foundation
Title:	"Deciphering the evolutionary origins of human brain uniqueness"
P.I.	James P. Noonan (Yale), Franck Polleux (Columbia)

Agency: NIH-NICHD
ID# 9R01 HD102030-10
Title: "Modeling uniquely human developmental gene regulatory networks using humanized mice"
P.I. James P. Noonan

Agency: Simons Foundation
ID#: 512694
Title: "Mapping ASD regulatory networks at cellular resolution in neurodevelopment"
P.I. James P. Noonan

Agency: NIH-NIMH
ID#: R01 MH114927
Title: "Neurogenetic investigations of obsessive-compulsive disorder"
P.I. Thomas Fernandez
Role on project: Co-Investigator

Agency: NIH-NICHD
ID# T32 HD007149-42
Title: Genetics and Genomics of Human Disease
P.I. James P. Noonan

Past (3 years)

Agency: Charles Hood Foundation
Title: "Discovering gene regulatory networks in early human brain development that contribute to autism spectrum disorder"
P.I. James P. Noonan

Invited Speaking Engagements, Presentations, Symposia and Workshops not Affiliated with Yale

International/National

2020 2nd Conference on Neurogenetics, Nature Conferences and New York University, New York, NY (online)

2020 Simons Foundation Autism Research Initiative Annual Meeting, New York, NY (online)

2020 Department of Physiology and Neurobiology, University of Connecticut, Storrs, CT (moved to 2021)

2019 Nomis Foundation, Zurich, Switzerland
"Deciphering the evolutionary origins of human brain uniqueness"

2019 Center for Pediatric Neuroscience, Cincinnati Children's Hospital, Cincinnati, OH
"Deciphering gene regulation at high resolution in human development and its disorders."

2019 Suddath Symposium, "Epigenetics: From Mechanisms to Tree of Life," Georgia Institute of Technology, Atlanta, GA

2017	Simons Foundation Autism Research Initiative Annual Meeting, New York, NY "Discovering *CHD8* regulatory networks disrupted in autism"
2017	ASBMB Special Symposium: Evolution and Core Processes in Gene Expression Stowers Institute "Genetic models of human evolution"
2017	Society for Molecular Biology and Evolution Annual Conference, Invited Symposium "Genetic models of human evolution"
2017	"Science at the Edge" Seminar Series, Michigan State University "Genetic models of human evolution"
2017	BBC Graduate Programs Seminar Series, University of California, San Francisco "Genetic models of human evolution"
2015	CSHL Meeting: Behavior and Neurogenetics of Nonhuman Primates “Comparative epigenetic analysis of early primate cortical development”
2015	Stanley Center for Cognitive Genomics, Cold Spring Harbor Laboratory “Gene regulatory mechanisms in human cognition and its disorders”
2015	Department of Biology and the Institute of Genetic Medicine, Johns Hopkins University, Baltimore, MD “Evolution of distinctly human phenotypes”
2014	Molecular Psychiatry 2nd Annual Conference, San Francisco, CA "Regulatory convergence in Autism Spectrum Disorder"
2014	The Jackson Laboratory for Genomic Medicine, Farmington, CT "Long-range regulatory interactions during embryonic development"
2014	Simons Foundation Autism Research Initiative Annual Meeting, New York, NY "Regulatory convergence in Autism Spectrum Disorder"
2014	Society for Molecular Biology and Evolution Annual Conference, Invited Symposium, "Evolution of the human developmental regulome"
2014	Advances in Genome Biology and Technology Annual Meeting, Marco Island, FL "Epigenomic analysis in the developing human embryo"
2013	Department of Genetics, University of Pennsylvania, Philadelphia, PA "Evolution of the human developmental regulome"
2013	Society for Molecular Biology and Evolution Annual Conference, Invited Symposium, "The molecular basis of human traits"
2013	Department of Genome Sciences, University of Washington, Seattle, WA "Human-specific gene regulation in the developing embryo"
2013	Department of Anthropology, University of New Mexico, Albuquerque, NM "The evolution of human-specific developmental gene regulation"
2012	Department of Developmental Biology, Stanford University, Palo Alto, CA "Evolution of the human developmental regulome"
2012	Department of Human Genetics, University of Chicago, Chicago, IL “Deciphering uniquely human developmental gene regulation”
2011	Genome Institute of Singapore, Singapore “Deciphering uniquely human gene regulation”
2011	22nd Meeting of the German Society of Human Genetics, Regensburg, Germany

	"Gene regulation and the origins of human biological uniqueness"
2011	CARTA Symposium: The Genetics of Humanness, San Diego, CA "Uniquely human gene regulation"
2011	Advances in Genome Biology and Technology Meeting, Marco Island, FL "Next generation sequencing at the Yale Center for Genome Analysis"
2011	Genome Sciences Symposia, Lawrence Berkeley National Laboratory "Deciphering uniquely human gene regulation"
2010	McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University, Baltimore, MD "Functional genomic analysis of human evolution"
2010	11th International Conference on Limb Development and Regeneration, Williamsburg, VA "Using next-generation sequencing to study gene regulation in human embryonic limb development"
2009	Human Genetics Gordon Conference, Biddeford, ME "Genome-wide identification and characterization of human-specific developmental regulatory functions"
2009	ISMB/ECCB Meeting, Stockholm, Sweden "The role of developmental *cis*-regulatory change in human evolution"
2009	National Center for Biotechnology Information, Bethesda, MD "Genome regulation and the evolution of human development"
2009	HudsonAlpha Institute for Biotechnology, Huntsville, AL "Exploring the genetic basis of human uniqueness"
2009	Department of Biology, Brandeis University, Waltham, MA "Genome regulation and the evolution of human development"
2008	CSHL Meeting: The Biology of Genomes (Session co-Chair) "Human-specific gain of function in a developmental enhancer"
2008	Department of Biology, Columbia University, New York, NY "The role of developmental gene regulatory change in human evolution"
2008	EvoS Seminar Series, State University of New York at Binghamton "Functional insights into human evolution from comparative genomics"
2008	Keystone Symposia: Complex Traits, Santa Fe, NM "Functional insights into the role of gene regulatory change in human evolution"
2007	Department of Biology, University of Utah, Salt Lake City, UT
2007	Department of Human Genetics, University of Michigan, Ann Arbor, MI
2007	Department of Genetics, University of Pennsylvania, Philadelphia, PA
2007	Department of Biology, Massachusetts Institute of Technology, Cambridge, MA
2007	Department of Molecular and Cell Biology, University of California, Berkeley, Berkeley, CA
2007	Division of Biology, California Institute of Technology, Pasadena, CA
2007	Department of Biology, University of Rochester, Rochester, NY

Peer-Reviewed Presentations & Symposia Given at Meetings Not Affiliated With Yale

International/National

2020	American College of Neuropsychopharmacology Annual Meeting "Functional and Systems Genomic Approaches to Psychiatric Disorders"
2015	International Meeting for Autism Research, Salt Lake City, UT "Deciphering regulatory networks that contribute to autism risk"
2015	CSHL Meeting on Systems Biology: Global Regulation of Gene Expression "Evolutionary changes in promoter and enhancer activity during human corticogenesis"
2014	International Meeting for Autism Research "The CHD8 network in the developing brain is enriched for ASD risk genes"
2012	CSHL Meeting on Systems Biology: Global Regulation of Gene Expression: "Chromatin profiling of human embryonic tissues identifies enhancers with human-specific functions"

Professional Service

Peer Review Groups/Grant Study Sections

2018	Human Developmental Biology Initiative, Wellcome Trust (UK)
2015	Genome Variation and Evolution Study Section (*ad hoc*), NIH
2013	External reviewer for Wellcome Trust Investigator Award program (UK)
2013	External reviewer for NSF CAREER program
2010	Genome Canada: Large-Scale Applied Research Project Competition

Journal Service

2008-present	Reviewer for: *Science*, *Nature*, *PNAS*, *Nature Neuroscience, PLoS Genetics, Nature Reviews Genetics, Genome Research, Genome Biology*, *Molecular Biology and Evolution*, others
2008-2014	Member, Editorial Board, *Genome Research*

Service for Professional Organizations

Human Genetics in NYC

2019-present	Yale School of Medicine Liaison; Program Committee, October 29 biannual meeting, American Museum of Natural History

American Society of Human Genetics

2012	Organizer, Invited Session, "Gene Regulatory Change: The Engine of Human Evolution," ASHG 62nd Annual Meeting
2010	Organizer, Invited Session, "Gene Regulation in Human Disease and Evolution," ASHG 60th Annual Meeting

Society for Molecular Biology and Evolution

2016 Organizer, Invited Symposium, "Genetic Mechanisms Influencing Morphological Evolution," SMBE Annual Meeting

Yale University Service

Yale University Committees

2015-present Implementation and Steering Committees, Yale Center for Biomedical Data Science

2009-2010, 2013 Admissions Committee, MCGD track, BBS

2008-present Yale Center for Genome Analysis Advisory Committee

Medical School Committees

2020 YSM Strategic Planning Committee, Developmental Brain Disorders, Autism and Schizophrenia

2015 Basic Science Strategic Planning Subcommittee on Bioinformatics (YSM)

2015-present Yale Center for Genome Editing Advisory Committee

2012 Faculty Advisory Committee, Deputy Chief Information Officer for Academic IT candidate search

Departmental Committees

2017-present Faculty Advisory Working Group, Department of Genetics

2014-present Genetics Faculty Search Committees

Public Service

2012 Consultant, NOVA/WGBH Production, *Cracking Your Genetic Code*

2010 Featured Expert and Consultant, NOVA/WGBH Production, *What Darwin Never Knew*

Bibliography

Peer-Reviewed Original Research

1: Neff NF, Ellis NA, Ye TZ, **Noonan J**, Huang K, Sanz M, Proytcheva M. The DNA helicase activity of BLM is necessary for the correction of the genomic instability of bloom syndrome cells. *Mol. Biol. Cell* 10(3): 665-76 (1999).

2: Wu Q, Zhang T, Cheng JF, Kim Y, Grimwood J, Schmutz J, Dickson M, **Noonan JP**, Zhang MQ, Myers RM, Maniatis T. Comparative DNA sequence analysis of mouse and human protocadherin gene clusters. *Genome Res*. 11(3): 389-404 (2001).

3: Yankiwski V, **Noonan JP**, Neff NF. The C-terminal domain of the Bloom syndrome DNA helicase is essential for genomic stability. *BMC Cell Biol*. 2: 11 (2001).

4: **Noonan JP**, Li J, Nguyen L, Caoile C, Dickson M, Grimwood J, Schmutz J, Feldman MW, Myers RM. Extensive linkage disequilibrium, a common 16.7-kilobase deletion, and evidence of balancing selection in the human protocadherin alpha cluster. *Am. J. Hum. Genet.* 72(3): 621-35 (2003).

5: **Noonan JP**, Grimwood J, Schmutz J, Dickson M, Myers RM. Gene conversion and the evolution of protocadherin gene cluster diversity. *Genome Res.* 14(3): 354-66 (2004).

6: Schmutz J, [47 additional authors], **Noonan JP**, Pitluck S, Pollard M, Predki P, Priest J, Ramirez L, Retterer J, Rodriguez A, Rogers S, Salamov A, Salazar A, Thayer N, Tice H, Tsai M, Ustaszewska A, Vo N, Wheeler J, Wu K, Yang J, Dickson M, Cheng JF, Eichler EE, Olsen A, Pennacchio LA, Rokhsar DS, Richardson P, Lucas SM, Myers RM, Rubin EM. The DNA sequence and comparative analysis of human chromosome 5. *Nature* 431(7006): 268-74 (2004).

7: **Noonan JP**, Grimwood J, Danke J, Schmutz J, Dickson M, Amemiya CT, Myers RM. Coelacanth genome sequence reveals the evolutionary history of vertebrate genes. *Genome Res.* 14(12): 2397-405 (2004).

8: **Noonan JP**, Hofreiter M, Smith D, Priest JR, Rohland N, Rabeder G, Krause J, Detter JC, Pääbo S, Rubin EM. Genomic sequencing of Pleistocene cave bears. *Science* 309(5734): 597-9 (2005).

9: Prabhakar S, **Noonan JP**, Pääbo S, Rubin EM. Accelerated evolution of conserved noncoding sequences in humans. *Science* 314(5800): 786 (2006).

10: **Noonan JP**, Coop G, Kudaravalli S, Smith D, Krause J, Alessi J, Chen F, Platt D, Pääbo S, Pritchard JK, Rubin EM. Sequencing and analysis of Neanderthal genomic DNA. *Science* 314(5802): 1113-8 (2006).

11: Prabhakar S, Visel A, Akiyama JA, Shoukry M, Lewis KD, Holt A, Plajzer-Frick I, Morrison H, Fitzpatrick DR, Afzal V, Pennacchio LA, Rubin EM, **Noonan JP**. Human-specific gain of function in a developmental enhancer. *Science* 321(5894): 1346-50 (2008).

12. Marlatt SA, Kong Y, Cammett TJ, Korbel G, **Noonan JP**, Dimaio D. Construction and maintenance of randomized retroviral expression libraries for transmembrane protein engineering. *Protein Eng. Des. Sel.* 24(3): 311-20 (2011).

13: Ayoub AE, Oh S, Xie Y, Leng J, Cotney J, Dominguez MH, **Noonan JP**, Rakic P. Transcriptional programs in transient embryonic zones of the cerebral cortex defined by high-resolution mRNA sequencing. *Proc. Natl. Acad. Sci. USA.* 108(36): 14950-5 (2011).

14: Cotney J, Leng J, Oh S, DeMare LE, Reilly SK, Gerstein MB, **Noonan JP**. Chromatin state signatures associated with tissue-specific gene expression and enhancer activity in the embryonic limb. *Genome Res.* 22(6): 1069-80 (2012).

15: Bandyopadhyay U, Cotney J, Nagy M, Oh S, Leng J, Mahajan M, Mane S, Fenton WA, **Noonan JP**, Horwich AL. RNA-Seq profiling of spinal cord motor neurons from a presymptomatic SOD1 ALS mouse. *PLoS One* 8(1): e53575 (2013).

16: Clark VE, Erson-Omay EZ, Serin A, Yin J, Cotney J, Ozduman K, Avşar T, Li J, Murray PB, Henegariu O, Yilmaz S, Günel JM, Carrión-Grant G, Yilmaz B, Grady C, Tanrikulu B, Bakircioğlu M, Kaymakçalan H, Caglayan AO, Sencar L, Ceyhun E, Atik AF, Bayri Y, Bai H, Kolb LE, Hebert RM, Omay SB, Mishra-Gorur K, Choi M, Overton JD, Holland EC, Mane S, State MW, Bilgüvar K, Baehring JM, Gutin PH, Piepmeier JM, Vortmeyer A, Brennan CW, Pamir MN, Kiliç T, Lifton RP, **Noonan JP**, Yasuno K, Günel M. Genomic analysis of non-NF2 meningiomas reveals mutations in TRAF7, KLF4, AKT1, and SMO. *Science* 339(6123): 1077-80 (2013).

17: Oh S, Song S, Grabowski G, Zhao H, **Noonan JP**. Time series expression analyses using RNA-seq: a statistical approach. *Biomed. Res. Int.* 2013: 203681 (2013).

18: DeMare LE, Leng J, Cotney J, Reilly SK, Yin J, Sarro R, **Noonan JP**. The genomic landscape of cohesin-associated chromatin interactions. *Genome Res.* 23(8): 1224-34 (2013).

19: Cotney J, Leng J, Yin J, Reilly SK, DeMare LE, Emera D, Ayoub AE, Rakic P, **Noonan JP**. The evolution of lineage-specific regulatory activities in the human embryonic limb. *Cell* 154(1): 185-96 (2013).

20: Pavlicev M, Wagner GP, **Noonan JP**, Hallgrímsson B, Cheverud JM. Genomic correlates of relationship QTL involved in fore- versus hind limb divergence in mice. *Genome Biol. Evol.* 5(10): 1926-36 (2013).

21: Willsey AJ, Sanders SJ, Li M, Dong S, Tebbenkamp AT, Muhle RA, Reilly SK, Lin L, Fertuzinhos S, Miller JA, Murtha MT, Bichsel C, Niu W, Cotney J, Ercan-Sencicek AG, Gockley J, Gupta AR, Han W, He X, Hoffman EJ, Klei L, Lei J, Liu W, Liu L, Lu C, Xu X, Zhu Y, Mane SM, Lein ES, Wei L, **Noonan JP**, Roeder K, Devlin B, Sestan N, State MW. Coexpression networks implicate human midfetal deep cortical projection neurons in the pathogenesis of autism. *Cell* 155(5): 997-1007 (2013).

22: Liu L, Lei J, Sanders SJ, Willsey AJ, Kou Y, Cicek AE, Klei L, Lu C, He X, Li M, Muhle RA, Ma'ayan A, **Noonan JP**, Sestan N, McFadden KA, State MW, Buxbaum JD, Devlin B, Roeder K. DAWN: a framework to identify autism genes and subnetworks using gene expression and genetics. *Mol. Autism* 5(1): 22 (2014).

23: Cotney J, **Noonan JP**. Chromatin immunoprecipitation with fixed animal tissues and preparation for high-throughput sequencing. *Cold Spring Harb. Protoc.* 2015(4): 419 (2015).

24: Cotney J, Muhle RA, Sanders SJ, Liu L, Willsey AJ, Niu W, Liu W, Klei L, Lei J, Yin J, Reilly SK, Tebbenkamp AT, Bichsel C, Pletikos M, Sestan N, Roeder K, State MW, Devlin B, **Noonan JP**. The autism-associated chromatin modifier CHD8 regulates other autism risk genes during human neurodevelopment. *Nat. Commun.* 6: 6404 (2015).

25: Reilly SK, Yin J, Ayoub AE, Emera D, Leng J, Cotney J, Sarro R, Rakic P, **Noonan JP**. Evolutionary changes in promoter and enhancer activity during human corticogenesis. *Science* 347 (6226): 1155-9 (2015).

26: Emera D, Yin J, Reilly SK, Gockley J, **Noonan JP**. Origin and evolution of developmental enhancers in the mammalian neocortex. *Proc. Natl. Acad. Sci. USA* 113(19): E2617-26 (2016).

27. Sousa AMM, Zhu Y, Raghanti MA, Kitchen RR, Onorati M, Tebbenkamp ATN, Stutz B, Meyer KA, Li M, Kawasawa YI, Liu F, Perez RG, Mele M, Carvalho T, Skarica M, Gulden FO, Pletikos M, Shibata A, Stephenson AR, Edler MK, Ely JJ, Elsworth JD, Horvath TL, Hof PR, Hyde TM, Kleinman JE, Weinberger DR, Reimers M, Lifton RP, Mane SM, **Noonan JP**, State MW, Lein ES, Knowles JA, Marques-Bonet T, Sherwood CC, Gerstein MB, Sestan N. Molecular and cellular reorganization of neural circuits in the human lineage. *Science* 358 (6366): 1027-32 (2017).

28. Sarro R, Kocher AA, Emera D, Uebbing S, Dutrow EV, Weatherbee SD, Nottoli T, **Noonan JP**. Disrupting the three-dimensional regulatory topology of the *Pitx1* locus results in overtly normal development. *Development* 145(7) pii: dev158550 (2018).

29. Wilderman A, Kron J, VanOudenhove J, **Noonan JP**, Cotney J. High resolution epigenomic analysis of early human craniofacial development. *Cell Reports* 23(5): 1581-1597 (2018).

30. Garcia-Santamarina S, Festa RA, Smith AD, Yu CH, Probst C, Ding C, Homer CM, Yin J, **Noonan JP**, Madhani H, Perfect JR, Thiele DJ. Genome-wide analysis of the regulation of Cu metabolism in *Cryptococcus neoformans*. *Mol. Microbiol*. Published online April 2, 2018. Available from doi.org/10.1111/mmi.13960.

31. Liu Y, Cicek AE, Liang Y, Li J, Muhle RA, Krenzer M, Mei Y, Wang Y, Knoblauch N, Morrison J, Jiang Y, Geller E, Li Z, Ionita-Laza I, Wu J, Xia K, **Noonan JP**, Sun ZS, He X. A statistical framework for mapping risk genes from de novo mutations in whole-genome sequencing studies. *Am. J. Hum. Genet*. 102(6): 1031-1047 (2018).

32. Blankvoort S, Witter MP, **Noonan J**, Cotney J, Kentros C. Marked diversity of unique cortical enhancers enables neuron-specific tools by Enhancer-Driven Gene Expression (EDGE). *Curr. Biol*. 28(13): 2103-2114 (2018).

33. Li M. *et al*. Integrative functional genomic analysis of human brain development and neuropsychiatric risks. *Science* 362(6420) pii: eaat7615 (2018).

34. Stewart TA, Liang C, Cotney J, **Noonan JP**, Sanger T, Wagner G. Evidence against tetrapod-wide digit identities and for a limited frame shift in bird wings. *Nat. Commun*. 10(1): 3244 (2019).

35. Geller E, Gockley J, Emera D, Uebbing S, Cotney J, **Noonan JP**. Massively parallel disruption of enhancers active during human corticogenesis. *bioRxiv* 852673. December 2, 2019. Available from: https://www.biorxiv.org/content/10.1101/852673v1. In revision.

36. Uebbing S, Gockley J, Reilly SK, Kocher AA, Geller E, Gandotra N, Scharfe C, Cotney J, **Noonan JP**. Massively parallel discovery of human-specific substitutions that alter neurodevelopmental enhancer activity. *bioRxiv* 865519. December 5, 2019. Available from: doi.org/10.1101/865519. In revision.

37. Dutrow EV, Emera D, Yim K, Uebbing S, Kocher AA, Krenzer M, Nottoli T, Burkhardt DB, Krishnaswamy S, Louvi A, **Noonan JP**. The Human Accelerated Region *HACNS1* modifies developmental gene expression in humanized mice. *bioRxiv* 873075. December 12, 2019. Available from: https://www.biorxiv.org/content/10.1101/2019.12.11.873075v1 In review.

Invited Reviews

1: **Noonan JP**. Regulatory DNAs and the evolution of human development. *Curr. Opin. Genet. Dev.* 19(6): 557-64 (2009).

2: **Noonan JP**, McCallion AS. Genomics of long-range regulatory elements. *Annu. Rev. Genomics Hum. Genet.* 11:1-23 (2010).

3: Sholtis SJ, **Noonan JP**. Gene regulation and the origins of human biological uniqueness. *Trends Genet.* 26(3):110-8 (2010).

4: **Noonan JP**. Neanderthal genomics and the evolution of modern humans. *Genome Res.* 20(5):547-53 (2010).

5: Reilly SK, **Noonan JP**. Evolution of gene regulation in humans. *Annu. Rev. Genomics Hum. Genet.* 17:45-67 (2016).

Mentoring

Postdoctoral Fellows and Associates

Justin Cotney, Ph.D.
Position and period of mentorship: Postdoctoral Associate, 2009-2013; Associate Research Scientist, 2013-2015
Research project: Identifying enhancers with human-specific function during limb development using comparative chromatin state mapping
Awards and honors: Rudolph J. Anderson postdoctoral fellowship; NIH K99 Pathway to Independence Award
Publications: 15 (References #13, 14, 15, 16, 18, 19, 21, 23, 24, 25, 29, 32, 34, 35, 36). Presentations: Talks at the 2013 Gordon Research Seminar on Human Genetics and Genomics; the 3rd Kavli Community Symposium, 2013; the 36th Annual Meeting of the Society of Craniofacial Genetics and Developmental Biology, 2013; UCSF Institute for Human Genetics, 2014
Current position: Assistant Professor, Department of Genetics and Genome Sciences, University of Connecticut Health Center

Jun Yin, Ph.D.
Position and period of mentorship: Postdoctoral Associate, 2012-2015
Research project: Integrated computational analysis of gene expression and chromatin state in human development, evolution and disease
Awards and honors: Brown-Coxe Postdoctoral Fellowship, Yale University
Publications: 7 (References #13, 18, 19, 24, 25, 26, 30).
Current position: Director, Bioinformatics Shared Resource at Sanford Burnham Prebys Medical Discovery Institute

Sunghee Oh, Ph.D.
Position and period of mentorship: Postdoctoral Associate, 2009-2011
Research project: Developing statistical methods for differential gene expression analysis using RNA-seq data
Publications: 4 (References #13, 14, 15, 17).
Current position: Instructor, Jeju National University, South Korea

Deena Emera, Ph.D.
Position and period of mentorship: Postdoctoral Associate, 2012-2017
Research project: Origin and evolution of developmental enhancers in the cortex
Awards and honors: NIH NRSA Postdoctoral Fellowship
Publications: 6 (References #19, 25, 26, 28, 35, 37). Presentations: Talks at the Society for Molecular Biology and Evolution annual meeting, 2014 and 2015.
Current position: Writer in Residence, Center for Reproductive Longevity and Equality, Buck Institute for Aging

Martina Krenzer, M.D.
Position and period of mentorship: Postdoctoral Fellow & Associate Research Scientist, 2016-present
Awards: Postdoctoral Fellowship from the German Research Foundation
Publications: 2 (References #31, 37). Presentations: Selected for a talk at 2019 Keystone Symposia: Windows on the Brain: Formation and Function of Synapses and Circuits and their Disruption in Disease

Severin Uebbing, Ph.D.
Position and period of mentorship: Postdoctoral Fellow, 2016-present
Awards: Postdoctoral Fellowship from the German Research Foundation
Publications: 4 (References #28, 35, 36, 37).

Marybeth Baumgartner, Ph.D.
Position and period of mentorship: Postdoctoral Associate, 2020-present

Associate Research Scientists and Residents

Rebecca Muhle, M.D., Ph.D.
Position and period of mentorship: Resident, Albert J. Solnit Integrated Adult and Child & Adolescent Psychiatry Program, Yale Child Study Center, 2011-2017; Instructor, Child Study Center, 2017-2019
Research project: The role of chromatin modifiers in autism spectrum disorder
Awards and honors: American Academy of Child and Adolescent Psychiatry Pilot Research Award; NIH K08 Mentored Clinical Scientist Research Career Award; Simons Foundation Bridge to Independence Award
Publications: 4 (References #21, 22, 24, 31). Presentations: Talks at the 2013 and 2015 International Meeting for Autism Research; the 2015, 2016, 2017 and 2018 Annual Meeting of the American Academy of Child and Adolescent Psychiatry; the 2018 and 2020 World Congress of Psychiatric Genetics
Current position: Assistant Professor, Columbia University Department of Psychiatry and the New York State Psychiatric Institute

Wei Niu, Ph.D.
Position and period of mentorship: Associate Research Scientist, 2013-2016
Research project: Genome editing tools to elucidate neurodevelopmental gene regulatory networks associated with autism risk
Publications: 2 (References #21, 24).
Current position: Research Scientist, University of Michigan

Graduate Students

Laura DeMare, Ph.D.
Position and period of mentorship: Graduate Student, 2008-2013
Thesis title: "A cohesin-mediated chromatin interactome during embryonic limb development."
Awards and Honors: Carolyn Slayman Prize for Outstanding Ph.D. Thesis in Genetics
Publications: 3 (References #14, 18, 19).
Current position: Product Manager, Single Cell Epigenomics, 10X Genomics

Steven K. Reilly, Ph.D.
Position and period of mentorship: Graduate Student, 2010-2015
Thesis title: "Evolutionary changes in promoter and enhancer activity during human corticogenesis."
Awards and Honors: NSF Graduate Research Fellowship; Carolyn Slayman Prize for Outstanding Ph.D. Thesis in Genetics
Publications: 9 (References #14, 18, 19, 21, 24, 25, 26, 36; Review #5).
Current position: NIH K99 Postdoctoral Fellow, Pardis Sabeti Lab, Broad Institute and Harvard University

Jing Leng, Ph.D. (Joint with Mark Gerstein, Computational Biology and Bioinformatics)
Position and period of mentorship: Graduate Student, 2009-2014
Thesis title: "Studying the evolution of gene regulation using next-generation sequencing: computational methods and data integration."
Publications: 6 (References #13, 14, 15, 18, 19, 25).
Current position: Bioinformatics Scientist, Illumina Inc.

Richard Sarro, Ph.D.
Position and period of mentorship: Graduate Student, 2012-2017
Thesis title: "Topologically directed disruption of a developmental enhancer through genome engineering."
Publications: 3 (References #18, 25, 28).
Current position: Senior Consultant, Simon-Kucher & Partners

Jake Gockley, Ph.D.
Position and period of mentorship: Graduate Student, 2013-2017
Thesis title: "Investigations into the genetic foundations of sex bias in Autism Spectrum Disorders and evolution of non-coding regulatory elements."
Publications: 4 (References #21, 26, 35, 36).
Current Position: Research Scientist, Sage Bionetworks

Evan Geller, Ph.D.
Position and period of mentorship: Graduate Student, 2014-2019
Thesis title: "Massively parallel genetic disruption of enhancers active during human corticogenesis."
Awards: Autism Speaks Weatherstone Fellowship
Publications: 3 (References #31, 35, 36). Presentations: Oral presentations at the 2019 CSHL Biology of Genomes Meeting and 2018 Society for Molecular Biology and Evolution Annual Meeting
Current Position: Postdoctoral Fellow, Neville Sanjana Lab, New York Genome Center/NYU

Emily Dutrow, Ph.D.
Position and period of mentorship: Graduate Student, 2015-2019
Thesis title: "Using humanized mouse models to study human evolution."

Publications: 2 (References #28, 37). Presentations: Oral presentation at the 2017 Society for the Study of Evolution Annual Meeting
Current Position: Postdoctoral Fellow, Elaine Ostrander Lab, NHGRI

Acadia Kocher
Position and period of mentorship: Graduate Student, 2016-present
Research project: Genetic and experimental characterization of enhancers with human-specific functions during brain development
Awards: NSF Graduate Research Fellowship
Publications: 3 (References #28, 36, 37).

Kristina Yim
Position and period of mentorship: Graduate Student, 2016-present
Research project: Characterizing autism risk gene regulatory networks at single-cell resolution during neurodevelopment
Awards: NSF Graduate Research Fellowship
Publications: 1 (Reference #37). Presentations: Short talk at the 2018 Molecular Psychiatry Association Annual Meeting

Mark Noble
Position and period of mentorship: Graduate Student, 2018-present
Research project: Genome-wide discovery of human-specific regulatory networks in human brain development and evolution
Awards: NSF Graduate Research Fellowship

Undergraduate Students

Sarah Abdallah, Honors thesis student, 2013-2014
Thesis title: "Identifying target genes of the autism-implicated transcription factor NFIA."
Current position: 1st Year Resident, Albert J. Solnit Integrated Adult and Child & Adolescent Psychiatry Program, Yale Child Study Center (Tom Fernandez, Research Mentor)
Awards: HHMI-ASHG Medical Research Fellows Award

Britt Bistis, Undergraduate Research Student, 2018-2020; Honors thesis student, 2020-2021
Awards: Yale College Dean's Research Fellowship, 2020

Teaching
2012-present: GENE 760: Genomic Methods for Genetic Analysis
Role: Course Director
Goal, content and major contribution: The purpose of this course is to provide first-year graduate students with an introduction to genomics and the analysis of high throughput sequence datasets. The focus is on next-generation sequencing (NGS) applications including RNA-seq, ChIP-seq, single cell genomics technologies, and exome and whole genome sequencing. The course consists of lectures, multiple problem sets, and a final project for which the students must propose a research topic, conduct analyses and write a report on their results. The lectures introduce students to genomics as a science and cover next-generation technologies for genome analysis, computational methods for understanding the massive datasets generated by those tools, and topics in global studies of gene regulation, genome architecture and the impact of human genetic variation on disease. The problem sets provide practical

training in genomic data analysis, including the use of existing pipeline and instruction on how to write custom scripts to process, analyze and interpret data. We use Python as our scripting language and R for statistical analysis.

I design the entire curriculum for this course each year, including selecting lecture topics and writing problem sets. I also give multiple lectures and recruit other Yale faculty to lecture on topics specific to their areas of expertise. As of 2020 over 170 students have completed the course.

Educational Leadership

I am the Director of the Training Program in the Genetics and Genomics of Human Disease, supported by a T32 grant from NICHD (T32 HD007149). This grant, now in its 42nd year, was successfully renewed under my direction for an additional 5-year term in 2019. The training program encompasses three core domains: 1) The genetics of human health and disease, encompassing disease gene discovery and the interpretation of human genetic variation; 2) Computational approaches to obtain biological insights from complex genomic datasets, both in human and model systems; and 3) Experimental studies in model organisms to understand how human genetic variation influences human biology and disease. To strengthen our program, we added additional training activities in our renewal, including required coursework in genomic data analysis, a Program Retreat, a Program Symposium, dedicated Research-in-Progress talks, and a Program Seminar Series. We currently support 7 Ph.D. students per year.