

Steven K. Reilly

Curriculum Vitae

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Harvard Organismic & Evolutionary Biology
Broad Institute of MIT and Harvard
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Education

Ph.D. in Genetics, **Yale University** 2015
Carolyn Slayman Prize for Outstanding Thesis

B.S. in Biological Sciences, **Carnegie Mellon University** 2009
University Honors
Chemistry Minor; molecular biology, genetics focus

Research

K99 & Kirschstein-NRSA Postdoctoral Fellow, **Sabeti Lab** 2016-Present
Broad Institute & Harvard University
Functional characterization of cis-regulatory variation, gene regulation, non-coding CRISPR screen development, machine learning methods to ID recent human adaptation.

NSF Graduate Research Fellow, **Noonan Lab** 2009-2015
Yale University School of Medicine
Comparative functional genomics of human brain evolution to identify human-specific enhancers.

Beckman Undergraduate Research Scholar, **López Lab** 2006-2009
Carnegie Mellon University
Functional Analysis of a Non-Exonic Recursive Splicing by Gene Replacement

HHMI Undergraduate Researcher, **Jones Lab** 2005
Carnegie Mellon University
Genomic characterization of the novel HK-97 bacteriophage.

Awards & Honors

NHGRI NIH Pathway to Independence Award (K99/R00) 2019

Travel Award - Broad Institute 2018

Kirschstein National Research Service Award (NRSA - F32) 2017

HHMI postdoctoral Fellowship Award 2016

Carolyn Slayman Thesis Prize for Distinguished Thesis 2015

Yale's Selection for Proquest National Thesis Award 2015

Research Feature by NIH Director Francis Collin	2015
Society of Molecular Biology & Evolution Poster Prize	2014
NSF Graduate Research Fellowship	2010
HHMI Undergraduate Travel Award	2009
Biology Advisory Committee Outreach Appreciate Award	2009
Carnegie Mellon University Honors	2009
Beckman Scholar Undergraduate Research Award	2008
Sigma Xi: Scientific Research Honors Society Induction	2008
HHMI Lab Resource Support Award	2007
HHMI Summer Research Institute Scholar	2006

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- Publications**
1. **Reilly SK**, Gosai SJ, Guitierrez A, Ulirsch JC, Kanai M, Berenzy D, Kales S, Butler GB, Gladden-Young A, Finucane HK, Sabeti PC, Tewhey R. Direct characterization of cis-regulatory elements and functional dissection of complex genetic associations using HCR-FlowFISH. (bioRxiv preprint) 2020. (*under revision at Nature Genetics*)
 2. Khetan S, Kales S, Kursawe R, Jillette A, **Reilly SK**, Ucar D, Tewhey R, Stitzel ML. Functional characterization of thousands of type 2 diabetes-associated and chromatin-modulating variants under steady state and endoplasmic reticulum stress. (bioRxiv preprint) 2020. <https://doi.org/10.1101/2020.02.12.939348> (*under revision at Nature Communications*)
 3. Lemieux J, Siddle KJ, Shaw BM, Loreth C, Schaffner S, Gladden-Young A, Adams G, Fink T, Tomkins-Tinch CH, Krasilnikova LA, Deruff KC, Rudy M, Bauer MR, Lagerborg KA, Normandin E, Chapman SB, **Reilly SK**, Anahtar MN, Lin AE, Carter A, Myhrvold C, Kembal M, Chaluvadi S, Cusick C, Flowers K, Neumann A, Cerrato F, Farhat M, Slater D, Harris JB, Branda J, Hooper D, Gaeta JM, Baggett TP, O'Connell J, Gnirke A, Lieberman TD, Philippakis A, Burns M, Brown C, Luban J, Ryan ET, Turbett SE, LaRocque RC, Hanage WP, Gallagher G, Madoff LC, Smole S, Pierce VM, Rosenberg ES, Sabeti P, Park DJ, MacInnis BL Phylogenetic analysis of SARS-CoV-2 in the Boston area highlights the role of recurrent importation and superspreading events (*medRxiv preprint*) 2020. <https://doi.org/10.1101/2020.08.23.20178236> (*under revision at Science*)
 4. Ray JP, de Boer CG, Fulco C, Lareau C, Kanai M, Ulirsch JU, Tewhey R, Ludwig L, **Reilly SK**, Bergman DT, Engreitz J, Issner R, Finucane HK, Lander ES, Regev A, Hacohen N. Prioritizing disease and trait causal variants at the TNFAIP3 locus using functional and genomic features. *Nature Communications* 2020. 11 (1), 1-13.
 5. 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 enhancer activity. 2019. (bioRxiv preprint) <https://doi.org/10.1101/865519> (*in press at Proceedings of the National Academy of Sciences*)

6. **Reilly SK**, Noonan JP. Evolution of Gene Regulation in Humans. *Annual Reviews of Genomics Human Genetics* 2016. 17:6.1–6.23
7. Tewhey R, Kotliar D, Park DS, Liu B, Winnicki S, **Reilly SK**, Andersen KG, Mikkelsen TS, Schaffner SF, Sabeti PC. Direct identification of hundreds of expression-modulating variants using a multiplexed reporter assay. *Cell* 165 (6), 1519-1529.
8. Emera D, Yin J, **Reilly SK**, Noonan JP. Origin and evolution of developmental enhancers in the mammalian neocortex. *Proceedings of the National Academy of Sciences*, April 25, 2016.
9. **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).
10. 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. *Nature Communications*. 6:6404 (2015).
11. 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).
12. 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).
13. DeMare LE, Leng J, Cotney J, **Reilly SK**, Yin J, Sarro R, Noonan JP. The genomic landscape of cohesin-associated chromatin interactions. *Genome Research*. 23(8):1224-34 (2013).
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 Research*. 22(6):1069-80 (2012).

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- Publications** (in prep.)
15. Griesemer D*, Xue J*, **Reilly SK***, Yang D, Ulirsch J, Novina C, Sabeti PC, Tewhey R, Genome-Wide Functional Screen of 3'UTR Variants Uncovers Putative Causal Variants Related to Selection and Disease. (*denotes equal contribution) (submitted to Cell)
 16. Vitti J, Gosai S, Griesmer, Brown EA, Xue J, Tewhey R, Sabeti PC, **Reilly SK.** Identification and characterization of adaptive regulatory variation in diverse human populations. (in preparation for Science)
 17. Gosai S, Vitti J, Xue J, Tewhey R, Sabeti PC, **Reilly SK.** DeepSweep: A novel deep convolutional neural network for detecting and localizing variants under positive selection. (in preparation for Nature Methods)
 18. **Reilly SK,** Xue J, Bauer M, Tewhey R, Sabeti PC. Multiplex non-coding CRISPR perturbations of the entire CTCF cistrome. (in preparation)
 19. Jagoda E, **Reilly SK,** Xue J, Dannemann M, Racimo F, Huerta-Sanchez E, Sankararaman S, Kelso J, Pagani L, Pardis Sabeti PC, Capellini TD. Direct Identification of Neanderthal Introgressed Genetic Variation Affecting Gene Expression in Modern Human Immune Cell Lines. (submitted to Nature Ecology and Evolution)
 20. Xue J, Tewhey R, Capellini T, Sabeti PC, **Reilly SK.** Comprehensive Characterization of hCONDELS Identified Human-Specific Gene Regulation. (in preparation)
 21. Kanai M, Ulirsch JC, Karjalainen J, Kurki M, Ishigaki K, Kamatani Y, **Reilly SK,** Sabeti PC, Tewhey RS, FinnGen, Okada Y, Daly MJ, Finucane HK. Insights into fine-mapping causal variants of complex traits from diverse populations. (in preparation)
 22. Ulirsch JC, Kanai M, Berenzy D, Kales S, Wang Q, Aguet F, **Reilly SK,** Mouri K, Dewey H, Lareau C, Cui R, K. Ardlie G, Sabeti PC, Daly MJ, Tewhey RS, Finucane HK. Functional and statistical fine-mapping of complex traits and gene expression across tissues reveals putative causal variants and nominates target genes. (in preparation)
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Research Presentations	Talk - American Society of Human Genetics (ASHG)	2020
	Invited Session Organizer - ASHG	2020
	Talk - Society of Molecular Biology and Evolution (SMBE)	2020
	Seminar - Allen Discovery Center for Brain Evolution	2019
	Talk - ASHG	2019
	Talk - ENCODE Annual Symposium	2019
	Seminar - Cell Circuits and Epigenetics	2019
	Talk - ENCODE Annual Symposium	2018
	Talk - NHGRI Trainee Meeting	2017
	Seminar - Medical and Population Genetics	2017
	Talk - ENCODE Consortium Meeting	2017
	Talk - ASHG	2016
	Poster - Allen Institute Human Brain Evolution Conference	2020
	Poster - ENCODE Consortium Meeting	2018
	Poster - NHGRI Trainee Meeting (Award Winner)	2018

Poster - SMBE	2015
Poster - SMBE	2014
Poster - CSHL Biology of Genomes	2014
Poster - Meeting of the Minds	2009
Poster - Beckman Scholars Research Symposium	2008
Poster - CMU Undergraduate Research Symposium	2008
Poster - HHMI Undergraduate Poster Session	2007
Poster - Meeting of the Minds	2008

Teaching	TA - Harvard, LS1B: Genetics, Genomics & Evolution	2017
	TA - Yale, Mechanisms of Developmental Biology	2014
	TA - Yale, Methods for Genomic Analysis (TA & course design)	2012
	TA - CMU, Experimental Biochemistry	2009
	TA - CMU, Summer Academy of Math + Sciences	2009
	TA - CMU, Experimental Techniques in Molecular Biology	2008

Mentoring	Ph.D. James Xue (Human-chimp conserved deletions)	2016-Present
	Ph.D. Sager Gosai (Machine learning sweep detection)	2017-Present
	Ph.D. Joe Vitti (Functional characterization of adaptation)	2016-2019
	B.S. Natalia Fuentes (Bayesian optimization of MPRA)	2018-Present
	High School Nico Santiago (Adaptation at <i>ERLIN1</i> locus)	2017-2018
	RAs Mentored four RAs in the Sabeti Lab	2017-Present

Leadership & Advocacy	NHGRI Research Training Expert Panel Member	2019-Present
	Inclusion, Diversity, Equity, and Allyship Ambassador	2020-Present
	Broad Summer Scholar Program Selection Committee	2018-Present
	Co-founder - Out@Broad: LGBTQ+ STEM advocacy group	2017-Present
	Co-founder - Broad Evolutionary Journal Club	2016-Present
	oSTEM: LGTQ+ advocacy group	2015
	Co-Chair Ivy+ Summit: Ivy League Graduate Leadership and Advocacy Conference	2014
	President, Yale Graduate & Professional Student Senate	2013-2014
	Board of Governors: Association of Yale Alumni	2013-2014
	Co-chair CMU Biology Student Advisory Committee	2007-2009
	CMU Biology Outreach Coordinator	2007-2008

Society Memberships	American Society of Human Genetics
	Society of Molecular Biology and Evolution

Press

NIH Director's Blog on 2015 Science Paper

- <https://directorsblog.nih.gov/2015/03/17/what-makes-our-brain-human-the-search-for-answers/>

Genome-wed article on human-specific brain development

- <https://www.genomeweb.com/epigenetics-research/study-links-regulatory-changes-during-development-human-cerebral-cortex>

Reporting on advocacy work around LGBTQ GWAS

- <https://www.nytimes.com/2019/08/29/science/gay-gene-sex.html>
- <https://www.wired.com/story/how-earnest-research-into-gay-genetics-went-wrong/>
- <https://www.broadinstitute.org/blog/opinion-all-science-what-did-we-learn>

Reporting on the development of DeepSweep

- <https://www.nature.com/articles/d41586-018-07225-z>

For work on COVID-19 genome sequencing in Boston

- <https://www.nytimes.com/2020/08/26/health/covid-19-superspreaders-boston.html>
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References

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