

Aaron R. Quinlan, Ph.D.

CONTACT INFORMATION	Associate Professor with tenure Department of Human Genetics Department of Biomedical Informatics Associate Director of the USTAR Center for Genetic Discovery University of Utah	(O): 801.585.0406 (Tw): @aaronquinlan aquinlan@genetics.utah.edu quinlanlab.org
EDUCATION	Boston College , Chestnut Hill, MA, USA Ph.D., Biology, 2008 College of William and Mary , Williamsburg, VA, USA B.S., Computer Science, 1997	
ACADEMIC EXPERIENCE	University of Virginia , Charlottesville, VA, USA <i>Assistant Professor of Public Health Sciences</i> Center for Public Health Genomics University of Virginia , Charlottesville, VA, USA <i>NRSA Postdoctoral Fellow (NHGRI)</i>	2011-2015 2008-2011
HONORS AND AWARDS	Editorial Board for Oxford Bioinformatics (2016-2018). Organizing Committee for the 2017 Wellcome Trust Genome Informatics meeting. Organizing Committee for the 2016 Wellcome Trust Genome Informatics meeting. Program Committee for the 2016 Intelligent Systems for Molecular Biology meeting. Finalist for the Benjamin Franklin for Open Access in the Life Sciences. 2016 Session Chair for Genome Informatics 2015 at Cold Spring Harbor Laboratories. Reviewer for 2015 American Society of Human Genetics Meeting. Reviewer for 2014 American Society of Human Genetics Meeting. Finalist (90 of >1000) for the Gordon and Betty Moore Data Driven Discovery Competition. 2014 Moderator for 2014 Genome Reference Consortium Meeting. Cambridge, England. Session Chair and reviewer for 2014 American Society of Human Genetics Meeting. Session Chair for Genome Informatics 2013 at Cold Spring Harbor Laboratories. Finalist for the Benjamin Franklin for Open Access in the Life Sciences. 2013 Co-chair, NHLBI Exome Sequence Project Structural Variation Group. 2011- Instructor for CSHL Advanced Sequencing Technologies Course. 2009- Fund for Excellence in Science and Technology Awardee, UVa. (1 of 5). 2011 Ruth L. Kirschstein (NRSA / F32) Postdoctoral Fellowship, NHGRI. 2009-2010 Presidential Fellowship, Boston College. 2004-2007	
JOURNAL REVIEW	<i>Ad hoc</i> reviewer for: <i>Nature Genetics</i> , <i>Nature Methods</i> , <i>Nature Biotechnology</i> , <i>AJHG</i> , <i>Genome Research</i> , <i>Genome Biology</i> , <i>Bioinformatics</i> , <i>BMC Bioinformatics</i> ,	

*Bioessays, Genes, and
IEEE Transactions On Computational Biology and Bioinformatics*

Guest editor for *PLoS Computational Biology*

GRANT REVIEW

NIH:

NIH GCAT Study Section, Washington, DC (10-Oct-2017 - 12-Oct-2017)

NIH GCAT Study Section, Washington, DC (8-Jun-2016 - 9-Jun-2016)

NIH GCAT Study Section, Chicago, IL (16-Oct-2013 - 17-Oct-2013)

Special Emphasis Panel for Computational Analysis of ENCODE data (U01). (16-May-2012)

Foreign:

Reviewer for Genome Canada's 2012 Bioinformatics and Computational Biology Competition (9-Dec-2012)

Ad hoc reviewer for Icelandic Research Fund (3-Nov-2015)

PUBLICATIONS

† denotes corresponding author

★ denotes joint first authors

★★ denotes consortium manuscript

2017

50. Eilbeck K, **Quinlan AR★**, Yandell M. Settling the score: variant prioritization and Mendelian disease. *Nature Reviews Genetics*, doi:10.1038/nrg.2017.52. 2017.
49. Pedersen BS, **Quinlan AR†**. Who's who? Detecting and resolving sample anomalies in human DNA sequencing studies with peddy. *AJHG*. DOI: 10.1016/j.ajhg.2017.01.017. 2017.
48. Pedersen BS, **Quinlan AR†**. cyvcf2: fast, flexible variant analysis with Python. *Bioinformatics*. DOI: 10.1093/bioinformatics/btx057. 2017.

2016

47. Pedersen BS, Layer RM, **Quinlan AR†**. Vcfanno: fast, flexible annotation of genetic variants. *Genome Biology*. 2016. doi: 10.1186/s13059-016-0973-5
46. Ge Y, Onengut-Gumuscu S, **Quinlan AR**, Mackey AJ, Wright JA, Buckner JH, Habib T, Rich SS, Concannon P. Targeted Deep Sequencing in Multiple-Affected Sibships of European Ancestry Identifies Rare Deleterious Variants in PTPN22 that Confer Risk for Type 1 Diabetes. *Diabetes*. 2016. pii: db150322
45. Layer RM, Kindlon N, Karczewski K, Exome Aggregation Consortium, **Quinlan AR†**. Efficient genotype compression and analysis of large genetic-variation data sets. *Nature Methods*. 2015. doi: 10.1038/nmeth.3654

2015

44. Layer R, **Quinlan AR†**. A parallel algorithm for N-way interval set intersection. *In press, IEEE Proceedings*.

43. Chiang C, Layer RM, Faust GG, Lindberg MR, Rose DB, Garrison EP, Marth GT, **Quinlan AR**, Hall IM. SpeedSeq: Ultra-fast personal genome analysis and interpretation *Nature Methods*. 2015. Oct;12(10):966-8. doi: 10.1038/nmeth.3505.
42. Auer PL, Nalls M, Meschia JF, Worrall BB, Longstreth WT Jr, Seshadri S, Kooperberg C, Burger KM, Carlson CS, Carty CL, Chen WM, Cupples LA, DeStefano AL, Fornage M, Hardy J, Hsu L, Jackson RD, Jarvik GP, Kim DS, Lakshminarayan K, Lange LA, Manichaikul A, **Quinlan AR**, Singleton AB, Thornton TA, Nickerson DA, Peters U, Rich SS; National Heart, Lung, and Blood Institute Exome Sequencing Project. Rare and Coding Region Genetic Variants Associated With Risk of Ischemic Stroke: The NHLBI Exome Sequence Project. *JAMA Neurol*. 2015. May 11. doi: 10.1001/jamaneurol.2015.0582.
41. Onengut-Gumuscu S, Chen WM, Burren O, Cooper NJ, **Quinlan AR**, et al. Fine mapping of type 1 diabetes susceptibility loci and evidence for colocalization of causal variants with lymphoid gene enhancers. *Nature Genetics*. 2015. Apr;47(4):381-6. doi: 10.1038/ng.3245
40. Lindberg MR, Hall IM, **Quinlan AR**†. Population-based structural variation discovery with Hydra-Multi. *Bioinformatics*. 2015. Apr 15;31(8):1286-9. doi: 10.1093/bioinformatics/btu771.
39. Church DM, Schneider VA, Steinberg KM, Schatz MC, **Quinlan AR**, Chin CS, Kitts PA, Aken B, Marth GT, Hoffman MM, Herrero J, Mendoza ML, Durbin R, Flicek P. Extending reference assembly models. *Genome Biol*. 2015. Jan 24;16:13. doi: 10.1186/s13059-015-0587-3.
38. Do R, Stitzel NO, Won HH, Jrgensen AB, Duga S, Angelica Merlini P, Kiezun A, Farrall M, Goel A, Zuk O, Guella I, Asselta R, Lange LA, Peloso GM, Auer PL; NHLBI Exome Sequencing Project, Girelli D, Martinelli N, Farlow DN, DePristo MA, Roberts R, Stewart AF, Saleheen D, Danesh J, Epstein SE, Sivapalaratnam S, Hovingh GK, Kastelein JJ, Samani NJ, Schunkert H, Erdmann J, Shah SH, Kraus WE, Davies R, Nikpay M, Johansen CT, Wang J, Hegele RA, Hechter E, Marz W, Kleber ME, Huang J, Johnson AD, Li M, Burke GL, Gross M, Liu Y, Assimes TL, Heiss G, Lange EM, Folsom AR, Taylor HA, Olivieri O, Hamsten A, Clarke R, Reilly DF, Yin W, Rivas MA, Donnelly P, Rossouw JE, Psaty BM, Herrington DM, Wilson JG, Rich SS, Bamshad MJ, Tracy RP, Cupples LA, Rader DJ, Reilly MP, Spertus JA, Cresci S, Hartiala J, Tang WH, Hazen SL, Allayee H, Reiner AP, Carlson CS, Kooperberg C, Jackson RD, Boerwinkle E, Lander ES, Schwartz SM, Siscovick DS, McPherson R, Tybjaerg-Hansen A, Abecasis GR, Watkins H, Nickerson DA, Ardissoni D, Sunyaev SR, O'Donnell CJ, Altshuler D, Gabriel S, Kathiresan S. *Nature*. 2015. Feb 5;518. doi: 10.1038/nature13917.

2014

37. Dai C, Deng Y, **Quinlan AR**, Gaskin F, Tsao B, Fu SM. Genetics of Systemic Lupus Erythematosus: Immune Responses and End Organ Resistance to Damage. *Current Opinion in Immunology*. doi:10.1016/j.coi.2014.10.004
36. Quick J, **Quinlan AR**, Loman N. A reference bacterial genome dataset generated on the MinION™ portable single-molecule nanopore sequencer. *GigaScience*. Oct 20;3:22. doi: 10.1186/2047-217X-3-22
35. Loman N, **Quinlan AR**†. PORETOOLS: a toolkit for working with nanopore sequencing data from Oxford Nanopore. *Bioinformatics*. doi:10.1093/bioinformatics/btu555, 2014.
34. Yi Qiao, **Quinlan AR**, Amir Jazaeri, Roeland Verhaak, David Wheeler, Gabor Marth. SubcloneSeeker: a computational framework for reconstructing tumor clone structure for

cancer variant interpretation and prioritization.

Genome Biology. Aug 26;15(8):443, 2014.

33. **Quinlan AR**†. BEDTools: the Swiss-army tool for genome interval arithmetic.
Curr Protoc Bioinformatics. doi: 10.1002/0471250953.bi1112s47, 2014.
32. Layer R, **Quinlan AR**†, Hall IM†. LUMPY: A probabilistic framework for sensitive detection of chromosomal rearrangements.
Genome Biology. doi:10.1186/gb-2014-15-6-r84, 2014.
31. Martin N, Nakamura K, Paila U, Woo J, Brown C, Wright J, Teraoka S, Haghayegh S, McCurdy D, Schneider M, Hu H, **Quinlan AR**, Gatti R, and Concannon P. Homozygous mutation of MTPAP causes cellular radiosensitivity and persistent DNA double strand breaks.
Cell Death Dis. doi: 10.1038/cddis.2014.99, 2014.
30. Farber CR, Reich A, Barnes AM, Becerra P, Rauch F, Cabral WA, Bae A, **Quinlan AR**, Glorieux FH, Clemens TL, and Marini JC. A Novel IFITM5 Mutation in Severe Osteogenesis Imperfecta Decreases PEDF Secretion by Osteoblasts.
J Bone Miner Res. doi: 10.1002/jbmr.2173, 2014.
29. Tabor HK, Auer PL, Jamal SM, Chong JX, Yu JH, Gordon AS, Graubert TA, O'Donnell CJ, Rich SS, Nickerson DA; **★NHLBI Exome Sequencing Project**, Bamshad MJ. Pathogenic variants for Mendelian and complex traits in exomes of 6,517 European and African Americans: implications for the return of incidental results..
Am J Hum Genet. doi: 10.1016/j.ajhg.2014.07.006, 2014.
28. Lange LA, Hu Y, Zhang H, Xue C, Schmidt EM, Tang ZZ, Bizon C, Lange EM, Smith JD, Turner EH, Jun G, Kang HM, Peloso G, Auer P, Li KP, Flannick J, Zhang J, Fuchsberger C, Gaulton K, Lindgren C, Locke A, Manning A, Sim X, Rivas MA, Holmen OL, Gottesman O, Lu Y, Ruderfer D, Stahl EA, Duan Q, Li Y, Durda P, Jiao S, Isaacs A, Hofman A, Bis JC, Correa A, Griswold ME, Jakobsdottir J, Smith AV, Schreiner PJ, Feitosa MF, Zhang Q, Huffman JE, Crosby J, Wassel CL, Do R, Franceschini N, Martin LW, Robinson JG, Assimes TL, Crosslin DR, Rosenthal EA, Tsai M, Rieder MJ, Farlow DN, Folsom AR, Lumley T, Fox ER, Carlson CS, Peters U, Jackson RD, van Duijn CM, Uitterlinden AG, Levy D, Rotter JI, Taylor HA, Gudnason V Jr, Siscovick DS, Fornage M, Borecki IB, Hayward C, Rudan I, Chen YE, Bottinger EP, Loos RJ, Strom P, Hveem K, Boehnke M, Groop L, McCarthy M, Meitinger T, Ballantyne CM, Gabriel SB, O'Donnell CJ, Post WS, North KE, Reiner AP, Boerwinkle E, Psaty BM, Altshuler D, Kathiresan S, Lin DY, Jarvik GP, Cupples LA, Kooperberg C, Wilson JG, Nickerson DA, Abecasis GR, Rich SS, Tracy RP, Willer CJ; **★NHLBI Grand Opportunity Exome Sequencing Project**. Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol.
Am J Hum Genet. doi: 10.1016/j.ajhg.2014.01.010, 2014.
27. Gordon AS, Tabor HK, Johnson AD, Snively BM, Assimes TL, Auer PL, Ioannidis JP, Peters U, Robinson JG, Sucheston LE, Wang D, Sotoodehnia N, Rotter JI, Psaty BM, Jackson RD, Herrington DM, O'Donnell CJ, Reiner AP, Rich SS, Rieder MJ, Bamshad MJ, Nickerson DA, **★NHLBI GO Exome Sequencing Project**.. Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol.
Quantifying rare, deleterious variation in 12 human cytochrome P450 drug-metabolism genes in a large-scale exome dataset. *Hum Mol Genet*, doi: 10.1093/hmg/ddt588, 2014.

2013

26. Paila U, Chapman BA, Kirchner R, **Quinlan AR**†. GEMINI: Integrative Exploration of Genetic Variation and Genome Annotations.
PLoS Comput Biol, 9(7): e1003153. doi:10.1371/journal.pcbi.1003153, 2013.

25. Rosenthal EA, Ranchalis J, Crosslin DR, Burt A, Brunzell JD, Motulsky AG, Nickerson DA; **★NHLBI GO Exome Sequencing Project**, Wijsman EM, Jarvik GP. Joint linkage and association analysis with exome sequence data implicates SLC25A40 in hypertriglyceridemia. *Am J Hum Genet.*, doi: 10.1016/j.ajhg.2013.10.019, 2013.
24. Guo DC, Regalado E, Casteel DE, Santos-Cortez RL, Gong L, Kim JJ, Dyack S, Horne SG, Chang G, Jondeau G, Boileau C, Coselli JS, Li Z, Leal SM, Shendure J, Rieder MJ, Bamshad MJ, Nickerson DA; GenTAC Registry Consortium; National Heart, Lung, and **★Blood Institute Grand Opportunity Exome Sequencing Project**, Kim C, Milewicz DM. Recurrent gain-of-function mutation in PRKG1 causes thoracic aortic aneurysms and acute aortic dissections. *Am J Hum Genet.*, doi: 10.1016/j.ajhg.2013.06.019, 2013.
23. O'Connor TD, Kiezun A, Bamshad M, Rich SS, Smith JD, Turner E; NHLBIGO Exome Sequencing Project; ESP Population Genetics, Statistical Analysis Working Group, Leal SM, Akey JM**★**. Fine-scale patterns of population stratification confound rare variant association tests. *PLoS One*, 8(7): e65834. doi:10.1371/journal.pone.0065834, 2013.
22. Johnsen JM, Auer PL, Morrison AC, Jiao S, Wei P, Haessler J, Fox K, McGee SR, Smith JD, Carlson CS, Smith N, Boerwinkle E, Kooperberg C, Nickerson DA, Rich SS, Green D, Peters U, Cushman M, Reiner AP; NHLBI Exome Sequencing Project.**★**. Common and rare von Willebrand factor (VWF) coding variants, VWF levels, and factor VIII levels in African Americans: the NHLBI Exome Sequencing Project. *Blood*, 122(4):590-7. doi:10.1182/blood-2013-02-485094, 2013.
21. Norton N, Li D, Rampersaud E, Morales A, Martin ER, Zuchner S, Guo S, Gonzalez M, Hedges DJ, Robertson PD, Krumm N, Nickerson DA, Hershberger RE; National Heart, Lung, and Blood Institute GO Exome Sequencing Project and the Exome Sequencing Project Family Studies Project Team.**★**. Exome sequencing and genome-wide linkage analysis in 17 families illustrate the complex contribution of TTN truncating variants to dilated cardiomyopathy. *Circ Cardiovasc Genet.*, 6(2):144-53. doi:10.1161/CIRCGENETICS.111.000062, 2013.
20. Malhotra A, Lindberg M, Leibowitz M, Clark R, Faust G, Layer R, **Quinlan AR**†, and Hall IM†. Breakpoint profiling of 64 cancer genomes reveals numerous complex rearrangements spawned by homology-independent mechanisms. *Genome Research*, doi:10.1101/gr.143677.112, 2013.
19. Fu W, O'Connor TD, Jun G, Kang HM, Abecasis G, Leal SM, Gabriel S, Rieder MJ, Altshuler D, Shendure J, Nickerson DA, Bamshad MJ; NHLBI Exome Sequencing Project, Akey JM. *Nature.*, 493(7431):216-20. doi:10.1038/nature11690, 2013.
18. Layer R, Robins G, Skadron K, **Quinlan AR**†. Binary Interval Search (BITS): A Scalable Algorithm for Counting Interval Intersections. *Bioinformatics*, 29(1):1-7, 2013.
17. Boileau C, Guo DC, Hanna N, Regalado ES, Detaint D, Gong L, Varret M, Prakash SK, Li AH, d'Indy H, Braverman AC, Grandchamp B, Kwartler CS, Gouya L, Santos-Cortez RL, Abifadel M, Leal SM, Muti C, Shendure J, Gross MS, Rieder MJ, Vahanian A, Nickerson DA, Michel JB; National Heart, Lung, and Blood Institute (NHLBI) Go Exome Sequencing Project, Jondeau G, Milewicz DM.**★**. TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. *Nature Genetics.*, 44(8):916-21. doi:10.1038/ng.2348, 2013.
16. Emond MJ, Louie T, Emerson J, Zhao W, Mathias RA, Knowles MR, Wright FA, Rieder MJ, Tabor HK, Nickerson DA, Barnes KC; National Heart, Lung, and Blood Institute (NHLBI) GO Exome Sequencing Project; Lung GO, Gibson RL, Bamshad MJ.**★**. Exome sequencing of extreme phenotypes identifies DCTN4 as a modifier of chronic *Pseudomonas aeruginosa*

infection in cystic fibrosis.
Nature Genetics., 44(8):886-9. doi:10.1038/ng.2344, 2013.

2010 - 2012

15. Krumm N, Sudmant PH, Ko A, O’Roak BJ, NHLBI Exome Sequencing Project, **Quinlan AR**, Nickerson DA, Eichler EE. Copy number variation detection and genotyping from exome sequence data.
Genome Research, 22(8):1525-32, 2012.
14. **Quinlan AR**, Hall IM. Characterizing complex structural variation in germline and somatic genomes.
Trends in Genetics, 18:43-53, 2012.
13. **Quinlan AR** and Hall IM. Detection and interpretation of genomic structural variation in mammals.
Methods in Molecular Biology, 838:225-48, 2012.
12. **Quinlan AR**, Boland MJ, Leibowitz ML, Shumilina S, Pehrson SM, Baldwin KK, Hall IM. Paired-end DNA sequencing of induced pluripotent stem cell genomes reveals rare structural mutations and retroelement stability.
Cell Stem Cell, 9:366-373, 2011.
11. Keene KL, **Quinlan AR**, Hou X, Hall IM, Mychaleckyj, Onengut-Gumuscu S, Concannon P. Evidence for two independent associations with type 1 diabetes at the 12q13 locus.
Genes and Immunity, 13:66-70, 2011.
10. Dale R, Pedersen B, **Quinlan AR**†. Pybedtools: a flexible Python library for manipulating genomic datasets and annotations.
Bioinformatics, 24:3423-3424, 2011.
packages.python.org/pybedtools/
9. Barnett D, Garrison E, **Quinlan AR**, Stromberg M, Marth G. BamTools: a C++ API and toolkit for analyzing and managing BAM files.
Bioinformatics, 12:1691-1692, 2011.
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8. 1000 Genomes Project Consortium.. A map of human genome variation from population-scale sequencing.
Nature 7319:1061-73, 2010.
7. **Quinlan AR** and Hall IM. BEDTools: A flexible framework for comparing genomic features.
Bioinformatics, 6:841-842, 2010.
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6. **Quinlan AR**, Clark RA, Sokolova, S, Leibowitz ML, Zhang Y, Hurles ME, Mell JC, Hall IM. Genome-wide mapping and assembly of structural variant breakpoints in the mouse genome.
Genome Research, 20:623-635, 2010.
code.google.com/p/hydra-sv

2007 - 2009

5. Sackton, TB, Kulathinal RJ, Bergman CM, Quinlan AR, Dopman E, Marth GT, Hartl DL, Clark AG. Population Genomic Inferences from Sparse High-Throughput Sequencing of Two Populations of *Drosophila melanogaster*.
Genome Biol Evol, 1:439-455, 2009.
4. Smith D, **Quinlan AR***, Peckham HR, *et al.* Rapid whole-genome mutational profiling using next-generation sequencing technologies.
Genome Research, 18:1638-1642, 2008.

3. Hillier LW, Marth GT, **Quinlan AR**, *et al.* Whole Genome Sequencing and SNP Discovery for *C. elegans* using massively parallel sequencing-by-synthesis. *Nature Methods*, 5:183-188, 2008.
2. **Quinlan AR**, Stewart D, Stromberg M, Marth GT. PyroBayes: Accurate quality scores for 454 Life Science pyrosequences. *Nature Methods*, 5:179-181, 2008.
1. **Quinlan AR**, Marth GT. Primer-site SNPs mask mutations. *Nature Methods*, 4:192, 2007.

ACTIVE RESEARCH
SUPPORT

Project Title: *New algorithms and tools for large-scale genomic analysis.*
 PI: Aaron Quinlan
 Source: NIH/NHGRI (R01 HG006693-05)
 Annual directs: \$331,000
 Period funded: 1-Apr-2016 - 30-Mar-2021

Project Title: *Monitoring tumor subclonal heterogeneity over time and space.*
 MPI: Gabor Marth (Contact) and Aaron Quinlan
 Source: NIH/NCI (UCA209999A)
 Annual directs: \$150,000
 Period funded: 1-Jul-2016 - 30-Jun-2021

Project Title: *Software for exploring all forms of genetic variation in any species.*
 PI: Aaron Quinlan
 Source: NIH/NHGMS (R01GM124355)
 Annual directs: \$476,000
 Period funded: 1-July-2017 - 30-Jun-2022
 Status: Pending IRG Review

Project Title: *The genetic basis of hypersensitivity to ionizing radiation*
 PI: Pat Concannon
 Source: NIH/NIEHS (R01 ES027121-01)
 Annual directs: \$55,000
 Period funded: 1-Oct-2016 - 30-Sep-2021

Project Title: *A scalable, integrative, multi-omic analysis platform*
 PI: Ryan Layer (Contact) and Aaron Quinlan
 Source: NIH/NHGRI (K99 HG009532-01)
 Annual directs: \$145,200
 Period funded: 1-July-2017 - 30-Jun-2021
 Status: Impact score = 13. Will very likely be funded in Feb 2017

Project Title: *Epigenetic engineering to identify and perturb gene regulatory regions involved in cancer etiology and therapy resistance.*
 MPI: Jay Gertz (Contact) and Aaron Quinlan
 Source: NIH/NHGRI (R01HG009141-01)
 Annual directs: \$85,000
 Period funded: 1-July-2017 - 30-Jun-2021

Status: 5th percentile. Will very likely be funded in April 2017

PENDING RESEARCH
SUPPORT

Project Title: *A powerful web-based discovery platform for rare disease genomics.*
MPI: Daniel MacArthur (Contact) and Aaron Quinlan
Source: NIH/NHGRI (R01HG009141-01)
Annual directs: \$249,700
Period funded: 1-July-2017 - 30-Jun-2021
Status: 4th percentile. Will very likely be funded in April 2017

Project Title: *Epigenetic engineering to identify and perturb gene regulatory regions involved in cancer etiology and therapy resistance.*
MPI: Jay Gertz (Contact) and Aaron Quinlan
Source: NIH/NHGRI (1U01CA217884-01)
Annual directs: \$65,000
Period funded: 1-July-2017 - 30-Jun-2021
Status: Pending IRG Review

COMPLETED
RESEARCH SUPPORT

Project Title: *The genetic basis of simplex autism*
PI: Hilary Coon
Source: Margolis Foundation
Annual directs: \$24,000
Period funded: 1-Mar-2016 - 30-June-2017

Project Title: *The genetic basis of simplex autism*
PI: Hilary Coon
Source: Simons Foundation
Annual directs: \$24,000
Period funded: 1-Mar-2016 - 31-Dec-2016

Project Title: *A clinical sequencing program to direct treatment of relapsed pediatric cancers.*
PI: Ira Hall and Aaron Quinlan
Source: UVA Health System Research Award
Period funded: 31-Apr-2013 - 30-Mar-2016

Project Title: *New oncogenes and regions of genome instability in ovarian cancer.*
PI: Aaron Quinlan
Source: University of Virginia Fund for Excellence in Science and Technology (FEST)
Period funded: 01-May-2011 - 30-Apr-2014

Project Title: *Defining the genomic architecture of glioblastoma for improved therapy.*
PI: Aaron Quinlan
Source: University of Virginia Cancer Center Pilot Fund
Period funded: 01-Apr-2011 - 31-Dec-2013

Project Title: *The Role of Copy Number Variants in Type 1 Diabetes.*
PI: Stephen Rich
Source: NIH/NIDDK (DP3 DK085695)
Period funded: 30-Sep-2009 - 30-Jun-2014

Project Title: *Expression and proteomic characterization of risk loci in type 1 diabetes.*

PI: Stephen Rich
Source: NIH/NIDDK (DP3 DK085678)
Period funded: 25-Sep-2009 - 30-Jun-2014

Project Title: *Identification of radiation sensitivity alleles by whole exome sequencing.*
PI: Pat Concannon
Co-investigator: Aaron Quinlan
Source: NIH/NIEHS (R21 ES020521-01)
Period funded: 19-Aug-2011 - 31-Jul-2013

Project Title: *Carry Out Physical Characterization of Contest Samples and Development and Test Bioinformatic Methods for Scoring and Judging Contestant Entries.*
PI: Dean Gaalaas
Source:
Period Archon Genomics X Prize : 27-Mar-2012 - 31-Dec-2012

Project Title: *Rates and patterns of recurrent structural variation in the mouse genome.*
PI: Aaron Quinlan
Source: NIH/NHGRI (F32 HG005197-02)
Period funded: 01-Aug-2009 - 31-Dec-2010

LECTURES

† *invited lecture*
★ *abstract selected lecture*

Inferring function from highly constrained coding regions.
UW Center for Mendelian Genomics; Seattle, WA;
Aug 17, 2017

★ *Inferring function from highly constrained coding regions.*
BIRS Conference; Banff, Alberta;
Mar 27, 2017

Variation deserts and recombination jungles.
BYU; Provo, UT;
Feb 23, 2017

Variation deserts and recombination jungles.
UCSD (Yeo Lab); San Diego, CA;
Dec 6, 2016

★ *Direct measurement of the mutagenic impact of recombination through deep genome sequencing of 519 families.*
ASHG 2016; Vancouver, BC;
Oct 22, 2016

★ *Genetic analysis software for any species.*
PAG 2016; San Diego, CA;
Jan 9, 2016

† *Making queries of the genome less difficult.*
BIOT 2015; Provo, UT;
Dec 11, 2015

† *Making queries of the genome less difficult.*

Genome Informatics 2015; Cold Spring Harbor, NY;
Oct 30, 2015

† *Querying the genome.*
Sanger Institute Seminar Series; Hinxton, England, UK;
Oct 24, 2015

Challenges and (some) solutions for scanning the genome in studies of human disease.
University of Utah Human Genetics Interest Group; Salt Lake City, UT;
Sep 24, 2015

† *Variant calling while accounting for alternate haplotypes*
Genome Reference Consortium Workshop 2014; Cambridge, England;
Sep 21, 2014

★ *How does ovarian cancer become resistant to chemotherapy?*
Biology of the Genome 2014; Cold Spring Harbor, NY
May 6-10, 2014

† *Prioritizing germline and somatic variation in studies of human disease.*
Johns Hopkins School of Medicine.
April 28, 2014

† *Comprehensive discovery and prioritization of genetic variation in studies of human disease.*
University of Virginia Biomedical Engineering Seminar Series.
April 4, 2014

† *Algorithms for chromosomal rearrangement detection and DNA classification.*
Cold Spring Harbor Laboratories Quantitative Biology Seminar Series.
March 19, 2014

† *Comprehensive discovery and prioritization of genetic variation in studies of human disease.*
University of Florida Genetics Institute.
March 12, 2014

† *Comprehensive discovery and prioritization of genetic variation in studies of human disease.*
University of Utah Department of Human Genetics.
February 26, 2014

★ *Disease variant interpretation and prioritization with GEMINI.*
Genome Informatics 2013; Cold Spring Harbor, NY
October 30, 2013

★ *Mining genomic feature sets and identifying significant biological relationships with BedTools2.*
American Society of Human Genetics; Boston, MA
October 22, 2013

★ *Disease variant interpretation and prioritization with GEMINI.*
Beyond the Genome 2013; San Francisco, CA
October 3, 2013

† *Detection and characterization of complex rearrangements in tumor genomes.*
BioConductor 2013; Seattle, WA

July 18, 2013

★ *Exploring disease genetics among thousands of human genomes with GEMINI.*

SciPy 2013; Austin, TX

June 26, 2013

Computational Genomics.

Big Data Summit 2 at UVa; Charlottesville, VA

May 14, 2013

Exploring genetic variation with a tour guide.

International Stroke Genetics Consortium Meeting; Charlottesville, VA

April 25, 2013

★ *LUMPY: A probabilistic framework for SV discovery.*

Advances in Genome Biology and Technology (AGBT); Marco Island, FL

February 22, 2013

Mining the genome.

UVa. Center for Public Health Genomics Genome Sciences Seminar Series

November 28, 2012

† *Mining the structure and function of the genome.*

Penn State, Dept. of Biochemistry and Molecular Biology

November 12, 2012; Host: Anton Nekreutenko

Exploring high-dimension genomic data.

Cold Spring Harbor Laboratories Advanced Sequencing Technologies Course

October 22, 2012

Towards a map of structural variation in the Exome Sequencing Project.

NHLBI Exome Sequencing Project In-Person Meeting

March 28, 2012

† *Exploring the origin and extent of structural variation in human genomes.*

Deans New Faculty Seminar Series, University of Virginia School of Medicine

Jan 19, 2012

ESP Structural Variation Project Group: goals, initial results, and future work.

NHLBI Exome Sequencing Project In-Person Meeting

June 9, 2011

★ *Large-Scale Characterization of SV Breakpoints in Cancer.*

Keystone Symposium on The Functional Impact of Structural Variation

Jan. 11, 2011

★ *Efficient discovery of structural instability in repetitive regions of mammalian genomes.*

Advances in Genome Biology and Technology

Feb. 2009

Approaches to rare allele discovery: More samples or more depth per sample?

1000 Genomes Analysis Meeting, Cold Spring Harbor Laboratories

May 2008.

ACADEMIC SERVICE

Health Sciences Research Council, University of Utah.
2017 - 2019.

Utah Genome Project Ambassador Program, University of Utah.
2017 - 2019.

Molecular Biology Admissions Committee, University of Utah.
2015 - 2017.

Judge for BIMS Graduate Student Poster Session.
19-Apr-2013.

Data Management Committee. Organized by Rick Horwitz, VPR.
12-Apr-2013.

Big Data Analytics Committee. Organized by Don Brown, Systems Engineering.
29-Mar-2013.

Member of Center for Public Health Genomics Executive Committee.
01-Aug-2012 - Present.

Member of the Univ. of Virginia Bioinformatics Core Advisory Committee.
01-Nov-2011 - Present.

Served on the Univ. of Virginia Bioinformatics Core Director Search Committee.
Summer 2011.

TEACHING

Applied Computational Genomics. University of Utah. Spring 2017.

Applied Genomics II (PH TX 7778-001). University of Utah, Spring 2016.

Evolutionary Genetics and Genomics (HGEN 6092). University of Utah, Spring 2016.

Advanced Genomics Journal Club (BMI 7010-003/HGEN 6810-001). University of Utah, Spring 2016.

Faculty for CSHL Advanced Sequencing Technologies Course, 2009 - 2017
- <http://meetings.cshl.edu/courses/2013/c-seqtech13.shtml>

Faculty for the University of Washington's Center for Mendelian Genetics Workshop. 2014 - 2017

Faculty for Canadian Bioinformatics Workshops. 2012, 2013, 2014
- <http://bioinformatics.ca/workshops/faculty>

Guest Lecturer for first year Computer Science graduate student core course (CS 6190), Fall 2013.
Taught by Kevin Skadron.

Lecturer for first year graduate student core course (BIMS 6000), Fall 2013
- *Genomics Lecture and practical session* - 04-Sep-2013
- *Research article discussions* - 5-Sep-2013

Guest Lecturer for undergraduate and graduate Biomedical Engineering course (BME 4806, 7806;

Prof. Brent French),
April 8, 2013
April 14, 2014

Guest Lecturer for graduate course in genomics (BIOCH 5080), Spring 2013.
March 20, March 22, April 29

Lecturer for first year medical student curriculum, Fall 2012.
- *Genomics research article discussions* - 09-Oct-2012

Lecturer for first year graduate student core course (BIMS 6000), Fall 2012
- *Genomics Lecture and practical session* - 07-Sep-2012
- *Research article discussions* - 10-Sep-2012, 11-Sep-2012

Guest Lecturer for graduate course in genomics (BIOCH 5080), 15-Feb-2011

MENTORSHIP

Current

James Havrilla (Ph.D. candidate, U. of Utah, Human Genetics, started January 2014)
Thomas Sasani (Ph.D. candidate, U. of Utah, Human Genetics, started April 2016)
Stephanie Kravitz (Ph.D. candidate, U. of Utah, Human Genetics, started April 2017)
Jonathan Belyeu (Ph.D. candidate, U. of Utah, Human Genetics, started April 2017)

Former

John Kubinski (Undergraduate Biology Senior Thesis candidate)
Ryan Layer (Ph.D. candidate, Computer Science, graduated 2014)
Phanwadee Sinthong (Undergraduate Computer Science researcher)

THESIS COMMITTEES

Current

Edwin Lin (MD/Ph.D. candidate, U. of Utah, Human Genetics)
Nicole Russell (MD/Ph.D. candidate, U. of Utah, Human Genetics)
Kristi Russell (Ph.D. candidate, U. of Utah, Human Genetics)
Julia Carleton (Ph.D. candidate, U. of Utah, Oncological Sciences)
Eric Bogenschutz (Ph.D. candidate, U. of Utah, Human Genetics)
Rachel Cosby (Ph.D. candidate, U. of Utah, Human Genetics)
Cassandra Garner (Ph.D. candidate, U. of Utah, Human Genetics)
Samuel Brady (M.S. candidate, U. of Utah, Biomedical Informatics)
Paris Vail (M.S. candidate, U. of Utah, Biomedical Informatics)

Former

Michael Lindberg (Ph.D. candidate, Biochemistry and Molecular Genetics, qualified 2012)
Lauren Mills (Ph.D., candidate Biochemistry and Molecular Genetics)
Johnny Gan (Ph.D., candidate Systems Engineering)