

## Aaron R. Quinlan, Ph.D.

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CONTACT INFORMATION	Professor Department of Human Genetics Department of Biomedical Informatics Thomas Dee Endowed Chair of Human Genetics Associate Director of the Utah Center for Genetic Discovery University of Utah	(O): 801.585.0406 (W): <a href="mailto:quinlanlab.org">quinlanlab.org</a> <a href="mailto:aquinlan@genetics.utah.edu">aquinlan@genetics.utah.edu</a>
RESEARCH INTERESTS	Mechanisms and consequences of genome mutation; Software and algorithms for genome research; Genetics of rare disease; Genetics of male infertility; Cancer genomics and targeted therapies; Purifying selection in the human genome.	
EDUCATION	<b>Boston College</b> , Chestnut Hill, MA, USA Ph.D., Biology, 2008  <b>College of William and Mary</b> , Williamsburg, VA, USA B.S., Computer Science, 1997	
EXPERIENCE	<b>University of Utah</b> , Salt Lake City, UT, USA <i>Professor of Human Genetics and Biomedical Informatics</i>	2019-Present
	<b>Base2 Genomics</b> , Salt Lake City, UT, USA <i>Co-Founder</i>	2017-2020
	<b>University of Utah</b> , Salt Lake City, UT, USA <i>Associate Professor of Human Genetics and Biomedical Informatics</i>	2015-2019
	<b>University of Virginia</b> , Charlottesville, VA, USA <i>Assistant Professor of Public Health Sciences</i>	2011-2015
	<b>University of Virginia</b> , Charlottesville, VA, USA <i>NRSA Postdoctoral Fellow (NHGRI)</i>	2008-2011
SERVICE, HONORS, AWARDS	<b>Current</b> Thomas Dee Endowed Chair of Human Genetics (2021-Present) Scientific Advisory Board for the NHGRI GREGoR Consortium (Sept 2021 - Present) SAB for the Utah Genome Project at the University of Utah (2018-Present) Human Genetic Faculty Search Committee (2021) Huntsman Cancer Institute Faculty Search Committee (2021) Editorial Board for Genome Research (2019-Present). Editorial Board for Oxford Bioinformatics (2016-Present). Executive Board for the Center for Genomic Medicine at U. of Utah (2018-Present)  <b>Past</b> Research Council for the University of Utah (2018-2020) SAB for European Bioinformatics Institute (2017-Present). Guest Member of the Editorial Board for Annual Reviews Genetics (April, 2020).	

Editorial Board for PeerJ (2017- ).

Organizing Committee for the CSHL/Wellcome Trust Genome Informatics meeting (2016-2018).

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

*Ad hoc* reviewer for:  
*Nature*, *Nature Genetics*, *Nature Methods*,  
*Nature Biotechnology*, *AJHG*,  
*Genome Research*, *Genome Biology*  
*Bioinformatics*, *BMC Bioinformatics*,  
*Bioessays*, *Genes*, and  
*IEEE Transactions On Computational Biology and Bioinformatics*

Guest editor for *PLoS Computational Biology*

#### GRANT REVIEW

**NIH:**  
 NIH Special Emphasis Panel, Oct, 2021  
 NIH BDMA Study Section, Feb, 2021  
 NCI ITCR Set-aside Funds Review, Oct 11, 2020  
 NIH Special Emphasis Panel (ZMH1-ERB-C-09) June 24, 2020  
 NIH GCAT Study Section, Feb 20, 2019  
 Co-chair of the American Heart Association Uncovering New Patterns Study Section, (Feb and March, 2018)  
 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)

#### TEACHING

Applied Computational Genomics. Full semester course. University of Utah. Spring 2022.  
 Applied Computational Genomics. Full semester course. University of Utah. Spring 2021.

Applied Computational Genomics. Full semester course. University of Utah. Spring 2020.  
 Salt Lake Learners of Biostatistics. Interest Group. University of Utah. Starting May, 2019.  
 Applied Computational Genomics. Full semester course. University of Utah. Spring 2018.  
 Applied Computational Genomics. Full semester course. University of Utah. Spring 2017.  
 Faculty for CSHL Advanced Sequencing Technologies Course, 2009 - Present  
 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.

- <http://meetings.cshl.edu/courses/2013/c-seqtech13.shtml>

Faculty for the University of Washington's Center for Mendelian Genetics Workshop. 2014 - 2018

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

## PUBLICATIONS

† *denotes corresponding author*

★ *denotes joint first authors*

★★ *denotes consortium manuscript*

Google Scholar Profile: <https://scholar.google.com/citations?user=QwVZZKsAAAAJ&hl=en>

## Preprints

86. Ryan M. Layer, Fritz J. Sedlazeck, Brent S. Pedersen, **Quinlan AR**. Mining Thousands of Genomes to Classify Somatic and Pathogenic Structural Variants. *Biorxiv*.

85. Peter McHale, **Quinlan AR**†. trfermikit: a tool to discover VNTR-associated deletions. *Biorxiv*.

84. Goldstein SA, Brown J, Pedersen BS, **Quinlan AR**, Elde NC. Extensive recombination-driven coronavirus diversification expands the pool of potential pandemic pathogens. *Biorxiv*.

83. Fixsen, Sarah M; Cone, Kelsey R; Goldstein, Stephen A; Sasani, Thomas A; **Quinlan AR**; Rothenburg, Stefan; Elde, Nels C. Poxviruses capture host genes by LINE-1 retrotransposition. *Biorxiv*.
82. Hou, Hao; Pedersen, Brent S; **Quinlan AR**†. Efficient storage and analysis of quantitative genomics data with the Dense Depth Data Dump (D4) format and d4tools. *Biorxiv*.

## 2021

81. Pedersen, Brent S; Brown, Joseph M; Dashnow, Harriet; Wallace, Amelia D; Velinder, Matt; Tvrdik, Tatiana; Mao, Rong; Best, Hunter D; Bayrak-Toydemir, Pinar; **Quinlan AR**†. Effective variant filtering and expected candidate variant yield in studies of rare human disease. *Nature Genomic Medicine*.
80. Meenal Gupta, Xiangfei Liu, Sharon N. Teraoka, Jocyndra A. Wright, Richard A. Gatti, **Quinlan AR**, and Patrick Concannon2. Genes affecting ionizing radiation survival identified through combined exome sequencing and functional screening. *Human Mutation*.
79. Jonathan R. Belyeu, Thomas A. Sasani, Brent S. Pedersen, **Quinlan AR**†. Unfazed: parent-of-origin detection for large and small de novo variants. *Bioinformatics*.
78. Wallace, Amelia; Sasani, Thomas; Swanier, Jordan; Gates, Brooke; Greenland, Jeffery; Pedersen, Brent; Varley, Katherine; **Quinlan AR**†. CaBagE: a Cas9-based Background Elimination strategy for targeted, long-read DNA sequencing. *PLoS One*. 2020
77. Nicholas, Thomas J; Cormier, Michael J; Huang, Xiaomeng; Qiao, Yi; Marth, Gabor T; **Quinlan AR**†. OncoGEMINI: Software for Investigating Tumor Variants From Multiple Biopsies With Integrated Cancer annotations. *Genome Medicine*. 2020
76. Cormier, Michael; Belyeu, Jonathan; Pedersen, Brent S; Brown, Joseph; Koster, Johannes; **Quinlan AR**†. Go Get Data (GGD): simple, reproducible access to scientific data. *Nature Communications*.
75. Belyeu JR, Brand H, Wang H, Zhao X, Pedersen BS, Feusier J, Gupta M, Nicholas TJ, Brown J, Baird L, Devlin B, Sanders SJ, Jorde LB, Talkowski ME, **Quinlan AR**†. De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. *AJHG*. 2020
74. Belyeu, Jonathan R; Chowdhury, Murad; Brown, Joseph; Pedersen, Brent S; Cormier, Michael J; **Quinlan AR**; Layer, Ryan M; . Samplot: A platform for structural variant visual validation and automated filtering *Genome Biology*. 2020

## 2020

73. PCarleton JB, Ginley-Hidinger M, Berrett KC, Layer RM, **Quinlan AR**, Gertz J. Regulatory sharing between estrogen receptor bound enhancers. *Nucleic Acids Research*. 2020
72. Pedersen BS, Bhetariya PJ, Brown J, Marth GT, Jensen RL, Bronner MP, Underhill HR, **Quinlan AR**†. Somalier: rapid relatedness estimation for cancer and germline studies using efficient genome sketches. *Genome Medicine*. 2020
71. Cawthon RM, Meeks HD, Sasani TA, Smith KR, Kerber RA, O'Brien E, Baird L, Dixon MM, Peiffer AP, Leppert MF, **Quinlan AR**, Jorde LB. Germline mutation rates in young adults predict longevity and reproductive lifespan. *Scientific Reports*. 2020

70. Berg, Jordan A; Belyeu, Jonathan R; Morgan, Jeffrey T; Ouyang, Yeyun; Bott, Alex J; **Quinlan AR**; Gertz, Jason; Rutter, Jared; . XPRESSyourself: Enhancing, standardizing, and automating ribosome profiling computational analyses yields improved insight into data *PLoS computational biology*. 2020

## 2019

69. Sasani TA, Pedersen BS, Gao Z, Baird L, Przeworski M, Jorde LB, **Quinlan AR**†. Large, three-generation CEPH families reveal post-zygotic mosaicism and variability in germline mutation accumulation. *eLife*. 2019
68. Pedersen BS, **Quinlan AR**†. Duphold: scalable, depth-based annotation and curation of high-confidence structural variant calls. *GigaScience*. 2019
67. Gao Z, Moorjani P, Sasani TA, Pedersen BS, **Quinlan AR**, Jorde JB, Przeworski M. Overlooked roles of DNA damage and maternal age in generating human germline mutations. *PNAS*. 2019
66. Boukas L, Havrilla JM, Hickey PF, **Quinlan AR**, Bjornsson HT, Hansen KT. Coexpression patterns define epigenetic regulators associated with neurological dysfunction. *Genome Research*. 2019
65. Havrilla JM, Pedersen BS, Layer RM, **Quinlan AR**†. A map of constrained coding regions in the human genome. *Nature Genetics*. 2019

## 2018

64. An JY, Lin K, Zhu L, Werling DM, Dong S, Brand H, Wang HZ, Zhao X, Schwartz GB, Collins RL, Currall BB, Dastmalchi C, Dea J, Duhn C, Gilson MC, Klei L, Liang L, Markenscoff-Papadimitriou E, Pochareddy S, Ahituv N, Buxbaum JD, Coon H, Daly MJ, Kim YS, Marth GT, Neale BM, **Quinlan AR**, Rubenstein JL, Sestan N, State MW, Willsey AJ, Talkowski ME, Devlin B, Roeder K, Sanders SJ. Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. *Science*. 2018
63. Westra HJ, Martnez-Bonet M, Onengut-Gumuscu S, Lee A, Luo Y, Teslovich N, Worthington J, Martin J, Huizinga T, Klareskog L, Rantapaa-Dahlqvist S, Chen WM, **Quinlan AR**, Todd JA, Eyre S, Nigrovic PA, Gregersen PK, Rich SS, Raychaudhuri S. Fine-mapping and functional studies highlight potential causal variants for rheumatoid arthritis and type 1 diabetes. *Nature Genetics*. 2018
62. Betsy E.P. Ostrander, Russell J. Butterfield, Brent S. Pedersen, Andrew J. Farrell, Ryan M. Layer, Alistair Ward, Chase Miller, Tonya DiSera, Francis M. Filloux, Meghan S. Candee, Tara Newcomb, Joshua L. Bonkowsky, Gabor T. Marth, **Quinlan AR**†. Whole-genome analysis for effective clinical diagnosis and gene discovery in early infantile epileptic encephalopathy. *Nature Genomic Medicine*. 2018
61. Thomas A. Sasani, Kelsey R. Cone, **Quinlan AR**†, Nels C. Elde. Long read sequencing reveals poxvirus evolution through rapid homogenization of gene arrays. *eLife*. 2018
60. Simovski B, Kanduri C, Gundersen S, Titov D, Domanska D, Bock C, Bossini-Castillo L, Chikina M, Favorov A, Layer RM, Mironov AA, **Quinlan AR**, Sheffield NC, Trynka G, Sandve GK. Coloc-stats: a unified web interface to perform colocalization analysis of genomic features. *Nucleic Acids Research*. 2018
59. Brent S Pedersen and **Quinlan AR**†, hts-nim: scripting high-performance genomic analyses. *Bioinformatics*. 2018

58. Jonathan R Belyeu, Thomas J Nicholas, Brent S Pedersen, Thomas A Sasani, James M Havrilla, Stephanie N Kravitz, Megan E Conway, Brian K Lohman, **Quinlan AR**<sup>†</sup>, Ryan M Layer. SV-plaudit: A cloud-based framework for manually curating thousands of structural variants. *Gigascience*. 2018
57. Ronna M Werling, Harrison Brand, Joon-Yong An, Matthew R Stone, Joseph T Glessner, Lingxue Zhu, Ryan L Collins, Shan Dong, Ryan M Layer, Eiriene-Chloe Markenscoff-Papadimitriou, Andrew Farrell, Grace B Schwartz, Benjamin B Currall, Jeanselle Dea, Clif Duhn, Carolyn Erdman, Michael Gilson, Robert E Handsaker, Seva Kashin, Lambertus Klei, Jeffrey D Mandell, Tomasz J Nowakowski, Yuwen Liu, Sirisha Pochareddy, Louw Smith, Michael F Walker, Harold Z Wang, Mathew J Waterman, Xin He, Arnold R Kriegstein, John L Rubenstein, Nenad Sestan, Steven A McCarroll, Ben M Neale, Hilary Coon, A. Jeremy Willsey, Joseph D Buxbaum, Mark J Daly, Matthew W State, Aaron Quinlan, Gabor T Marth, Kathryn Roeder, Bernie Devlin, Michael E Talkowski, Stephan J Sanders. An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. *Nature Genetics*. 2018
56. Miten Jain, Sergey Koren, Josh Quick, Arthur C Rand, Thomas A Sasani, John R Tyson, Andrew D Beggs, Alexander T Dilthey, Ian T Fiddes, Sunir Malla, Hannah Marriott, Karen H Miga, Tom Nieto, Justin O'Grady, Hugh E Olsen, Brent S Pedersen, Arang Rhie, Hollan Richardson, **Aaron Quinlan**, Terrance P Snutch, Louise Tee, Benedict Paten, Adam M. Phillippy, Jared T Simpson, Nicholas James Loman, Loose. Nanopore sequencing and assembly of a human genome with ultra-long reads. *Nature Biotechnology*. 2017
55. Ryan M. Layer, Brent S. Pedersen, Tonya DiSera, Gabor T. Marth, Jason Gertz, **Quinlan AR**<sup>†</sup>. GIGGLE: a search engine for large-scale integrated genome analysis. *Nature Methods*. 2017.
54. Pedersen BS, **Quinlan AR**<sup>†</sup>. mosdepth: quick coverage calculation for genomes and exomes. *Bioinformatics*. 2017.

## 2017

53. Andrea Bild, Samuel Brady, Jasmine McQuerry, Yi Qiao, Stephen Piccolo, Gajendra Shrestha, Ryan Layer, Brent Pedersen, David Jenkins, Ryan Miller, Amanda Esch, Sara Selitsky, Joel Parker, Layla Anderson, Chakravarthy Reddy, Jonathan Boltax, Dean Li, Philip Moos, Joe Gray, Laura Heiser, W. Evan Johnson, Saurada Buys, Adam Cohen, **Quinlan AR**, Gabor Marth, Theresa Werner, Brian Dalley, and Rachel Factor. Combating subclonal evolution of resistant cancer phenotypes. *Nature Communications*. 2017.
52. Pedersen BS, **Quinlan AR**<sup>†</sup>. Indexcov: fast coverage quality control for whole-genome sequencing: fast, flexible variant analysis with Python. *GigaScience*, *in press*. 2017.
51. Xiangfei Liu, Uma Devi Paila, Sharon N. Teraoka, Jocyndra A. Wright, Xin Huang, **Quinlan AR**, Richard A. Gatti and Patrick Concannon. Identification of ATIC as a novel target for chemoradiosensitization. *International Journal of Radiation Oncology*, *in press*. 2017.
50. Eilbeck K, **Quinlan AR**<sup>★</sup>, 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**<sup>†</sup>. 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**<sup>†</sup>. cyvcf2: fast, flexible variant analysis with Python. *Bioinformatics*. DOI: 10.1093/bioinformatics/btx057. 2017.

## 2016

47. Pedersen BS, Layer RM, **Quinlan AR**<sup>†</sup>. 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**<sup>†</sup>. 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**<sup>†</sup>. 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**<sup>†</sup>. 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, Ardisson 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.
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18. Layer R, Robins G, Skadron K, **Quinlan AR**<sup>†</sup>. Binary Interval Search (BITS): A Scalable Algorithm for Counting Interval Intersections. *Bioinformatics*, 29(1):1-7, 2013.
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## 2010 - 2012

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13. **Quinlan AR** and Hall IM. Detection and interpretation of genomic structural variation in mammals. *Methods in Molecular Biology*, 838:225-48, 2012.
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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.  
[code.google.com/p/bedtools](http://code.google.com/p/bedtools)
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*Genome Research*, 20:623-635, 2010.  
[code.google.com/p/hydra-sv](http://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.

## PENDING RESEARCH FUNDING

Project Title: *New algorithms and tools for large-scale genomic analyses.*

PIs: Aaron Quinlan

Source: NIH/NHGRI (R01 HG012252)

Annual directs: \$485,106

Period funded: 1-Sep-2021 - 31-Aug-2026 Status: 1st percentile, council review completed, funding imminent.

## ACTIVE RESEARCH FUNDING

Project Title: *Scalable detection and interpretation of structural variation in human genomes.*

PI: Aaron Quinlan

Source: NIH/NHGRI (R01 HG010757)

Annual directs: \$472,551

Period funded: 1-Apr-2020 - 30-Mar-2025

Project Title: *GEMS: Genomic approach to connecting Elevated germline Mutation rates with male infertility and Somatic health.*

MPIs: James Hotelling, Kenneth Aston, Aaron Quinlan

Source: NIH/NICHD (R01 HD106112)

Annual directs: \$609,608

Period funded: 1-Sep-2021 - 31-Aug-2026

Project Title: *A data and software ecosystem for bedtools.*

PI: Aaron Quinlan

Source: Chan Zuckerberg Institute  
Annual directs: \$100,000  
Period funded: 1-Aug-2021 - 31-Jul-2023

Project Title: *Software for exploring all forms of genetic variation in any species.*  
PI: Aaron Quinlan  
Source: NIH/NHGMS (R01GM124355)  
Annual directs: \$300,000  
Period funded: 1-July-2017 - 30-Jun-2022

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-2022

COMPLETED  
RESEARCH SUPPORT

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: *Interaction of WGS Variation and Polygenic Risk.*  
MPI: Hilary Coon, Anna Docherty, Gabor Marth, and Aaron Quinlan  
Source: Simons Foundation  
Annual directs: \$250,000  
Period funded: 1-Sep-2017 - 30-Aug-2021

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: *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

Project Title: *A data and software ecosystem for bedtools.*  
PI: Aaron Quinlan  
Source: Chan Zuckerberg Institute  
Annual directs: \$100,000  
Period funded: 1-May-2019 - 30-Apr-2020

Project Title: *A scalable, integrative, multi-omic analysis platform*  
PI: Ryan Layer (Contact)  
Source: NIH/NHGRI (K99 HG009532-01)  
Annual directs: \$145,200  
Period funded: 1-July-2017 - 30-Jun-2021

Project Title: *New algorithms and tools for large-scale genomic analysis.*  
PI: Aaron Quinlan  
Source: NIH/NHGRI (R01 HG006693-05)  
Annual directs: \$329,604  
Period funded: 1-Apr-2016 - 30-Mar-2019

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*

† *Patterns and consequences of mutation in the human germline*  
Medical University of South Carolina; (via Webex)  
April, 2021

† *Patterns and consequences of mutation in the human germline*  
UCONN Health; (via Webex)  
December 3, 2020

† *Strategies for diagnosing rare disease*  
2nd year medical students at UU; (via Webex)  
November 30, 2020; (via Webex)

† *Germline mutation rates in young adults predict longevity*  
EMGS 2020; (via Webex)

† *Annotation and curation of high-confidence structural variation*  
ESHG 2020; (via Webex)

† *We are all mutants: patterns of mutation in the human genome revealed by DNA sequencing of large, multigenerational families*  
Western Colorado University; Gunnison, CO;  
April 22, 2019

*Constrained coding regions and implications for somatic genome evolution*  
Huntsman Cancer Institute; Salt Lake City, UT;  
April 2, 2019

† *Large, three-generation CEPH families reveal post-zygotic mosaicism and variability in germline mutation accumulation*  
University of Michigan; Ann Arbor, MI;  
March 3, 2019

† *Tools for undiagnosed disease research*  
NIH; Bethesda, MD;  
January 30, 2019

† *Constrained coding regions and patterns of mutation in the human genome.*  
Penn State University; State College, PA;  
October 3, 2018

*Mutation in the human genome.*  
University of Utah, Dept. of Human Genetics; Salt Lake City, UT;  
September 14, 2018

† *A map of constrained coding regions in the human genome.*  
23AndMe; Mountain View, CA;

May 17, 2018

† *Computing the human genome.*  
Goldman Sachs; Salt Lake City, UT;  
April 2, 2018

† *A map of constrained coding regions in the human genome.*  
UCLA; Los Angeles, CA;  
March 12, 2018

*Computing the genome.*  
Chan Zuckerberg Initiative; Palo Alto, CA;  
Feb 14, 2018

*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    Utah Genome Project Advisory Board.  
2018 - Present

Center for Genomic Medicine Steering Committee.  
2017 - Present.

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

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.

#### MENTORSHIP

##### **Current**

Stephanie Kravitz (Ph.D. candidate, U. of Utah, Human Genetics, started April 2017)  
Michael Cormier (Ph.D. candidate, U. of Utah, Human Genetics, starting April 2018)  
Simone Longo (Ph.D. candidate, U. of Utah, Biomedical Informatics, started August 2019)  
Jason Kunisaki (MD/Ph.D. candidate, U. of Utah, Human Genetics, started May 2020)  
Laurel Hiatt (MD/Ph.D. candidate, U. of Utah, Human Genetics, started May 2021)  
John Chamberlin (Ph.D. candidate, U. of Utah, Human Genetics, started May 2019)  
Amelia Wallace (Postdoc, T32 Fellow)  
Harriet Dashnow (Postdoc)

##### **Former**

Jonathan Belyeu (Ph.D. candidate, U. of Utah, Human Genetics, graduated 2021)  
James Havrilla (Ph.D. candidate, U. of Utah, Human Genetics, graduated 2109)  
Thomas Sasani (Ph.D. candidate, U. of Utah, Human Genetics, graduated 2020)  
John Kubinski (Undergraduate Biology Senior Thesis candidate)  
Ryan Layer (Ph.D. candidate, Computer Science, graduated 2014)  
Phanwadee Sinthong (Undergraduate Computer Science researcher)  
Brian Lohman (Postdoc)

#### THESIS COMMITTEES

##### **Current**

Megan Conway (Ph.D. candidate, U. of Utah, Oncological Sciences)  
Ryan Miller (Ph.D. candidate, U. of Utah, Oncological Sciences)  
Spencer Arnesen (Ph.D. candidate, U. of Utah, Human Genetics)  
Rosalie Waller (Ph.D. candidate, U. of Utah, Biomedical Informatics)  
Nicole Russell (Ph.D. candidate, U. of Utah, Human Genetics)  
Kristi Russell (Ph.D. candidate, U. of Utah, Human Genetics)  
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)  
Michael Cormier (Ph.D. candidate, U. of Utah, Human Genetics, starting April 2018)

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