Aaron R. Quinlan, Ph.D.

CONTACT INFORMATION

Assistant Professor
Department of Public Health Sciences
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RESEARCH INTERESTS

Chromosome stability and somatic genome evolution; Genomics software development; Cancer genomics; Population genomics; Genetics of complex disease

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
Assistant Professor of Public Health Sciences 2011-
Center for Public Health Genomics

University of Virginia, Charlottesville, VA, USA
NRSA Postdoctoral Fellow (NHGRI) 2008-2011

HONORS AND AWARDS

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 Methods*, *Genome Research*, *Genome Biology Bioinformatics*, *BMC Bioinformatics*, *Bioessays*, *Genes*, and *IEEE Transactions On Computational Biology and Bioinformatics*.

GRANT REVIEW

NIH:
Special Emphasis Panel for Computational Analysis of ENCODE (U01) (16-May-2012)

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

PUBLICATIONS

† *denotes corresponding author*
⋆ *denotes joint first authors*

17. 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.

16. 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.
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.
`code.google.com/p/bamtools`
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`
6. **Quinlan AR**, Clark RA, Sokolova, S, Leibowitx 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`
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.

Research Support

Project Title: *New algorithms and tools for large-scale genomic analysis.*
PI: Aaron Quinlan
Source: NIH/NHGRI (R01 HG006693-01)
Amount: $437,112; Period funded: 19-Apr-2012 - 31-Mar-2016

Project Title: *Identification of radiation sensitivity alleles by whole exome sequencing.*
PI: Pat Concannon
Co-investigator: Aaron Quinlan
Source: NIH/NIEHS (R21 ES020521-01)
Amount: $153,376; Period funded: 19-Aug-2011 - 31-Jul-2013

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)
Amount: $50,000; Period funded: 01-May-2011 - 30-Apr-2012

Project Title: *Defining the genomic architecture of glioblastoma for improved therapy.*
PI: Aaron Quinlan
Source: University of Virginia Cancer Center Pilot Fund
Amount: $53,125; Period funded: 01-Apr-2011 - 31-Mar-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

Exploring genetic variation with a tour guide.
International Stroke Genetics Consortium Meeting; Charlottesville, VA
April 25, 2013

LUMPY: A probablistic 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

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

Guest Lecturer for undergraduate and graduate Biomedical Engineering course (BME 4806, 7806; Prof. Brent French), Spring 2013.
April 8

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

Instructor for CSHL Advanced Sequencing Technologies Course, 2009 - Present

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

Mentorship

Current
Ryan Layer (Ph.D. candidate, Computer Science, qualified 2012)
John Kubinski (Undergraduate Biology Senior Thesis candidate)
Phanwadee Sinthong (Undergraduate Computer Science researcher)

Thesis Committees **Current**

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)