Department of Molecular and Systems Biology Geisel School of Medicine Dartmouth College Hanover, NH

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### Aaron McKenna

Education

Genome Sciences, University of Washington, Seattle, Washington

August, 2017

Ph.D. in Genome Sciences,

Certificates in Computational Biology and Statistical Genetics

Advisor: Jay Shendure M.D. Ph.D.

Boston University, Boston, MA

December, 2008

M.S. in Bioinformatics

Worcester Polytechnic Institute, Worcester, MA

May, 2003

B.S. in Computer Science

**Employment** 

Assistant Professor

May, 2019 -

Dartmouth College. Hanover, NH, USA

Department of Molecular and Systems Biology, Geisel School of Medicine.

Senior Fellow

August, 2017 - April, 2019

University of Washington. Seattle, WA Department of Genome Sciences Advisor: Jay Shendure M.D. Ph.D.

Software Engineer / Computational Biologist

September 2008 - July 2012

The Broad Institute of Harvard and MIT, Cambridge, MA

I designed and implemented the Genome Analysis Toolkit (GATK) a Java framework for next-generation sequencing projects, which is now used at the Broad and other large sequencing centers. This was published as first author in *Genome Research*. In addition, I implemented a method for detecting inter-sample contamination in next generation sequencing samples, published as a first author in *Bioinformatics*, while leading the analysis of The Cancer Genome Atlas (TCGA) Glioblastoma project, published as a first author in *Cell*.

Software Engineer

September 2006 – September 2008

Adaptive Optic Associates, Cambridge, MA

I design, implemented, and tested a measurement and control system for a portable fiber-optic interrogation platform in Java. I also designed, implemented, and tested an embedded Linux system to control a fiber optic dispersion compensator, coded in assembly and C++.

Software Engineer

July 2003 - September 2006

General Dynamics C4 Systems, Needham, MA

I was part of a team that designed, implemented, and tested a packet processor engine for TACLANE inline gigabit IP encryption device under National Security Agency contract, coding in Assembly and C/C++. I developed a prototype Java application for the National Security Agency, incorporating their HMI device interface requirements into a rapid prototype, and I developed an application to automate configuration of encryption devices, greatly reducing production testing schedules. This program selected for excellence, receiving 100% award fee from the National Security Agency.

#### Software Engineer Intern

Summer 1999, 2000, 2001

International Business Machines (IBM), Essex, VT

I worked on a team to develop timing analysis and noise prediction tools for ASIC circuit design on IBM AIX UNIX. I also migrated IBM Circuit Design Tools from AIX UNIX to the Linux platform. Lastly I was nominated for IBM Extreme Blue program.

#### **Patents**

Methods and Apparatus for Analyzing and Quantifying DNA Alterations in Cancer.

Scott L. Carter, Gad Getz, **Aaron McKenna**, and Matthew Meyerson, inventor. The Broad Institute, Inc., Dana-Farber Cancer Institute, Inc, applicant. Patent WO2014026096 A1

Whole-organism lineage tracing by combinatorial and cumulative genome editing. Aaron McKenna, Gregory M. Findlay, James A. Gagnon, Alexander F. Schier, Jay Shendure, inventor. Harvard University and University of Washington, applicant. Provisional Patent

### First Author Publications

FlashFry: a fast and flexible tool for large-scale CRISPR target design

**Aaron McKenna** and Jay Shendure *BMC Biology* 2018 Jul 5;16(1):74.

Whole-organism lineage tracing by combinatorial and cumulative genome editing McKenna A\*, Findlay GM\*, Gagnon JA\*, Horwitz MS, Schier AF, Shendure J *Science* 2016 Jul 29;353(6298):aaf7907

The somatic genomic landscape of glioblastoma

Brennan CW\*, Verhaak RG\*, McKenna A\*, Campos B, Noushmehr H, Salama SR, Zheng S, Chakravarty D, Sanborn JZ, Berman SH, Beroukhim R, Bernard B, Wu CJ, Genovese G, Shmulevich I, Barnholtz-Sloan J, Zou L, Vegesna R, Shukla SA, Ciriello G, Yung WK, Zhang W, Sougnez C, Mikkelsen T, Aldape K, Bigner DD, Van Meir EG, Prados M, Sloan A, Black KL, Eschbacher J, Finocchiaro G, Friedman W, Andrews DW, Guha A, Iacocca M, O'Neill BP, Foltz G, Myers J, Weisenberger DJ, Penny R, Kucherlapati R, Perou CM, Hayes DN, Gibbs R, Marra M, Mills GB, Lander E, Spellman P, Wilson R, Sander C, Weinstein J, Meyerson M, Gabriel S, Laird PW, Haussler D, Getz G, Chin L; TCGA Research Network

Cell 2013 Oct 10;155(2):462-77

ContEst: estimating cross-contamination of human samples in next-generation sequencing data

Ĉibulskis K\*, **McKenna A**\*, Fennell T, Banks E, DePristo M, Getz G *Bioinformatics* 2011 Sep 15;27(18):2601-2

# The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data

McKenna A, Hanna M, Banks E, Sivachenko A, Cibulskis K, Kernytsky A, Garimella K, Altshuler D, Gabriel S, Daly M, DePristo MA *Genome Res* 2010 Sep;20(9):1297-303

#### **Publications**

#### Estimation of cell lineage trees by maximum-likelihood phylogenetics

Jean Feng, William S DeWitt III, **Aaron McKenna**, Noah Simon, Amy Willis, Frederick A Matsen IV *BioRxiv* March, 2019

# Massively parallel profiling and predictive modeling of the outcomes of CRISPR/Cas9-mediated double-strand break repair

Wei Chen, **Aaron McKenna**, Jacob Schreiber, Yi Yin, Vikram Agarwal, William Stafford Noble, Jay Shendure *BioRxiv* November, 2018

# Simultaneous single-cell profiling of lineages and cell types in the vertebrate brain Raj B, Wagner DE, McKenna A, Pandey S, Klein AM, Shendure J, Gagnon JA, Schier AF *Nat Biotechnology* 2018 Jun;36(5):442-450

# CRISPR/Cas9-Mediated Scanning for Regulatory Elements Required for HPRT1 Expression via Thousands of Large, Programmed Genomic Deletions

Gasperini M, Findlay GM, McKenna A, Milbank JH, Lee C, Zhang MD, Cusanovich DA, Shendure J

Am J Hum Genetics 2017 Aug 3;101(2):192-205

#### Systematic genomic and translational efficiency studies of uveal melanoma

Jóhnson CP, Kim IK, Esmaeli B, Amin-Mansour A, Treacy DJ, Carter SL, Hodis E, Wagle N, Seepo S, Yu X, Lane AM, Gragoudas ES, Vazquez F, Nickerson E, Cibulskis K, McKenna A, Gabriel SB, Getz G, Van Allen EM, 't Hoen PAC, Garraway LA, Woodman SE

PLoS One 2017 Jun 8;12(6):e0178189 A comparative assessment of clinical whole exome

# and transcriptome profiling across sequencing centers: implications for precision cancer medicine

Van Allen EM, Robinson D, Morrissey C, Pritchard C, Imamovic A, Carter S, Rosenberg M, McKenna A, Wu YM, Cao X, Chinnaiyan A, Garraway L, Nelson PS Oncotarget 2016 Aug 16;7(33):52888-52899

### Genomic Characterization of Brain Metastases Reveals Branched Evolution and Potential Therapeutic Targets

Brastianos PK, Carter SL, Santagata S, Cahill DP, Taylor-Weiner A, Jones RT, Van Allen EM, Lawrence MS, Horowitz PM, Cibulskis K, Ligon KL, Tabernero J, Seoane J, Martinez-Saez E, Curry WT, Dunn IF, Paek SH, Park SH, McKenna A, Chevalier A, Rosenberg M, Barker FG 2nd, Gill CM, Van Hummelen P, Thorner AR, Johnson BE, Hoang MP, Choueiri TK, Signoretti S, Sougnez C, Rabin MS, Lin NU, Winer EP, Stemmer-Rachamimov A, Meyerson M, Garraway L, Gabriel S, Lander ES, Beroukhim R, Batchelor TT, Baselga J, Louis DN, Getz G, Hahn WC

Cancer Discovery 2015 Nov;5(11):1164-1177

#### Paired exome analysis of Barrett's esophagus and adenocarcinoma

Stachler MD, Taylor-Weiner A, Peng S, McKenna A, Agoston AT, Odze RD, Davison JM, Nason KS, Loda M, Leshchiner I, Stewart C, Stojanov P, Seepo S, Lawrence MS, Ferrer-Torres D, Lin J, Chang AC, Gabriel SB, Lander ES, Beer DG, Getz G, Carter SL, Bass AJ *Nat Genetics* 2015 Sep;47(9):1047-55

### Genomic Correlate of Exceptional Erlotinib Response in Head and Neck Squamous Cell Carcinoma

Van Allen EM, Lui VW, Egloff AM, Goetz EM, Li H, Johnson JT, Duvvuri U, Bauman JE, Stransky N, Zeng Y, Gilbert BR, Pendleton KP, Wang L, Chiosea S, Sougnez C, Wagle N, Zhang F, Du Y, Close D, Johnston PA, McKenna A, Carter SL, Golub TR, Getz G, Mills GB, Garraway LA, Grandis JR

JAMA Oncology 2015 May;1(2):238-44

#### Prospective derivation of a living organoid biobank of colorectal cancer patients

van de Wetering M, Francies HE, Francis JM, Bounova G, Iorio F, Pronk A, van Houdt W, van Gorp J, Taylor-Weiner A, Kester L, McLaren-Douglas A, Blokker J, Jaksani S, Bartfeld S, Volckman R, van Sluis P, Li VS, Seepo S, Sekhar Pedamallu C, Cibulskis K, Carter SL, McKenna A, Lawrence MS, Lichtenstein L, Stewart C, Koster J, Versteeg R, van Oudenaarden A, Saez-Rodriguez J, Vries RG, Getz G, Wessels L, Stratton MR, McDermott U, Meyerson M, Garnett MJ, Clevers H

Cell 2015 May 7;161(4):933-45

# Complementary genomic approaches highlight the PI<sub>3</sub>K/mTOR pathway as a common vulnerability in osteosarcoma

Perry JA, Kiezun A, Tonzi P, Van Allen EM, Carter SL, Baca SC, Cowley GS, Bhatt AS, Rheinbay E, Pedamallu CS, Helman E, Taylor-Weiner A, McKenna A, DeLuca DS, Lawrence MS, Ambrogio L, Sougnez C, Sivachenko A, Walensky LD, Wagle N, Mora J, de Torres C, Lavarino C, Dos Santos Aguiar S, Yunes JA, Brandalise SR, Mercado-Celis GE, Melendez-Zajgla J, C⊠rdenas-Card⊠s R, Velasco-Hidalgo L, Roberts CW, Garraway LA, Rodriguez-Galindo C, Gabriel SB, Lander ES, Golub TR, Orkin SH, Getz G, Janeway KA *Proc Natl Acad Sci U S A* 2014 Dec 23;111(51):E5564-73

#### The genomic landscape of pediatric Ewing sarcoma

Crompton BD, Stewart C, Taylor-Weiner A, Alexe G, Kurek KC, Calicchio ML, Kiezun A, Carter SL, Shukla SA, Mehta SS, Thorner AR, de Torres C, Lavarino C, Su\( \text{Sol} M\), McKenna A, Sivachenko A, Cibulskis K, Lawrence MS, Stojanov P, Rosenberg M, Ambrogio L, Auclair D, Seepo S, Blumenstiel B, DeFelice M, Imaz-Rosshandler I, Schwarz-Cruz Y Celis A, Rivera MN, Rodriguez-Galindo C, Fleming MD, Golub TR, Getz G, Mora J, Stegmaier K

Cancer Discovery 2014 Nov;4(11):1326-41

# Whole-exome sequencing and clinical interpretation of formalin-fixed, paraffin- embedded tumor samples to guide precision cancer medicine

Van Allen EM, Wagle N, Stojanov P, Perrin DL, Cibulskis K, Marlow S, Jane-Valbuena J, Friedrich DC, Kryukov G, Carter SL, **McKenna** A, Sivachenko A, Rosenberg M, Kiezun A, Voet D, Lawrence M, Lichtenstein LT, Gentry JG, Huang FW, Fostel J, Farlow D, Barbie D, Gandhi L, Lander ES, Gray SW, Joffe S, Janne P, Garber J, MacConaill L, Lindeman N, Rollins B, Kantoff P, Fisher SA, Gabriel S, Getz G, Garraway LA *Nat Med* 2014 Jun;20(6):682-8

### Genetic and clonal dissection of murine small cell lung carcinoma progression by genome sequencing

McFadden DG, Papagiannakopoulos T, Taylor-Weiner A, Stewart C, Carter SL, Cibulskis K, Bhutkar A, McKenna A, Dooley A, Vernon A, Sougnez C, Malstrom S, Heimann M, Park J, Chen F, Farago AF, Dayton T, Shefler E, Gabriel S, Getz G, Jacks T *Cell* 2014 Mar 13;156(6):1298-1311

### Widespread genetic heterogeneity in multiple myeloma: implications for targeted therapy

Lohr JG, Stojanov P, Carter SL, Cruz-Gordillo P, Lawrence MS, Auclair D, Sougnez C, Knoechel B, Gould J, Saksena G, Cibulskis K, **McKenna A**, Chapman MA, Straussman R, Levy J, Perkins LM, Keats JJ, Schumacher SE, Rosenberg M; Multiple Myeloma Research Consortium, Getz G, Golub TR

Cancer Cell 2014 Jan 13;25(1):91-101

#### Landscape of genomic alterations in cervical carcinomas

Ojesina AI, Lichtenstein L, Freeman SS, Pedamallu CS, Imaz-Rosshandler I, Pugh TJ, Cherniack AD, Ambrogio L, Cibulskis K, Bertelsen B, Romero-Cordoba S, Treviळo V, Vazquez-Santillan K, Guadarrama AS, Wright AA, Rosenberg MW, Duke F, Kaplan B, Wang R, Nickerson E, Walline HM, Lawrence MS, Stewart C, Carter SL, McKenna A, Rodriguez-Sanchez IP, Espinosa-Castilla M, Woie K, Bjorge L, Wik E, Halle MK, Hoivik EA, Krakstad C, Gabiळo NB, Gळmez-Macळas GS, Valdez-Chapa LD, Garza-Rodrळguez ML, Maytorena G, Vazquez J, Rodea C, Cravioto A, Cortes ML, Greulich H, Crum CP, Neuberg DS, Hidalgo-Miranda A, Escareno CR, Akslen LA, Carey TE, Vintermyr OK, Gabriel SB, Barrera-Saldaळa HA, Melendez-Zajgla J, Getz G, Salvesen HB, Meyerson M Nature 2014 Feb 20;506(7488):371-5

### Integrative and comparative genomic analysis of lung squamous cell carcinomas in East Asian patients

Kim Y, Hammerman PS, Kim J, Yoon JA, Lee Y, Sun JM, Wilkerson MD, Pedamallu CS, Cibulskis K, Yoo YK, Lawrence MS, Stojanov P, Carter SL, **McKenna A**, Stewart C, Sivachenko AY, Oh IJ, Kim HK, Choi YS, Kim K, Shim YM, Kim KS, Song SY, Na KJ, Choi YL, Hayes DN, Kim J, Cho S, Kim YC, Ahn JS, Ahn MJ, Getz G, Meyerson M, Park K

J Clin Oncology 2014 Jan 10;32(2):121-8

# MAP kinase pathway alterations in BRAF-mutant melanoma patients with acquired resistance to combined RAF/MEK inhibition

Wagle N, Van Allen EM, Treacy DJ, Frederick DT, Cooper ZA, Taylor-Weiner A, Rosenberg M, Goetz EM, Sullivan RJ, Farlow DN, Friedrich DC, Anderka K, Perrin D, Johannessen CM, **McKenna A**, Cibulskis K, Kryukov G, Hodis E, Lawrence DP, Fisher S, Getz G, Gabriel SB, Carter SL, Flaherty KT, Wargo JA, Garraway LA *Cancer Discovery* 2014 Jan;4(1):61-8

The genetic landscape of clinical resistance to RAF inhibition in metastatic melanoma Van Allen EM, Wagle N, Sucker A, Treacy DJ, Johannessen CM, Goetz EM, Place CS, Taylor-Weiner A, Whittaker S, Kryukov GV, Hodis E, Rosenberg M, McKenna A, Cibulskis K, Farlow D, Zimmer L, Hillen U, Gutzmer R, Goldinger SM, Ugurel S, Gogas HJ, Egberts F, Berking C, Trefzer U, Loquai C, Weide B, Hassel JC, Gabriel SB, Carter SL, Getz G, Garraway LA, Schadendorf D; Dermatologic Cooperative Oncology Group of

Germany (DeCOG) Cancer Discovery 2014 Jan;4(1):94-109

#### Somatic mutation of CDKN<sub>1</sub>B in small intestine neuroendocrine tumors

Francis JM, Kiezun A, Ramos AH, Serra S, Pedamallu CS, Qian ZR, Banck MS, Kanwar R, Kulkarni AA, Karpathakis A, Manzo V, Contractor T, Philips J, Nickerson E, Pho N, Hooshmand SM, Brais LK, Lawrence MS, Pugh T, McKenna A, Sivachenko A, Cibulskis K, Carter SL, Ojesina AI, Freeman S, Jones RT, Voet D, Saksena G, Auclair D, Onofrio R, Shefler E, Sougnez C, Grimsby J, Green L, Lennon N, Meyer T, Caplin M, Chung DC, Beutler AS, Ogino S, Thirlwell C, Shivdasani R, Asa SL, Harris CR, Getz G, Kulke M, Meyerson M

Nat Genetics 2013 Dec;45(12):1483-6

### A survey of intragenic breakpoints in glioblastoma identifies a distinct subset associated with poor survival

Zheng S, Fu J, Vegesna R, Mao Y, Heathcock LE, Torres-Garcia W, Ezhilarasan R, Wang S, **McKenna** A, Chin L, Brennan CW, Yung WK, Weinstein JN, Aldape KD, Sulman EP, Chen K, Koul D, Verhaak RG

Genes Dev 2013 Jul 1;27(13):1462-72

#### Mutational heterogeneity in cancer and the search for new cancer-associated genes

Lawrence MS, Stojanov P, Polak P, Kryukov GV, Cibulskis K, Sivachenko A, Carter SL, Stewart C, Mermel CH, Roberts SA, Kiezun A, Hammerman PS, McKenna A, Drier Y, Zou L, Ramos AH, Pugh TJ, Stransky N, Helman E, Kim J, Sougnez C, Ambrogio L, Nickerson E, Shefler E, Cort\( \Sigma \) ML, Auclair D, Saksena G, Voet D, Noble M, DiCara D, Lin P, Lichtenstein L, Heiman DI, Fennell T, Imielinski M, Hernandez B, Hodis E, Baca S, Dulak AM, Lohr J, Landau DA, Wu CJ, Melendez-Zajgla J, Hidalgo-Miranda A, Koren A, McCarroll SA, Mora J, Crompton B, Onofrio R, Parkin M, Winckler W, Ardlie K, Gabriel SB, Roberts CWM, Biegel JA, Stegmaier K, Bass AJ, Garraway LA, Meyerson M, Golub TR, Gordenin DA, Sunyaev S, Lander ES, Getz G

Nature 2013 Jul 11;499(7457):214-218

# Genomic analysis of diffuse pediatric low-grade gliomas identifies recurrent oncogenic truncating rearrangements in the transcription factor MYBL1

Ramkissoon LA, Horowitz PM, Craig JM, Ramkissoon SH, Rich BE, Schumacher SE, McKenna A, Lawrence MS, Bergthold G, Brastianos PK, Tabak B, Ducar MD, Van Hummelen P, MacConaill LE, Pouissant-Young T, Cho YJ, Taha H, Mahmoud M, Bowers DC, Margraf L, Tabori U, Hawkins C, Packer RJ, Hill DA, Pomeroy SL, Eberhart CG, Dunn IF, Goumnerova L, Getz G, Chan JA, Santagata S, Hahn WC, Stiles CD, Ligon AH, Kieran MW, Beroukhim R, Ligon KL

Proc Natl Acad Sci U S A 2013 May 14;110(20):8188-93

# Exome and whole-genome sequencing of esophageal adenocarcinoma identifies recurrent driver events and mutational complexity

Dulak AM, Stojanov P, Peng S, Lawrence MS, Fox C, Stewart C, Bandla S, Imamura Y, Schumacher SE, Shefler E, **McKenna A**, Carter SL, Cibulskis K, Sivachenko A, Saksena G, Voet D, Ramos AH, Auclair D, Thompson K, Sougnez C, Onofrio RC, Guiducci C, Beroukhim R, Zhou Z, Lin L, Lin J, Reddy R, Chang A, Landrenau R, Pennathur A, Ogino S, Luketich JD, Golub TR, Gabriel SB, Lander ES, Beer DG, Godfrey TE, Getz G, Bass AJ *Nat Genetics* 2013 May;45(5):478-86

#### Evolution and impact of subclonal mutations in chronic lymphocytic leukemia

Landau DA, Carter SL, Stojanov P, **McKenna A**, Stevenson K, Lawrence MS, Sougnez C, Stewart C, Sivachenko A, Wang L, Wan Y, Zhang W, Shukla SA, Vartanov A, Fernandes SM, Saksena G, Cibulskis K, Tesar B, Gabriel S, Hacohen N, Meyerson M, Lander ES, Neuberg D, Brown JR, Getz G, Wu CJ

Cell 2013 Feb 14;152(4):714-26

#### Integrative eQTL-based analyses reveal the biology of breast cancer risk loci

Li Q, Seo JH, Stranger B, McKenna A, Pe'er I, Laframboise T, Brown M, Tyekucheva S, Freedman ML

Cell 2013 Jan 31;152(3):633-41

### Genomic sequencing of meningiomas identifies oncogenic SMO and AKT1 mutations Brastianos PK, Horowitz PM, Santagata S, Jones RT, McKenna A, Getz G, Ligon KL,

Palescandolo E, Van Hummelen P, Ducar MD, Raza A, Sunkavalli A, Macconaill LE, Stemmer-Rachamimov AO, Louis DN, Hahn WC, Dunn IF, Beroukhim R *Nat Genetics* 2013 Mar;45(3):285-9

The genetic landscape of high-risk neuroblastoma

Pugh TJ, Morozova O, Attiyeh EF, Asgharzadeh S, Wei JS, Auclair D, Carter SL, Cibulskis

K, Hanna M, Kiezun A, Kim J, Lawrence MS, Lichenstein L, McKenna A, Pedamallu CS, Ramos AH, Shefler E, Sivachenko A, Sougnez C, Stewart C, Ally A, Birol I, Chiu R, Corbett RD, Hirst M, Jackman SD, Kamoh B, Khodabakshi AH, Krzywinski M, Lo A, Moore RA, Mungall KL, Qian J, Tam A, Thiessen N, Zhao Y, Cole KA, Diamond M, Diskin SJ, Mosse YP, Wood AC, Ji L, Sposto R, Badgett T, London WB, Moyer Y, Gastier-Foster JM, Smith MA, Guidry Auvil JM, Gerhard DS, Hogarty MD, Jones SJ, Lander ES, Gabriel SB, Getz G, Seeger RC, Khan J, Marra MA, Meyerson M, Maris JM *Nat Genetics* 2013 Mar;45(3):279-84

### Whole-exome sequencing identifies a recurrent NAB2-STAT6 fusion in solitary fibrous tumors

Chmielecki J, Crago AM, Rosenberg M, O'Connor R, Walker SR, Ambrogio L, Auclair D, McKenna A, Heinrich MC, Frank DA, Meyerson M

Nat Genetics 2013 Feb;45(2):131-2

### Comprehensive genomic characterization of squamous cell lung cancers

Cancer Genome Atlas Research Network *Nature* 2012 Sep 27;489(7417):519-25

Medulloblastoma exome sequencing uncovers subtype-specific somatic mutations

Pugh TJ, Weeraratne SD, Archer TČ, Pomeranz Krummel DA, Auclair D, Bochicchio J, Carneiro MO, Carter SL, Cibulskis K, Erlich RL, Greulich H, Lawrence MS, Lennon NJ, McKenna A, Meldrim J, Ramos AH, Ross MG, Russ C, Shefler E, Sivachenko A, Sogoloff B, Stojanov P, Tamayo P, Mesirov JP, Amani V, Teider N, Sengupta S, Francois JP, Northcott PA, Taylor MD, Yu F, Crabtree GR, Kautzman AG, Gabriel SB, Getz G, J\(\text{\textit{Z}}\)ger N, Jones DT, Lichter P, Pfister SM, Roberts TM, Meyerson M, Pomeroy SL, Cho YJ Nature 2012 Aug 2;488(7409):106-10

Comprehensive molecular characterization of human colon and rectal cancer

Cancer Genome Atlas Network

Nature 2012 Jul 18;487(7407):330-7

#### Absolute quantification of somatic DNA alterations in human cancer

Carter SL, Cibulskis K, Helman E, **McKenna A**, Shen H, Zack T, Laird PW, Onofrio RC, Winckler W, Weir BA, Beroukhim R, Pellman D, Levine DA, Lander ES, Meyerson M, Getz G

*Nat Biotechnology* 2012 May;30(5):413-21

### The 1000 Genomes Project: data management and community access

Clarke L, Zheng-Bradley X, Smith R, Kulesha E, Xiao C, Toneva I, Vaughan B, Preuss D, Leinonen R, Shumway M, Sherry S, Flicek P; 1000 Genomes Project Consortium *Nat Methods* 2012 Apr 27;9(5):459-62

### The functional spectrum of low-frequency coding variation

Marth GT, Yu F, Îndap AR, Garimella K, Gravel S, Leong WF, Tyler-Smith C, Bainbridge M, Blackwell T, Zheng-Bradley X, Chen Y, Challis D, Clarke L, Ball EV, Cibulskis K, Cooper DN, Fulton B, Hartl C, Koboldt D, Muzny D, Smith R, Sougnez C, Stewart C, Ward A, Yu J, Xue Y, Altshuler D, Bustamante CD, Clark AG, Daly M, DePristo M, Flicek P, Gabriel S, Mardis E, Palotie A, Gibbs R; 1000 Genomes Project *Genome Biol* 2011 Sep 14;12(9):R84

### The mutational landscape of head and neck squamous cell carcinoma

Stransky N, Egloff AM, Tward AD, Kostic AD, Čibulskis K, Sivachenko A, Kryukov GV, Lawrence MS, Sougnez C, McKenna A, Shefler E, Ramos AH, Stojanov P, Carter SL, Voet D, Cort\( \text{S} & ML, Auclair D, Berger MF, Saksena G, Guiducci C, Onofrio RC, Parkin M, Romkes M, Weissfeld JL, Seethala RR, Wang L, Rangel-Escare\( \text{S} & C, Fernandez-Lopez JC, Hidalgo-Miranda A, Melendez-Zajgla J, Winckler W, Ardlie K, Gabriel SB, Meyerson M, Lander ES, Getz G, Golub TR, Garraway LA, Grandis JR Science 2011 Aug 26;333(6046):1157-60

### Demographic history and rare allele sharing among human populations

Gravel S, Henn BM, Gutenkunst RN, Indap AR, Marth GT, Clark AG, Yu F, Gibbs RA; 1000 Genomes Project, Bustamante CD *Proc Natl Acad Sci U S A* 2011 Jul 19;108(29):11983-8

#### Variation in genome-wide mutation rates within and between human families

Conrad DF, Keebler JE, DePristo MA, Lindsay SJ, Zhang Y, Casals F, Idaghdour Y, Hartl CL, Torroja C, Garimella KV, Zilversmit M, Cartwright R, Rouleau GA, Daly M, Stone EA, Hurles ME, Awadalla P; 1000 Genomes Project *Nat Genetics* 2011 Jun 12;43(7):712-4

#### The variant call format and VCFtools

Danecek P, Auton A, Abecasis G, Albers CA, Banks E, DePristo MA, Handsaker RE, Lunter G, Marth GT, Sherry ST, McVean G, Durbin R; 1000 Genomes Project Analysis Group

Bioinformatics 2011 Aug 1;27(15):2156-8

A framework for variation discovery and genotyping using next-generation DNA sequencing data

DePristo MA, Banks E, Poplin R, Garimella KV, Maguire JR, Hartl C, Philippakis AA, del Angel G, Rivas MA, Hanna M, **McKenna A**, Fennell TJ, Kernytsky AM, Sivachenko AY, Cibulskis K, Gabriel SB, Altshuler D, Daly MJ *Nat Genetics* 2011 May;43(5):491-8

Mapping copy number variation by population-scale genome sequencing

Mills RE, Walter K, Stewart C, Handsaker RE, Chen K, Alkan C, Abyzov A, Yoon SC, Ye K, Cheetham RK, Chinwalla A, Conrad DF, Fu Y, Grubert F, Hajirasouliha I, Hormozdiari F, Iakoucheva LM, Iqbal Z, Kang S, Kidd JM, Konkel MK, Korn J, Khurana E, Kural D, Lam HY, Leng J, Li R, Li Y, Lin CY, Luo R, Mu XJ, Nemesh J, Peckham HE, Rausch T, Scally A, Shi X, Stromberg MP, St\(\text{Ztz AM}\), Urban AE, Walker JA, Wu J, Zhang Y, Zhang ZD, Batzer MA, Ding L, Marth GT, McVean G, Sebat J, Snyder M, Wang J, Ye K, Eichler EE, Gerstein MB, Hurles ME, Lee C, McCarroll SA, Korbel JO; 1000 Genomes Project Nature 2011 Feb 3;470(7332):59-65

#### Diversity of human copy number variation and multicopy genes

Sudmant PH, Kitzman JO, Antonacci F, Alkan C, Malig M, Tsalenko A, Sampas N, Bruhn L, Shendure J; 1000 Genomes Project, Eichler EE *Science* 2010 Oct 29;330(6004):641-6

A map of human genome variation from population-scale sequencing

1000 Genomes Project Consortium, Abecasis GR, Altshuler D, Auton A, Brooks LD, Durbin RM, Gibbs RA, Hurles ME, McVean GA
Nature 2010 Oct 28;467(7319):1061-73

### Honors and Funding

NIH Pathway to Independence Award K99/Roo (1K99HG010152-01) 2018-2023

Graduate School's Distinguished Dissertation Award Winner

2018

National Institute of Health (NIH) National Research Service Award (T<sub>32</sub>) Trainee for Experimental Pathology of Cardiovascular Disease (5T<sub>32</sub>HL007<sub>312</sub>-37) 2015-2017

National Institute of Health (NIH) Institutional National Research Service Award (T<sub>32</sub>) (5T<sub>32</sub>HG0000<sub>35</sub>-20) 2012-2014

#### Select Talks

"Improvements to CRISPR-based lineage tracing: A potpourri of success, challenges, and opportunities". High-Throughput Dense Reconstruction of Cell Lineages meeting, Janelia Farms / HHMI.

April, 2019

"Whole organism lineage tracing in Drosophila with GESTALT". Second Fly Cell Atlas meeting.

March, 2019

Cellular 'phylogenetics' - decoding the developmental history and relationships among individual cells. Phylogenetics seminar.

March, 2019

Single cell state and fate: application to tumor heterogeneity and metastasis. National Cancer Institute Workshop on Linear and Non-Linear Metastases. September, 2018

Capturing high-dimensional cell state and cell fate from single cells. Machine Learning Strategies for Disease Prediction. Technical University of Denmark. May, 2018

Recovering single-cell state in conjunction with lineage in Drosophila. Fly Cell Atlas meeting. University of Leuven.

December, 2017

Information storage and recovery using a diversity of second-generation sequencing technologies. Genome 10K and Genome Science 2017. Earlham Institute. September, 2017

Whole-organism lineage tracing with CRISPR-Cas9 barcodes. Genome Engineering Workshop 5.0. The Broad Institute of Harvard and MIT.

May, 2017

Contiguity in Genome Sequencing – Methods and Applications. NHGRI Sequencing Technology Development Meeting.

May, 2015

Mutation discovery in tumor sequencing. Our team led a two-day training on cancer genomics at the Carlos Slim Institute of Health in Mexico City.

June, 2012

#### Service

**Reviewed for:** Science, Cell, Bioinformatics, Molecular Oncology, BMC Medical Genomics, eLife, GigaScience, Nucleotide Acids Research, PLOS Computational Biology

#### **Program committee for:**

AlCoB 2019, May 2019, University of California, Berkeley, CA

### Other professional service:

Co-founder of the Young Alliance Against Cancer (YAAC), a Germany-based non-profit focused on bridging the gap between basic science and clinical training for young cancer researchers

### Teaching Experience

LecturerSpring 2019QBS.146.01: Molecular and Computational GenomicsDartmouth College

Teaching AssistantWinter 2016Genome Sciences 466: Cancer GeneticsUniversity of Washington

Teaching AssistantFall 2015Genome 371: Introductory GeneticsUniversity of Washington