

# DeannaMChurch

Genomics, Technology and Bioinformatics Leader

## contact

3538 22nd St.  
Boulder, CO 80304  
USA

(301) 233 2991

deanna.church@gmail.com

## skills

Leadership, Mentorship,  
Product  
Commercialization,  
Team Player,  
Project Management,  
Curiosity & Initiative

## genomics

Genome editing,  
Genome assembly,  
Genome annotation,  
variant analysis &  
Single-cell 'omics

## bioinformatics

Python, R, Git,  
HTML, XML/XSLT,  
Javascript,  
CSS3, SQL & Perl  
github.com/deannachurch

## languages

english, mother tongue

## focus

Leading interdisciplinary teams to deliver cutting edge life sciences tools.

## experience

2021–present **Inscripta, Inc**

Boulder, CO

*Vice President, Mammalian Business Area and Software Strategy*

Driving customer voice and commercialization of our mammalian editing platform. Ensuring our software platform supports all of our platforms and provides customers with an excellent experience.

2019–2021 **Inscripta, Inc**

Boulder, CO

*Senior Director, Applications Mammalian*

Driving customer voice of our mammalian editing platform.

2016–2018 **10x Genomics**

Pleasant, CA

*Senior Director of Applications*

Led an interdisciplinary team of top scientists in demonstrating the scientific value of the full 10x product suite. This work covers a variety of scientific areas including single cell 'omics, genome assembly, genome analysis, tumor microenvironment analysis, tumor heterogeneity and immune profiling.

2014–2016 **Personalis, Inc**

Menlo Park, CA

*Senior Director of Genomics and Content*

Led a team developing novel genome analysis algorithms and pipelines and knowledge bases built to support genome interpretation.

2001–2014 **National Center for Biotechnology Information, NIH**

Bethesda, MA

*Staff Scientist*

Participated in and led numerous international collaborations including helping to found the Genome Reference Consortium. Developed tools and resources for the scientific community.

1990–1992 **MIT and MGH**

Boston, MA

*Laboratory Technician*

Managed laboratory and performed experiments, including helping to develop the exon amplification protocol.

## education

1999–2001 **National Center for Biotechnology Information, NIH**

Bethesda, MD

*Postdoctoral fellow*

1997–1999 **Laboratory of Janet Rossant, Mt. Sinai Hospital**

Toronto, Ontario

*Postdoctoral fellow*

1992–1997 **PhD of Biological Sciences**

The University of California, Irvine

*Phenotype-genotype correlation in the Cri-du-Chat region of chromosome 5*

1986–1990 **Bachelor of Liberal Arts**

University of Virginia

*Research project focused on cell cycle regulation using yeast*

## advisory boards and professional service

2020-present	<b>Human Pangenome Reference Consortium</b>	Scientific Advisory Board
2018-present	<b>Genome Quebec</b>	Scientific Advisory Board
2018-2020	<b>American Association for Cancer Research</b>	Member
2012-present	<b>Ensembl</b>	Scientific Advisory Board
2014-2016	<b>American College of Medical Genetics</b>	Member
2013-2016	<b>The Genome Analysis Center/Earlham Institute</b>	Scientific Advisory Board
2010-2014	<b>Database of Genomic Variants</b>	Scientific Advisory Board
2010-present	<b>American Society of Human Genetics</b>	Member
2008-2013	<b>Advances in Genome Biology and Technology</b>	Organizing Committee Member
2008-2013	<b>International Standards Cytogenomic Arrays</b>	Committee Member
2008-2012	<b>International Mammalian Genome Society</b>	Nomenclature Committee Member
2004-2012	<b>International Mammalian Genome Society</b>	Member
2006-2011	<b>European Conditional Mouse Mutagenesis Program</b>	Scientific Advisory Board
2006-2009	<b>Genome Canada-MORGEN project</b>	Scientific Advisory Board
2005	<b>Mouse Genome Finishing Meeting</b>	Organizer

## presentations

Selected highlights from over 50 presentations.

2019-Nov	<b>Rosalind Franklin Society</b> Invited Speaker, "The impact of technology on our view of biology"	Philadelphia, PA
2018-Feb	<b>Beckman Symposium, Technology Innovation and Human Genomics</b> Invited Speaker, "The impact of technology on our view of biology"	Palo Alto, CA
2017-Nov	<b>Australasian Genomic Technologies Association Annual Meeting</b> Keynote, "The impact of technology on biology"	Hobart, AUS
2016-Feb	<b>Stanford Big Data in Genomics</b> Invited Speaker, "Truly personalized genomics"	Palo Alto, CA
2015-Oct	<b>Genome informatics</b> Invited Speaker, "The Rumsfeldian challenge of developing clinical sequencing tests"	Cold Spring Harbor, NY
2014-Nov	<b>Biological Data Sciences</b> Keynote Speaker, "Analog reporting in a digital age"	Cold Spring Harbor, NY
2013-Oct	<b>American Society of Human Genetics</b> Invited Speaker, "Navigating clinical genomic resources at NCBI"	Boston, MA
2013- May	<b>Sequencing Finishing and Analysis in the Future</b> Keynote Address, "Keep calm and carry on sequencing"	Santa Fe, NM
2011-Jul	<b>BIO-IT APAC Conference and Expo</b> Keynote Speaker, "The evolution of the reference human genome"	Shenzhen, China

## teaching

Selected courses and lectures.

- |            |   |
|------------|---|
| 2017       | <b>Stanford, Informatics in Industry</b><br>Prepared lecture for 50 students.   |
| 2013       | <b>54th Annual Short Course on Medical and Experimental Mammalian Genetics</b><br>Prepared 6 hours of lecture and online tutorials for 75 students.     |
| 2012, 2008 | <b>Genome Reference Consortium Workshop at ASHG</b><br>Prepared 4 hours of lecture on the human reference assembly for 250 people.                      |
| 2011-2012  | <b>Frontiers in Reproduction</b><br>Prepared 4 hours of lecture on online tutorials for 25 students.  |
| 2003-2012  | <b>International Mammalian Genome Conference</b><br>Co-organized and prepared 4 hours of lecture and online tutorials for 50-75 students.               |
| 2008       | <b>NIDA Short Course on Genetics and Epigenetics</b><br>Prepared 2 hours of lecture for 100 students  |
| 2004-2005  | <b>AGBT workshop</b><br>Prepared 1 hour lecture for the pre-meeting workshop.   |
| 2003-2005  | <b>The Jackson Laboratory Bioinformatics for Bench Scientists</b><br>Co-organized this two week course to introduce bench scientists to bioinformatics. |

## publications

### Articles

De novo assembly of the olive fruit fly (*Bactrocera oleae*) genome with linked-reads and long-read technologies minimizes gaps and provides exceptional Y chromosome assembly  
Anthony Bayega, Haig Djambazian, Konstantina T Tsoumani, Maria-Eleni Gregoriou, Efthimia Sagri, Eleni Drosopoulou, Penelope Mavragani-Tsipidou, Kristina Giorda, George Tsiamis, Kostas Bourtzis, Spyridon Oikonomopoulos, Ken Dewar, Deanna M Church, Alexie Papanicolaou, Kostas D Mathiopoulos, and Jiannis Ragoussis  
*BMC Genomics* 21.1 p. 259. 2020

Thousands of human sequences provide deep insight into single genomes  
Deanna M Church  
*Nature* 581.7809 pp. 385–386. 2020

Single-Cell Transcriptomics Reveals Early Emergence of Liver Parenchymal and Non-parenchymal Cell Lineages  
Jeremy Lotto, Sibyl Drissler, Rebecca Cullum, Wei Wei, Manu Setty, Erin M Bell, Stéphane C Boutet, Sonja Nowotschin, Ying-Yi Kuo, Vidur Garg, Dana Pe'er, Deanna M Church, Anna-Katerina Hadjantonakis, and Pamela A Hoodless  
*Cell* 183.3 702–716.e14. 2020

Haplotyping the *Vitis* collinear core genome with rhAmpSeq improves marker transferability in a diverse genus  
Cheng Zou, Avinash Karn, Bruce Reisch, Allen Nguyen, Yongming Sun, Yun Bao, Michael S Campbell, Deanna Church, Stephen Williams, Xia Xu, Craig A Ledbetter, Sagar Patel, Anne Fennell, Jeffrey C Glaubitz, Matthew Clark, Doreen Ware, Jason P Londo, Qi Sun, and Lance Cadle-Davidson  
*Nat. Commun.* 11.1 p. 413. 2020

Multi-platform discovery of haplotype-resolved structural variation in human genomes

Mark J P Chaisson, Ashley D Sanders, Xuefang Zhao, Ankit Malhotra, David Porubsky, Tobias Rausch, Eugene J Gardner, Oscar L Rodriguez, Li Guo, Ryan L Collins, Xian Fan, Jia Wen, Robert E Handsaker, Susan Fairley, Zev N Kronenberg, Xiangmeng Kong, Fereydoon Hormozdiani, Dillon Lee, Aaron M Wenger, Alex R Hastie, Danny Antaki, Thomas Anantharaman, Peter A Audano, Harrison Brand, Stuart Cantsilieris, Han Cao, Eliza Cerveira, Chong Chen, Xintong Chen, Chen-Shan Chin, Zeichen Chong, Nelson T Chuang, Christine C Lambert, Deanna M Church, Laura Clarke, Andrew Farrell, Joey Flores, Timur Galeev, David U Gorkin, Madhusudan Gujral, Victor Guryev, William Haynes Heaton, Jonas Korlach, Sushant Kumar, Jee Young Kwon, Ernest T Lam, Jong Eun Lee, Joyce Lee, Wan-Ping Lee, Sau Peng Lee, Shantao Li, Patrick Marks, Karine Viaud-Martinez, Sascha Meiers, Katherine M Munson, Fabio C P Navarro, Bradley J Nelson, Conor Nodzak, Amina Noor, Sofia Kyriazopoulou-Panagiotopoulou, Andy W C Pang, Yunjiang Qiu, Gabriel Rosanio, Mallory Ryan, Adrian Stütz, Diana C J Spierings, Alistair Ward, Annemarie E Welch, Ming Xiao, Wei Xu, Chengsheng Zhang, Qihui Zhu, Xiangqun Zheng-Bradley, Ernesto Lowy, Sergei Yakneen, Steven McCarroll, Goo Jun, Li Ding, Chong Lek Koh, Bing Ren, Paul Flicek, Ken Chen, Mark B Gerstein, Pui-Yan Kwok, Peter M Lansdorp, Gabor T Marth, Jonathan Sebat, Xinghua Shi, Ali Bashir, Kai Ye, Scott E Devine, Michael E Talkowski, Ryan E Mills, Tobias Marschall, Jan O Korbel, Evan E Eichler, and Charles Lee

*Nat. Commun. 10.1 p. 1784. Nature Publishing Group, 2019*

### Resolving the full spectrum of human genome variation using Linked-Reads

Patrick Marks, Sarah Garcia, Alvaro Martinez Barrio, Kamila Belhocine, Jorge Bernate, Rajiv Bhargava, Keith Bjornson, Claudia Catalanotti, Josh Delaney, Adrian Fehr, Ian T Fiddes, Brendan Galvin, Haynes Heaton, Jill Herschleb, Christopher Hindson, Esty Holt, Cassandra B Jabara, Susanna Jett, Nikka Keivanfar, Sofia Kyriazopoulou-Panagiotopoulou, Monkol Lek, Bill Lin, Adam Lowe, Shazia Mahamdallie, Shamoni Maheshwari, Tony Makarewicz, Jamie Marshall, Francesca Meschi, Christopher J O'Keefe, Heather Ordonez, Pranav Patel, Andrew Price, Ariel Royall, Elise Ruark, Sheila Seal, Michael Schnall-Levin, Preyas Shah, David Stafford, Stephen Williams, Indira Wu, Andrew Wei Xu, Nazneen Rahman, Daniel MacArthur, and Deanna M Church

*Genome Res. 29.4 pp. 635–645. 2019*

### The emergent landscape of the mouse gut endoderm at single-cell resolution

Sonja Nowotschin, Manu Setty, Ying-Yi Kuo, Vincent Liu, Vidur Garg, Roshan Sharma, Claire S Simon, Nestor Saiz, Rui Gardner, Stéphane C Boutet, Deanna M Church, Pamela A Hoodless, Anna-Katerina Hadjantonakis, and Dana Pe'er

*Nature. 2019*

### A general approach for detecting expressed mutations in AML cells using single cell RNA-sequencing

Allegra A Petti, Stephen R Williams, Christopher A Miller, Ian T Fiddes, Sridhar N Srivatsan, David Y Chen, Catrina C Fronick, Robert S Fulton, Deanna M Church, and Timothy J Ley

*Nat. Commun. 10.1 p. 3660. 2019*

### Birth, expansion, and death of VCY-containing palindromes on the human Y chromosome

Wentao Shi, Andrea Massaia, Sandra Louzada, Juliet Handsaker, William Chow, Shane McCarthy, Joanna Collins, Pille Hallast, Kerstin Howe, Deanna M Church, Fengtang Yang, Yali Xue, and Chris Tyler-Smith

*Genome Biol. 20.1 p. 207. 2019*

### Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly

Tatsiana Aneichyk, William T Hendriks, Rachita Yadav, David Shin, Dadi Gao, Christine A Vaine, Ryan L Collins, Aloysius Domingo, Benjamin Currall, Alexei Stortchevoi, Trisha Multhaupt-Buell, Ellen B Penney, Lilian Cruz, Jyotsna Dhakal, Harrison Brand, Carrie Hanscom, Caroline Antolik, Marisela Dy, Ashok Ragavendran, Jason Underwood, Stuart Cantsilieris, Katherine M Munson, Evan E Eichler, Patrick Acuña, Criscely Go, R Dominic G Jamora, Raymond L Rosales, Deanna M Church, Stephen R Williams, Sarah Garcia, Christine Klein, Ulrich Müller, Kirk C Wilhelmsen, H T Marc Timmers, Yechiam Sapir, Brian J Wainger, Daniel Henderson, Naoto Ito, Neil Weisenfeld, David Jaffe, Nutan Sharma, Xandra O Breakefield, Laurie J Ozelius, D Christopher Bragg, and Michael E Talkowski

*Cell 172.5 897–909.e21. Cold Spring Harbor Laboratory, 2018*

## Genomes for all

Deanna M Church

*Nat. Biotechnol.* 36.9 pp. 815–816. 2018

## Reference quality assembly of the 3.5-Gb genome of *Capsicum annuum* from a single linked-read library

Amanda M Hulse-Kemp, Shamoni Maheshwari, Kevin Stoffel, Theresa A Hill, David Jaffe, Stephen R Williams, Neil Weisenfeld, Srividya Ramakrishnan, Vijay Kumar, Preyas Shah, Michael C Schatz, Deanna M Church, and Allen Van Deynze

*Hortic Res* 5 p. 4. 2018

## Principles and Recommendations for Standardizing the Use of the Next-Generation Sequencing Variant File in Clinical Settings

Ira M Lubin, Nazneen Aziz, Lawrence J Babb, Dennis Ballinger, Himani Bisht, Deanna M Church, Shaun Cordes, Karen Eilbeck, Fiona Hyland, Lisa Kalman, Melissa Landrum, Edward R Lockhart, Donna Maglott, Gabor Marth, John D Pfeifer, Heidi L Rehm, Somak Roy, Zivana Tezak, Rebecca Truty, Mollie Ullman-Cullere, Karl V Voelkerding, Elizabeth A Worthey, Alexander W Zaranek, and Justin M Zook

*J. Mol. Diagn.* 19.3 pp. 417–426. 2017

## Evaluation of GRCh38 and de novo haploid genome assemblies demonstrates the enduring quality of the reference assembly

Valerie A Schneider, Tina Graves-Lindsay, Kerstin Howe, Nathan Bouk, Hsiu-Chuan Chen, Paul A Kitts, Terence D Murphy, Kim D Pruitt, Françoise Thibaud-Nissen, Derek Albracht, Robert S Fulton, Milinn Kremitzki, Vincent Magrini, Chris Markovic, Sean McGrath, Karyn Meltz Steinberg, Kate Auger, William Chow, Joanna Collins, Glenn Harden, Timothy Hubbard, Sarah Pelan, Jared T Simpson, Glen Threadgold, James Torrance, Jonathan M Wood, Laura Clarke, Sergey Koren, Matthew Boitano, Paul Peluso, Heng Li, Chen-Shan Chin, Adam M Phillippy, Richard Durbin, Richard K Wilson, Paul Flicek, Evan E Eichler, and Deanna M Church

*Genome Res.* 27.5 pp. 849–864. 2017

## Building and Improving Reference Genome Assemblies

K Meltz Steinberg, V A Schneider, C Alkan, M J Montague, W C Warren, D M Church, and R K Wilson

*Proc. IEEE* 105.3 pp. 422–435. 2017

## Direct determination of diploid genome sequences

Neil I Weisenfeld, Vijay Kumar, Preyas Shah, Deanna M Church, and David B Jaffe

*Genome Res.* 27.5 pp. 757–767. 2017

## A variant by any name: quantifying annotation discordance across tools and clinical databases

Jennifer L Yen, Sarah Garcia, Aldrin Montana, Jason Harris, Stephen Chervitz, Massimo Morra, John West, Richard Chen, and Deanna M Church

*Genome Med.* 9.1 p. 7. 2017

## Alternate-locus aware variant calling in whole genome sequencing

Marten Jäger, Max Schubach, Tomasz Zemojtel, Knut Reinert, Deanna M Church, and Peter N Robinson

*Genome Med.* 8.1 p. 130. 2016

## A global reference for human genetic variation

Adam Auton et al.

*Nature* 526.7571 pp. 68–74. 2015

## Extending reference assembly models

Deanna M Church, Valerie a Schneider, Karyn Meltz Steinberg, Michael C Schatz, Aaron R Quinlan, Chen-Shan Chin, Paul a Kitts, Bronwen Aken, Gabor T Marth, Michael M Hoffman, and Javier Herrero

*Genome Biol.* 16.13 pp. 2–6. 2015

## Good laboratory practice for clinical next-generation sequencing informatics pipelines

Amy S Gargis, Lisa Kalman, David P Bick, Cristina da Silva, David P Dimmock, Birgit H Funke, Sivakumar Gowrisankar, Madhuri R Hegde, Shashikant Kulkarni, Christopher E Mason, Rakesh Nagarajan, Karl V Voelkerding, Elizabeth a Worthey, Nazneen Aziz, John Barnes, Sarah F Bennett, Himani Bisht,

Deanna M Church, Zoya Dimitrova, Shaw R Gargis, Nabil Hafez, Tina Hambuch, Fiona C L Hyland, Ruth Ann Luna, Duncan MacCannell, Tobias Mann, Megan R McCluskey, Timothy K McDaniel, Lilia M Ganova-Raeva, Heidi L Rehm, Jeffrey Reid, David S Campo, Richard B Resnick, Perry G Ridge, Marc L Salit, Pavel Skums, Lee-Jun C Wong, Barbara a Zehnbaauer, Justin M Zook, and Ira M Lubin  
*Nat. Biotechnol.* 33.7 pp. 689–693. 2015

#### **Assembly: a resource for assembled genomes at NCBI**

P A Kitts, D M Church, F Thibaud-Nissen, J Choi, V Hem, V Sapojnikov, R G Smith, T Tatusova, C Xiang, A Zherikov, M DiCuccio, T D Murphy, K D Pruitt, and A Kimchi  
*Nucleic Acids Res.* Pp. 1–8. 2015

#### **Achieving high-sensitivity for clinical applications using augmented exome sequencing**

Anil Patwardhan, Jason Harris, Nan Leng, Gabor Bartha, Deanna M Church, Shujun Luo, Christian Haudenschild, Mark Pratt, Justin Zook, Marc Salit, Jeanie Tirsch, Massimo Morra, Stephen Chervitz, Ming Li, Michael Clark, Sarah Garcia, Gemma Chandratillake, Scott Kirk, Euan Ashley, Michael Snyder, Russ Altman, Carlos Bustamante, Atul J Butte, John West, and Richard Chen  
*Genome Med.* 7.1 p. 71. *Genome Medicine*, 2015

#### **ClinVar: public archive of relationships among sequence variation and human phenotype**

Melissa J Landrum, Jennifer M Lee, George R Riley, Wonhee Jang, Wendy S Rubinstein, Deanna M Church, and Donna R Maglott  
*Nucleic Acids Res.* 42.Database issue pp. D980–5. 2014

#### **Database resources of the National Center for Biotechnology Information**

NCBI Resource Coordinators  
*Nucleic Acids Res.* 42.Database issue pp. D7–17. 2014

#### **Single haplotype assembly of the human genome from a hydatidiform mole**

Karyn Meltz Steinberg, Valerie A Schneider, Tina A Graves-Lindsay, Robert S Fulton, Richa Agarwala, John Huddleston, Sergey A Shiryev, Aleksandr Morgulis, Urvashi Surti, Wesley C Warren, Deanna M Church, Evan E Eichler, and Richard K Wilson  
*Genome Res.* 24.12 pp. 2066–2076. 2014

#### **DbVar and DGVa: public archives for genomic structural variation**

Ilkka Lappalainen, John Lopez, Lisa Skipper, Timothy Hefferon, J Dylan Spalding, John Garner, Chao Chen, Michael Maguire, Matt Corbett, George Zhou, Justin Paschall, Victor Ananiev, Paul Flicek, and Deanna M Church  
*Nucleic Acids Res.* 41.Database issue pp. D936–41. 2013

#### **Clone DB: an integrated NCBI resource for clone-associated data**

Valerie A Schneider, Hsiu-Chuan Chen, Cliff Clausen, Peter A Meric, Zhigang Zhou, Nathan Bouk, Nora Husain, Donna R Maglott, and Deanna M Church  
*Nucleic Acids Res.* 41.Database issue pp. D1070–8. 2013

#### **Towards an evidence-based process for the clinical interpretation of copy number variation**

E R Riggs, D M Church, K Hanson, V L Horner, E B Kaminsky, R M Kuhn, K E Wain, E S Williams, S Aradhya, H M Kearney, D H Ledbetter, S T South, E C Thorland, and C L Martin  
*Clin. Genet.* 81(5).November pp. 403–412. 2012

#### **Online resources for genomic structural variation**

Tam P Sneddon and Deanna M Church  
*Methods Mol. Biol.* 838 pp. 273–289. 2012

#### **Modernizing Reference Genome Assemblies**

Deanna M Church, Valerie a Schneider, Tina Graves, Katherine Auger, Fiona Cunningham, Nathan Bouk, Hsiu-Chuan Chen, Richa Agarwala, William M McLaren, Graham R S Ritchie, Derek Albracht, Milinn Kremitzki, Susan Rock, Holland Kotkiewicz, Colin Kremitzki, Aye Wollam, Lee Trani, Lucinda Fulton, Robert Fulton, Lucy Matthews, Siobhan Whitehead, Will Chow, James Torrance, Matthew Dunn, Glenn Harden, Glen Threadgold, Jonathan Wood, Joanna Collins, Paul Heath, Guy Griffiths, Sarah Pelan, Darren Grafham, Evan E. Eichler, George Weinstock, Elaine R Mardis, Richard K Wilson, Kerstin Howe, Paul Flicek, and Tim Hubbard  
*PLoS Biol.* 9.7 e1001091. 2011

### An evidence-based approach to establish the functional and clinical significance of copy number variants in intellectual and developmental disabilities

Erin B Kaminsky, Vineith Kaul, Justin Paschall, Deanna M Church, Brian Bunke, Dawn Kunig, Daniel Moreno-De-Luca, Andres Moreno-De-Luca, Jennifer G Mulle, Stephen T Warren, Gabriele Richard, John G Compton, Amy E Fuller, Troy J Gliem, Shuwen Huang, Morag N Collinson, Sarah J Beal, Todd Ackley, Diane L Pickering, Denae M Golden, Emily Aston, Heidi Whitby, Shashirekha Shetty, Michael R Rossi, M Katharine Rudd, Sarah T South, Arthur R Brothman, Warren G Sanger, Ramaswamy K Iyer, John a Crolla, Erik C Thorland, Swaroop Aradhya, David H Ledbetter, and Christa L Martin

*Genet. Med.* 13.9 pp. 777–784. 2011

### Public data archives for genomic structural variation

Deanna M Church, Ilkka Lappalainen, Tam P Sneddon, Jonathan Hinton, John Lopez, John Garner, Justin Paschall, Michael Dicuccio, Stephen W Scherer, Lars Feuk, and Paul Flicek

*Nat. Genet.* 42.10 pp. 813–814. 2010

### Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies

David T Miller, Margaret P Adam, Swaroop Aradhya, Leslie G Biesecker, Arthur R Brothman, Nigel P Carter, Deanna M Church, John a Crolla, Evan E Eichler, Charles J Epstein, W Andrew Faucett, Lars Feuk, Jan M Friedman, Ada Hamosh, Laird Jackson, Erin B Kaminsky, Klaas Kok, Ian D Krantz, Robert M Kuhn, Charles Lee, James M Ostell, Carla Rosenberg, Stephen W Scherer, Nancy B Spinner, Dimitri J Stavropoulos, James H Tepperberg, Erik C Thorland, Joris R Vermeesch, Darrel J Waggoner, Michael S Watson, Christa Lese Martin, and David H Ledbetter

*Am. J. Hum. Genet.* 86.5 pp. 749–764. *The American Society of Human Genetics*, 2010

### Completing the map of human genetic variation

David Altshuler, Anne M Bowcock, Lisa D Brooks, Nigel P Carter, M Church, Adam Felsenfeld, Mark Guyer, Charles Lee, James R Lupski, C James, Jonathan K Pritchard, Jonathan Sebat, Stephen T Sherry, David Valle, and Robert H Waterston

*Nature* 447.7141 pp. 161–165. 2009

### Lineage-specific biology revealed by a finished genome assembly of the mouse

Deanna M Church, Leo Goodstadt, Ladeana W Hillier, Michael C Zody, Steve Goldstein, Xinwe She, Carol J Bult, Richa Agarwala, Joshua L Cherry, Michael DiCuccio, Wratko Hlavina, Yuri Kapustin, Peter Meric, Donna Maglott, Zoë Birtle, Ana C Marques, Tina Graves, Shiguo Zhou, Brian Teague, Konstantinos Potamiosis, Christopher Churas, Michael Place, Jill Herschleb, Ron Runnheim, Daniel Forrest, James Amos-Landgraf, David C Schwartz, Ze Cheng, Kerstin Lindblad-Toh, Evan E Eichler, and Chris P Ponting

*PLoS Biol.* 7.5 e1000112. 2009

### Back to Bermuda: how is science best served?

Deanna M Church and Ladeana W Hillier

*Genome Biol.* 10.4 p. 105. 2009

### Database resources of the National Center for Biotechnology Information

Eric W Sayers, Tanya Barrett, Dennis a Benson, Stephen H Bryant, Kathi Canese, Vyacheslav Chetvernin, Deanna M Church, Michael DiCuccio, Ron Edgar, Scott Federhen, Michael Feolo, Lewis Y Geer, Wolfgang Helmberg, Yuri Kapustin, David Landsman, David J Lipman, Thomas L Madden, Donna R Maglott, Vadim Miller, Ilene Mizrahi, James Ostell, Kim D Pruitt, Gregory D Schuler, Edwin Sequeira, Stephen T Sherry, Martin Shumway, Karl Sirotkin, Alexandre Souvorov, Grigory Starchenko, Tatiana a Tatusova, Lukas Wagner, Eugene Yaschenko, and Jian Ye

*Nucleic Acids Res.* 37.Database issue pp. D5–15. 2009

### Mouse segmental duplication and copy number variation

Xinwei She, Ze Cheng, Sebastian Zöllner, Deanna M Church, and Evan E Eichler

*Nat. Genet.* 40.7 pp. 909–914. 2008

### Candidate single nucleotide polymorphism selection using publicly available tools: a guide for epidemiologists

Parveen Bhatti, Deanna M Church, Joni L Rutter, Jeffery P Struewing, and Alice J Sigurdson

*Am. J. Epidemiol.* 164.8 pp. 794–804. 2006

### Database resources of the National Center for Biotechnology Information

David L Wheeler, Tanya Barrett, Dennis a Benson, Stephen H Bryant, Kathi Canese, Vyacheslav Chetvernin, Deanna M Church, Michael DiCuccio, Ron Edgar, Scott Federhen, Lewis Y Geer, Wolfgang Helmberg, Yuri Kapustin, David L Kenton, Oleg Khovayko, David J Lipman, Thomas L Madden, Donna R Maglott, James Ostell, Kim D Pruitt, Gregory D Schuler, Lynn M Schriml, Edwin Sequeira, Stephen T Sherry, Karl Sirotkin, Alexandre Souvorov, Grigory Starchenko, Tugba O Suzek, Roman Tatusov, Tatiana a Tatusova, Lukas Wagner, and Eugene Yaschenko

*Nucleic Acids Res. 34.Database issue pp. D173–80. 2006*

### A genome-wide comparison of recent chimpanzee and human segmental duplications

Ze Cheng, Mario Ventura, Xinwei She, Philipp Khaitovich, Tina Graves, Kazutoyo Osoegawa, Deanna Church, Pieter DeJong, Richard K Wilson, Svante Pääbo, Mariano Rocchi, and Evan E Eichler

*Nature 437.7055 pp. 88–93. 2005*

### Analysis of Segmental Duplications and Genome Assembly in the Mouse

Jeffrey A Bailey, Deanna M Church, Mario Ventura, Mariano Rocchi, and Evan E Eichler

*Genome Res. 14.216 pp. 789–801. 2004*

### Shotgun sequence assembly and recent segmental duplication within the human genome

Xinwei She, Zhaoshi Jiang, Royden A Clark, Ge Liu, Ze Cheng, Eray Tuzun, Deanna M Church, Granger Sutton, Aaron L Halpern, and Evan E Eichler

*Nature 431.7011 pp. 927–930. 2004*

### Database resources of the National Center for Biotechnology Information: update

David L Wheeler, Deanna M Church, Ron Edgar, Scott Federhen, Wolfgang Helmberg, Thomas L Madden, Joan U Pontius, Gregory D Schuler, Lynn M Schriml, Edwin Sequeira, Tugba O Suzek, Tatiana a Tatusova, and Lukas Wagner

*Nucleic Acids Res. 32.Database issue pp. D35–40. 2004*

### Connecting Sequence and Biology in the Laboratory Mouse

Richard M Baldarelli, David P Hill, Judith A Blake, Jun Adachi, Masaaki Furuno, Dirck Bradt, Lori E Corbani, Sharon Cousins, Kenneth S Frazer, Dong Qi, Longlong Yang, Sridhar Ramachandran, Deborah Reed, Yunxia Zhu, Takeya Kasukawa, Martin Ringwald, Benjamin L King, Lois J Maltais, Louise M Mckenzie, Lynn M Schriml, Donna Maglott, Deanna M Church, Kim Pruitt, Janan T Eppig, Joel E Richardson, Jim A Kadin, and Carol J Bult

*Genome Res. 13.6b pp. 1505–1519. 2003*

### Cross-Species Sequence Comparisons : A Review of Methods and Available Resources

Kelly A Frazer, Laura Elnitski, Deanna M Church, Inna Dubchak, and Ross C Hardison

*Genome Res. 13.1 pp. 1–12. 2003*

### Database resources of the National Center for Biotechnology

David L Wheeler, Deanna M Church, Scott Federhen, Alex E Lash, Thomas L Madden, Joan U Pontius, Gregory D Schuler, Lynn M Schriml, Edwin Sequeira, Tatiana A Tatusova, and Lukas Wagner

*Nucleic Acids Res. 31.1 pp. 28–33. 2003*

### Initial sequencing and comparative analysis of the mouse genome

Robert H Waterston et al.

*Nature 420.6915 pp. 520–562. 2002*

### Database resources of the National Center for Biotechnology Information: 2002 update

David L Wheeler, Deanna M Church, Alex E Lash, Detlef D Leipe, Thomas L Madden, Joan U Pontius, Gregory D Schuler, Lynn M Schriml, Tatiana a Tatusova, Lukas Wagner, and Barbara a Rapp

*Nucleic Acids Res. 30.1 pp. 13–16. 2002*

### A radiation hybrid map of mouse genes

T J Hudson, D M Church, S Greenaway, H Nguyen, A Cook, R G Steen, W J Van Etten, a B Castle, M a Strivens, P Trickett, C Heuston, C Davison, A Southwell, R Hardisty, A Varela-Carver, a R Haynes, P Rodriguez-Tome, H Doi, M S Ko, J Pontius, L Schriml, L Wagner, D Maglott, S D Brown, E S Lander, G Schuler, and P Denny

*Nat. Genet. 29.2 pp. 201–205. 2001*

### Initial sequencing and analysis of the human genome



E S Lander et al.  
*Nature* 409.6822 pp. 860–921. 2001

Generation of RCAS vectors useful for functional genomic analyses  
S K Loftus, D M Larson, D Watkins-Chow, D M Church, and W J Pavan  
*DNA Res.* 8.5 pp. 221–226. 2001

Spidey : A Tool for mRNA-to-Genomic Alignments  
Sarah J Wheelan, Deanna M Church, and James M Ostell  
*Genome Res.* 11.11 pp. 1952–1957. 2001

Rapid generation of nested chromosomal deletions on mouse chromosome 2  
D F LePage, D M Church, E Millie, T J Hassold, and R a Conlon  
*Proc. Natl. Acad. Sci. U. S. A.* 97.19 pp. 10471–10476. 2000

Gene identification by exon amplification  
D M Church and A J Buckler  
*Methods Enzymol.* 303 pp. 83–99. 1999

Transcript mapping of the human chromosome 11q12-q13.1 gene-rich region identifies several newly described conserved genes  
P R Cooper, N J Nowak, M J Higgins, D M Church, and T B Shows  
*Genomics* 49.3 pp. 419–429. 1998

A High-Resolution Physical and Transcript Map of the Cri du Chat A High-Resolution Physical and Transcript Map of the Cri du Chat Region of Human Chromosome 5p  
Deanna M Church, Julie Yang, Maureen Bocian, Rita Shiang, and John J Wasmuth  
*Genome Res.* 7.8 pp. 787–801. 1997

A Radiation Hybrid Map of Human Chromosome 5 with Integration of Cytogenetic , Genetic , and Transcript Maps  
John D Mcpherson, Barbara Apostol, Caryn B Wagner-mcpherson, Simin Hakim, Richard G Del Mas-tro, Naeema Aziz, Elizabeth Baer, Genalyn Gonzales, Mary Carol Krane, Rachelle Markovich, Peter Masny, Miguel Ortega, John Vu, Marco Vujicic, Deanna M Church, Allan Segal, Deborah L Grady, Robert K Moyzis, M Anne Spence, Michael Lovett, and John J Wasmuth  
*Genome Res.* 7.9 pp. 897–909. 1997

Exon trapping and sequence-based methods of gene finding in transcript mapping of human 4p16.3  
I Pribill, G T Barnes, J Chen, D Church, A Buckler, S Baxendale, G P Bates, H Lehrach, M J Gusella, M P Duyao, C M Ambrose, J F Gusella, and M E MacDonald  
*Somat. Cell Mol. Genet.* 23.6 pp. 413–427. 1997

The Ras GTPase-activating-protein-related human protein IQGAP2 harbors a potential actin binding domain and interacts with calmodulin and Rho family GTPases  
S Brill, S Li, C W Lyman, D M Church, J J Wasmuth, L Weissbach, A Bernards, and a J Snijders  
*Mol. Cell. Biol.* 16.9 pp. 4869–4878. 1996

Identification and characterization of two novel tetratricopeptide repeat-containing genes  
A E Murthy, A Bernards, D Church, J Wasmuth, and J F Gusella  
*DNA Cell Biol.* 15.9 pp. 727–735. 1996

Molecular definition of deletions of different segments of distal 5p that result in distinct phenotypic features  
D M Church, U Bengtsson, K V Nielsen, J J Wasmuth, and E Niebuhr  
*Am. J. Hum. Genet.* 56.5 pp. 1162–1172. 1995

Isolation of genes from complex sources of mammalian genomic DNA using exon amplification  
Deanna M Church, Christy J Stotler, Joni L Rutter, Jill R Murrell, James A Trofatter, and Alan J Buckler  
*Nat. Genet.* 6.1 pp. 98–105. 1994

Mutations in the transmembrane domain of FGFR3 cause the most common genetic form of dwarfism, achondroplasia  
R Shiang, L M Thompson, Y Z Zhu, D M Church, T J Fielder, M Bocian, S T Winokur, and J J Wasmuth

Cell 78.2 pp. 335–342. 1994

**Identification of human chromosome 9 specific genes using exon amplification**

D M Church, L T Banks, a C Rogers, S L Graw, D E Housman, J F Gusella, and a J Buckler

Hum. Mol. Genet. 2.11 pp. 1915–1920. 1993

**A gene from chromosome 4p16.3 with similarity to a superfamily of transporter proteins**

Mabel P Duyao, Sherryl A M Tayler, Alan J Buckler, Christine M Ambrose, Carol Lin, Nicolet Groot, Deanna Church, Glenn Barnes, John J Wasmuth, David E Housman, Marcy E MacDonald, and James F Gusella

Hum. Mol. Genet. 2.6 pp. 673–676. 1993

**Efficiency and specificity of gene isolation by exon amplification**

M A North, P Sanseau, A J Buckler, D Church, A Jackson, K Patel, J Trowsdale, and H Lehrach

Mamm. Genome 4.9 pp. 466–474. 1993

**Molecular basis of myotonic dystrophy: expansion of a trinucleotide (CTG) repeat at the 3' end of a transcript encoding a protein kinase family member**

J D Brook, M E McCurrach, H G Harley, a J Buckler, D Church, H Aburatani, K Hunter, V P Stanton, J P Thirion, and T Hudson

Cell 69.2 p. 385. 1992

**Cloning of the a-adducin gene from the Huntington's disease candidate region of chromosome 4 by exon amplification**

S A M Taylor, R G Snell, a J Buckler, C Ambrose, M Duyao, D Church, C S Lin, M Altherr, G P Bates, N Groot, G Barnes, D J Shaw, H Lehrach, J J Wasmuch, P S Harper, D E Housman, M E MacDonald, and J F Gusella

Nat. Genet. 2.3 pp. 223–227. 1992

**Protein synthesis requirements for nuclear division, cytokinesis, and cell separation in *Saccharomyces cerevisiae***

D J Burke and D Church

Mol. Cell. Biol. 11.7 pp. 3691–3698. 1991

## Preprints

**Charting the emergent organotypic landscape of the mammalian gut endoderm at single-cell resolution**

Sonja Nowotschin, Manu Setty, Ying-Yi Kuo, Vincent Lui, Vidur Garg, Roshan Sharma, Claire S Simon, Nestor Saiz, Rui Gardner, Stéphane C Boutet, Deanna M Church, Pamela A Hoodless, Anna-Katerina Hadjantonakis, and Dana Pe'er

bioRxiv p. 471078. 2018

**Mutation detection in thousands of acute myeloid leukemia cells using single cell RNA-sequencing**

Allegra A Petti, Stephen R Williams, Christopher A Miller, Ian T Fiddes, Sridhar N Srivatsan, David Y Chen, Catrina C Fronick, Robert S Fulton, Deanna M Church, and Timothy J Ley

bioRxiv p. 434746. 2018

**Multi-platform discovery of haplotype-resolved structural variation in human genomes**

Mark J P Chaisson, Ashley D Sanders, Xuefang Zhao, Ankit Malhotra, David Porubsky, Tobias Rausch, Eugene J Gardner, Oscar Rodriguez, Li Guo, Ryan L Collins, Xian Fan, Jia Wen, Robert E Handsaker, Susan Fairley, Zev N Kronenberg, Xiangmeng Kong, Fereydoun Hormozdiani, Dillon Lee, Aaron M Wenger, Alex Hastie, Danny Antaki, Peter Audano, Harrison Brand, Stuart Cantsilieris, Han Cao, Eliza Cerveira, Chong Chen, Xintong Chen, Chen-Shan Chin, Zechen Chong, Nelson T Chuang, Deanna M Church, Laura Clarke, Andrew Farrell, Joey Flores, Timur Galeev, David Gorkin, Madhusudan Gural, Victor Guryev, William Haynes Heaton, Jonas Korlach, Sushant Kumar, Jee Young Kwon, Jong Eun Lee, Joyce Lee, Wan-Ping Lee, Sau Peng Lee, Patrick Marks, Karine Viaud-Martinez, Sascha Meiers, Katherine M Munson, Fabio Navarro, Bradley J Nelson, Conor Nodzak, Amina Noor, Sofia Kyriazopoulou-Panagiotopoulou, Andy Pang, Yunjiang Qiu, Gabriel Rosanio, Mallory Ryan, Adrian Stütz, Diana C J Spierings, Alistair Ward, Annemarie E Welch, Ming Xiao, Wei Xu, Chengsheng Zhang, Qihui Zhu, Xiangqun Zheng-Bradley, Goo Jun, Li Ding, Chong Lek Koh, Bing Ren, Paul Flicek, Ken

Chen, Mark B Gerstein, Pui-Yan Kwok, Peter M Lansdorp, Gabor Marth, Jonathan Sebat, Xinghua Shi, Ali Bashir, Kai Ye, Scott E Devine, Michael Talkowski, Ryan E Mills, Tobias Marschall, Jan Korb, Evan E Eichler, and Charles Lee  
*bioRxiv p. 193144. 2017*

#### **Linked-Read sequencing resolves complex structural variants**

Sarah Garcia, Stephen Williams, Andrew Wei Xu, Jill Herschleb, Patrick Marks, David Stafford, and Deanna M Church  
*bioRxiv p. 231662. 2017*

#### **Resolving the Full Spectrum of Human Genome Variation using Linked-Reads**

Patrick Marks, Sarah Garcia, Alvaro Martinez Barrio, Kamila Belhocine, Jorge Bernate, Rajiv Bhargava, Keith Bjornson, Claudia Catalanotti, Josh Delaney, Adrian Fehr, Brendan Galvin, Haynes Heaton, Jill Herschleb, Christopher Hindson, Esty Holt, Cassandra B Jabara, Susanna Jett, Nikka Keivanfar, Sofia Kyriazopoulou-Panagiotopoulou, Monkol Lek, Bill Lin, Adam Lowe, Shazia Mahamdallie, Shamoni Maheshwari, Tony Makarewicz, Jamie Marshall, Francesca Meschi, Chris O'keefe, Heather Ordonez, Pranav Patel, Andrew Price, Ariel Royall, Elise Ruark, Sheila Seal, Michael Schnall-Levin, Preyas Shah, Stephen Williams, Indira Wu, Andrew Wei Xu, Nazneen Rahman, Daniel MacArthur, and Deanna M Church  
*bioRxiv p. 230946. 2017*

#### **Improved de novo Genome Assembly: Linked-Read Sequencing Combined with Optical Mapping Produce a High Quality Mammalian Genome at Relatively Low Cost**

David W Mohr, Ahmed Naguib, Neil Weisenfeld, Vijay Kumar, Preyas Shah, Deanna M Church, David Jaffe, and Alan F Scott  
*bioRxiv p. 128348. 2017*