DeannaMChurch

Genomics, Technology and Bioinformatics Leader

contact

2241 Edgewood Dr.
Boulder, CO 80304
USA

(301) 233 2991

deanna.church@gmail.com

skills

Leadership, Team Player, Project Management (Jira), Curiosity & Iniative

genomics

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 make genomics and technology accessible to all.

experience

2019–present **Inscripta** Boulder, Colorado
Senior Director, Applications Mammalian
Leading an interdisciplinary team of top scientists in demonstrating the power of genome engineering to open new insights in biology.

2016–2018 **10x Genomics** Pleasant, California
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 heterogeneticy and immune profiling.

2014-2016 **Personalis, Inc** Menlo Park, CA
Senior Director of Genomics and Content
Led a team developing novel genome analysis algorthms and pipelines. Led the development of knowledge bases built using automated data processing coupled with expert data curation to support genome interpretation.

2001-2014 **National Center for Biotechnology Information, NIH** Bethesda, MA
Staff Scientist
Led a team of biologists and software developers in developing databases and tools to make genomic data accessible to the research and clinical communities. Participated in numerous international collaborations including helping to found the Genome Reference Consortium, the group tasked with maintaining the human reference assembly.

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 Coordinated mouse genome resources.

1997-1999 **Laboratory of Janet Rossant, Mt. Sinai Hospital** Toronto, Ontario
Postdoctoral fellow Applying genomics methods to understanding mouse developmental biology

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

additional training

2010	**Client-Side Web Development**	University of Illinois (O'Reilly)
2009	**XSLT/XPATH: Introduction and Syntax**	Mulberry Technologies
2007	**Introduction to XML**	University of Illinois (O'Reilly)

advisory boards and professional service

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

awards

2012 **Special Service Award** — National Library of Medicine
For extraordinary effort in providing access to the 1000 genomes project data.

1997-1999 **Human Frontiers Postdoctoral Fellowship** — Mt. Sinai Hospital
Awarded to early career scientist to enable them to broaden their skills in a new country.

1986-1990 **Echols Scholar** — University of Virginia
Awarded to select undergraduates who exhibit academic independence.

presentations

Selected highlights from over 50 presentations.

2019-Nov	**Rosalind Franklin Society** Invited Speaker, "The impact of technology on our view of biology"	Philadelphia, PA
2018-Feb	**Beckman Symposium, Technology Innovation and Human Genomics** Invited Speaker, "The impact of technology on our view of biology"	Palo Alto, CA
2017-Nov	**Australasian Genomic Technologies Association Annual Meeting** Keynote, "High resolution biology with 10x"	Hobart, AUS
2017-Nov	**Australasian Genomic Technologies Association Annual Meeting** Keynote, "The impact of technology on biology"	Hobart, AUS
2017-Apr	**NGS Barcelona** Keynote, "Advancing genomics drop by drop"	Barcelona, SP
2017-Feb	**Advance in Genomics and Biotechnology** Abstract Selected, "More complete genome analysis"	Hollywood, FL
2017-Jan	**Plant and Animal Genome Conference** Invited Speaker, "Quantitative profiling of large and complex populations by single cell RNA-sequencing"	San Diego, CA
2016-Oct	**American Society of Human Genetics** Abstract Selected, "De novo assembly of individual human haplotypes from diploid samples"	Vancouver, CA
2016-Sep	**Genome Informatics** Abstract Selected, "Improving genome analysis using Linked-Reads"	Hinxton, UK
2016-Aug	**Cancer Genomics Consortium** Invited Speaker, "Complete genome analysis"	Denver, CO
2016-Feb	**Stanford Big Data in Genomics** Invited Speaker, "Truly personalized genomics"	Palo Alto, CA
2015-Oct	**Genome informatics** Invited Speaker, "The Rumsfeldian challenge of developing clinical sequencing tests"	Cold Spring Harbor, NY
2015-Mar	**American College of Medical Genetics Annual Meeting** Invited Speaker, "Technical limitations of variant identification and annotation"	Salt Lake City, UT
2014-Nov	**Biological Data Sciences** Keynote Speaker, "Analog reporting in a digital age"	Cold Spring Harbor, NY
2014-Oct	**American Society of Human Genetics** Abstract Selected, "The impact of GRCh38 on clinical sequencing"	San Diego, CA
2013-Oct	**American Society of Human Genetics** Invited Speaker, "Navigating clinical genomic resources at NCBI"	Boston, MA
2013- May	**Sequencing Finishing and Analysis in the Future** Keynote Address, "Keep calm and carry on sequencing"	Santa Fe, NM
2012-Nov	**American Society of Human Genetics** Invited Speaker, "Improving the accuracy of variant identification"	San Francisco, CA
2012-Apr	**Generic Model Organism Database Conference** Keynote Address, "Navigating genome resources at NCBI"	Washington, DC
2011-Jul	**BIO-IT APAC Conference and Expo** Keynote Speaker, "The evolution of the reference human genome"	Shenzhen, China
2011-Jan	**Plant and Animal Genome Conference** Keynote Address, "The evolution of the reference human genome"	San Diego, CA
2008-May	**Finishing in the Future** Keynote Speaker, "Great expectations: fulfilling the promises of the human genome project"	Santa Fe, NM
2006-Jun	**Human Genome Variation Society** Keynote Speaker, "The evolution of the reference human genome"	Someplace, WO

teaching

Selected courses and lectures.

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

publications

Articles

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, Fereydoun Hormozdiari, 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, Zechen 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 Bharadwaj, 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 Cristopher 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 ofCapsicum annuumfrom 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 Zehnbauer, 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 Tirch, 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 Potamousis, 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 Mizrachi, 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 Mastro, 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 Hormozdiari, 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 Gujral, 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 Korbel, 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 Bharadwaj, 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*