# DeannaMChurch Genomics, Technology and Bioinformatics Leader

contact

focus

550 Iris St. Redwood City, CA 94062 USA

Leading interdisciplinary teams to make genomics and technology accessible to all.

(301) 233 2991

**experience** 

deanna.church@gmail.com

Leadership, Team Player,

2016-Present 10x Genomics Senior Director of Applications Pleasant, California

skills

Leads an interdisciplinary team of top rate 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.

Project Management (Jira), Curiosity & Iniative

2014-2016 Personalis, Inc Menlo Park, CA

genomics

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.

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

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.

bioinformatics

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

1990-1992 MIT and MGH

Boston, MA

Laboratory Technician

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

languages english, mother tongue

### education

1999-2001	National Center for Biotechnology Information, NIH Postdoctoral fellow Coordinated mouse genome re	Bethesda, MD sources.
1997-1999	Laboratory of Janet Rossant, Mt. Sinai Hospital Postdoctoral fellow Applying genomics methods to velopmental biology	Toronto, Ontario understanding mouse de-
1992-1997	<b>PhD</b> of Biological Sciences  Phenotype-genotype correlation in the Cri-du-Chat	The University of California, Irvine region of chromosome 5
1986-1990	<b>Bachelor</b> of Liberal Arts Research project focused on cell cycle regulation us	University of Virginia sing 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 <b>Genome Quebec</b>	Scientific Advisory Board
2018-present American Association for Cancer Re	search Member
2012-present <b>Ensembl</b>	Scientific Advisory Board
2014-2016 American College of Medical Genetic	Member Member
2013-2016 The Genome Analysis Center/Earlha	m Institute Scientific Advisory Board
2010-2014 <b>Database of Genomic Variants</b>	Scientific Advisory Board
2010-present American Society of Human Genetic	<b>S</b> Member
2008-2013 Advances in Genome Biology and Te	<b>chnology</b> Organizing Committee Member
2008-2013 International Standards Cytogenomi	<b>c Arrays</b> Committee Member
2008-2012 International Mammalian Genome S	ociety Nomenclature Committee Member
2004-2012 International Mammalian Genome S	ociety Member
2006-2011 European Conditional Mouse Mutage	enesis 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	0 genomes project data.
1997-1999	Human Frontiers Postdoctoral Fellowship	Mt. Sinai Hospital
	Awarded to early career scientist to enable them to brocountry.	paden their skills in a new
1986-1990	<b>Echols Scholar</b> Awarded to select undergraduates who exhibit acader	University of Virginia mic independence.

### presentations

Selected highlights from over 50 presentations.

2018-Feb	Beckman Symposium, Technology Innovation and Human Genomics	Palo Alto, CA
	Invited Speaker, "The impact of technology on our view of biology"	
2017-Nov	Australasian Genomic Technologies Association Annual Meeting	Hobart, AUS
	Keynote, "High resolution biology with 10x"	

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

# 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 bioinfor-

matics.

### publications

#### **Articles**

#### 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, 2018

Deanna M Church

Genomes for all

Nat. Biotechnol. 36 p. 815. Nature Publishing Group, a division of Macmillan Publishers Limited. All Rights Re-

#### Reference quality assembly of the 3.5-Gb genome of Capsicum annum from a single linkedread 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

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

Paul A Kitts, Deanna M Church, Françoise Thibaud-Nissen, Jinna Choi, Vichet Hem, Victor Sapojnikov, Robert G Smith, Tatiana Tatusova, Charlie Xiang, Andrey Zherikov, Michael DiCuccio, Terence D Murphy, Kim D Pruitt, and Avi Kimchi

Nucleic Acids Res. 44.D1 pp. D73-80. 2016

#### 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, Javier Herrero, M Lisandra Zepeda Mendoza, Richard Durbin, and Paul Flicek

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

#### 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

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

Ncbi Resource Coordinators

Nucleic Acids Res. 41. Database issue pp. D8-D20. 2013

#### 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

ER Riggs, DM Church, KHanson, VL Horner, EB Kaminsky, RM Kuhn, KE Wain, ES Williams, SAradhya, HM Kearney, DH Ledbetter, ST South, EC Thorland, and CL 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

### 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

ES 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

PR Cooper, NJ Nowak, MJ Higgins, DM Church, and TB 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

# A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes. The Huntington's Disease Collaborative Research Group

Cell 72.6 pp. 971-983. 1993

#### 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

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

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

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