Jay Shendure, MD, PhD

Updated December 1, 2019

Current Positions

Investigator, Howard Hughes Medical Institute
Professor, Genome Sciences, University of Washington
Director, Allen Discovery Center for Cell Lineage
Director, Brotman Baty Institute for Precision Medicine

Contact Information

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Education

- 2007 M.D., Harvard Medical School (Boston, Massachusetts)
- 2005 Ph.D. in Genetics, Harvard University (Cambridge, Massachusetts)
  Research Advisor: George M. Church
  Thesis entitled "*Multiplex Genome Sequencing and Analysis*"
- 1996 A.B., *summa cum laude* in Molecular Biology, Princeton University (Princeton, NJ)
  Research Advisor: Lee M. Silver

Professional Experience

- 2017 – present Scientific Director
  Brotman-Baty Institute for Precision Medicine
- 2017 – present Scientific Director
  Allen Discovery Center for Cell Lineage Tracing
- 2015 – present Investigator
  Howard Hughes Medical Institute
- 2015 – present Full Professor (with tenure)
  Department of Genome Sciences, University of Washington, Seattle, WA
- 2010 – present Affiliate Professor
  Division of Human Biology, Fred Hutchinson Cancer Research Center, Seattle, WA
- 2011 – 2015 Associate Professor (with tenure)
  Department of Genome Sciences, University of Washington, Seattle, WA
- 2007 – 2011 Assistant Professor
  Department of Genome Sciences, University of Washington, Seattle, WA
- 1998 – 2007 Medical Scientist Training Program (MSTP) Candidate
  Department of Genetics, Harvard Medical School, Boston, WA
- 1997 – 1998 Research Scientist
  Vaccine Division, Merck Research Laboratories, Rahway, NJ
- 1996 – 1997 Fulbright Scholar to India
  Department of Pediatrics, Sassoon General Hospital, Pune, India

Honors, Awards, Named Lectures

- 2019 Richard Lounsbery Award (for extraordinary scientific achievement in biology & medicine)
  National Academy of Sciences
- 2019 Jeffrey M. Trent Lectureship in Cancer Research
  National Human Genome Research Institute, National Institutes of Health
- 2019 Paul D. Gottlieb Distinguished Lectureship
  University of Texas, Austin
- 2019 AAAS Fellow
  American Association for the Advancement of Science
- 2018 Allan C. Wilson Memorial Lectureship
  University of California, Berkeley
- 2018 Richard and Carol Hertzberg Prize (for technology innovation)
  University of California, San Diego
- 2018 Dr. Nancy C. Andrews Physician-Scientist Lectureship
  Duke University
- 2017 British Society of Genetic Medicine Lectureship
  British Society of Genetic Medicine
- 2015 HHMI Investigator
  Howard Hughes Medical Institute
- 2014 Cell “40 under 40”
  Cell 40th Anniversary, Cell Press
- 2014 7th Annual Scripps Genomic Medicine Award
  Scripps Health
- 2014 HudsonAlpha Prize for Life Sciences
  HudsonAlpha Institute for Biotechnology
- 2013 FEDERAprijs
  Federation of Dutch Medical Scientific Societies
- 2013 NIH Director’s Pioneer Award
  National Institutes of Health
- 2012 Curt Stern Award
  American Society of Human Genetics
- 2010 Lowell Milken Young Investigator (2010-2013)
  Prostate Cancer Foundation
- 2008 Science in Medicine New Investigator Lecture
  University of Washington
- 2008 3rd Annual Tomorrow’s PIs
  Genome Technology Magazine
- 2007 James Tolbert Shipley Prize
  Harvard Medical School
- 2006 TR35 Young Innovator Award
  M.I.T. Technology Review
- 1998 Medical Science Training Program Fellowship
  National Institutes of Health

- 1996 Fulbright Scholarship
  U.S. State Department
- 1996 *summa cum laude*
  Princeton University
- 1996 Honorary Major in Anthropology
  Princeton University
- 1996 Sigma Chi Book Award for Molecular Biology Senior Thesis ("*The Genetics of Alcohol Consumption: QTLs Affecting Ethanol Consumption in Inbred Mice*")
  Princeton University
- 1996 Senior Prize for Best Thesis in Anthropology ("*Homunculi, Polyps and the Generation of Beings: Interpreting Theory Change in Biology*")
  Princeton University
- 1996 Phi Beta Kappa
  Princeton University
- 1992 National Merit Scholar
  Solon High School

Academic Consortium Leadership & Scientific Advisory Roles

- 2018 – present Allen Institute for Immunology, Scientific Advisory Board
- 2018 – present Chan Zuckerberg Initiative, Human Cell Atlas Scientific Advisory Board
- 2017 – present Advisory Committee to NIH Director (ACD), National Institutes of Health
- 2017 – present Science, Board of Reviewing Editors
- 2017 – present Allen Institute for Cell Science, Stem Cells and Gene Editing Advisory Council
- 2014 – 2018 NIH/NHGRI National Advisory Council for Human Genome Research
- 2015 Advisory Committee to NIH Director: Working Group on US Precision Medicine Initiative
- 2015 – present NIH/NHGRI Center of Excellence in Genomic Science (Stanford University; PI: Howard Chang) (Scientific Advisory Board)
- 2015 – 2018 NIH/OD 4D Nucleome Network (Steering Committee)
- 2014 – 2019 NIH/NIAID Center of Excellence in Translational Research (Harvard School of Public Health; PI: Megan Murray) (Scientific Advisory Board)
- 2012 – 2014 Joint Genome Institute, Department of Energy (Scientific Advisory Board)
- 2012 – 2015 NIH/NHGRI Centers for Mendelian Genomics (Steering Committee)
- 2009 – 2012 NIH/NHLBI Exome Sequencing Project (Steering Committee)

Journal Editorial Boards

- 2017 – present Science (Board of Reviewing Editors)
- 2015 – present Genome Medicine (Editorial Board)
- 2015 – present Molecular Case Studies (Editorial Board)
- 2014 – present Genetics (Associate Editor)
- 2014 – present Human Molecular Genetics (Editorial Board)

- 2011 – present Human Genetics (Editorial Board)
- 2010 – present Genome Biology (Editorial Advisory Board)
- 2009 – present Genome Research (Editorial Board)
- 2011 – 2018 Biotechniques (Editorial Board)
- 2009 – 2012 American Journal of Human Genetics (Associate Editor)

Meeting & Symposium Organization

- 2019 – present Co-organizer, Biology of Genomes, Cold Spring Harbor Labs (Cold Spring Harbor, NY)
- 2015 – 2019 Co-organizer, Genomics of Rare Diseases, Wellcome Genome Campus (Hinxton, UK)
- 2018 Co-organizer, Genome Sciences / Brotman Baty Institute Symposium – "*The Personal Genome: Sequencing, Understanding and Editing the Genome to Improve Human Health*"
- 2014 Organizer, Genome Sciences Symposium – "*Genetic Networks - From Model Organisms to Human Disease*"
- 2010 Co-organizer & Moderator, Symposium & Panel Discussion – "*New Discoveries in Medicine: Implications for the Cost and Quality of American Healthcare*"

Commercial Scientific Advisory Board and Consulting Roles

- 2016 – present Guardant Health (Scientific Consultant)
- 2018 – present Maze Therapeutics (Scientific Advisory Board)
- 2018 – present Camp4 Therapeutics (Scientific Advisory Board)
- 2016 – present Nanostring (Scientific Advisory Board)
- 2015 – present Phase Genomics (Founder; Scientific Advisory Board)
- 2010 – present Adaptive Biotechnologies (Scientific Advisory Board)
- 2009 – present Stratos Genomics (Scientific Advisory Board)
- 2016 – 2019 Bellwether Bio (Founder; Scientific Consultant)
- 2016 – 2019 Cambridge Epigenetix (Scientific Advisory Board)
- 2013 – 2018 GenePeeks (Scientific Advisory Board)
- 2009 – 2017 Good Start Genetics (Scientific Advisory Board)
- 2013 – 2017 Gen9 (Scientific Advisory Board)
- 2010 – 2015 Ariosa Diagnostics (Scientific Consultant)
- 2013 – 2015 Ingenuity Systems (Scientific Advisory Board)
- 2013 – 2015 Rubicon Genomics (Scientific Advisory Board)
- 2012 Merck Research Laboratories (Scientific Consultant)
- 2010 – 2011 Halo Genomics (Scientific Advisory Board)
- 2008 – 2009 Complete Genomics (Scientific Consultant)
- 2006 Highland Capital Partners (Scientific Consultant)
- 2004 – 2005 Agencourt Biosciences (Scientific Consultant)

Faculty Administrative Responsibilities (University of Washington)

- 2014 – 2015 Member, Seminar Series Committee (Genome Sciences)
- 2013 – 2014 Chair, Seminar Series Committee (Genome Sciences)
- 2008 – 2009 Member, Seminar Series Committee (Genome Sciences)

- 2017 – 2018 Member, Faculty Search Committee (Genome Sciences)
- 2016 – 2017 Member, Faculty Search Committee (Biology)
- 2013 – 2017 Member, Faculty Search Committee (Institute for Protein Design)
- 2008 – 2013 Member, Faculty Search Committee (Medical Genetics)
- 2010 – 2012 Member, Faculty Search Committee (Genome Sciences)
- 2008 – 2009 Member, Faculty Search Committee (Genome Sciences)

- 2009 Organizer, Departmental Retreat (Genome Sciences)

- 2012 – 2013 Co-chair, Scientific Discovery Subcommittee for Curriculum Renewal
- 2009 Member, U.W. "Two Years to Two Decades" (2y2d) initiative, Discovery focus group

Reviewer (ad hoc)

American Journal of Human Genetics
Analytical Chemistry
Bioinformatics
Biotechniques
BMC Genomics
Cell
Cell Stem Cell
Cellular & Molecular Biology Letters
Genetics in Medicine
Genome Biology
Genome Research
Genomics
Human Mutation
Mammalian Genome
Molecular Cell
Nature
Nature Biotechnology
Nature Genetics
Nature Medicine
Nature Methods
Nature Neuroscience
Nature Protocols
Nature Reviews Genetics
Neuron
New England Journal of Medicine
Nucleic Acids Research
PLoS Computational Biology
PLoS Genetics
Proceedings of the National Academy of Sciences
Science
Science Translational Medicine
Trends in Genetics

Grant Review & Related Service

- 2017 GGG-L(50) Advanced Genomic Technology Development Study Section, NIH
- 2014 Grant reviewer, Paul G. Allen Family Foundation ADI 2014 Life Science Focus
- 2014 Grant reviewer, TEDDY Whole Genome Sequencing Lab RFP
- 2014 Grant reviewer, National Institute of Diabetes and Digestive and Kidney Diseases Special Emphasis Panel
- 2013 Grant reviewer, National Institute of Child Health and Human Development Special Emphasis Panel
- 2013 Abstract reviewer, 63th Annual Meeting of American Society of Human Genetics
- 2013 Grant reviewer, The Wellcome Trust

- 2011 Grant reviewer, W. M. Keck Foundation
- 2011 Grant reviewer, Lasker Clinical Research Scholars Program
- 2010 Grant reviewer, UK Medical Research Council, Molecular and Cellular Medicine Board
- 2009 Grant reviewer, National Science Foundation
- 2009 Grant reviewer, NIH ARRA Challenge Grants (Genes, Genomes and Genetics IRG)
- 2009 Grant reviewer, Ontario Research Fund (GL2 Competition)
- 2008 Grant reviewer, Genome BritishColumbia

Postdoctoral Fellows Trained (University of Washington)

- 2019 – present Junhong Choi, Ph.D.
- 2019 – present Diego Calderon, Ph.D. (joint trainee with Cole Trapnell)
- 2018 – present Ronnie Blecher, Ph.D.
- 2018 – present Jacob Tome, Ph.D.
- 2018 – present Silvia Domcke Ph.D.
- 2016 – present Yi Yin, Ph.D.

- 2015 – 2019 Vikram Agarwal, Ph.D. (current: Computational Biologist, Calico Labs)
- 2014 – 2019 Bridget Kulasekara, Ph.D. (current: Research Scientist, Nanostring)
- 2014 – 2018 Jes Alexander, Ph.D.
- 2016 – 2018 Malte Spielmann, M.D. (current: Group Leader, Max Planck Institute for Molecular Genetics)
- 2014 – 2018 Darren Cusanovich, Ph.D. (current: Research Assistant Professor, Cellular and Molecular Medicine, University of Arizona)
- 2012 – 2017 Martin Kircher, Ph.D. (current: Junior Group Leader in Bioinformatics, Berlin Institute of Health)
- 2014 – 2016 Ron Hause, Ph.D. (current: Cell Therapy Predictive Sciences Group Lead, Celgene)
- 2011 – 2015 Stephen Salipante, M.D., Ph.D. (current: Assistant Professor, Department of Laboratory Medicine, University of Washington)
- 2009 – 2013 Jerrod Schwartz, Ph.D. (current: Head of Genomics, Verily Life Sciences)
- 2009 – 2013 Brian O'Roak, Ph.D. (joint trainee with Evan Eichler; current: Assistant Professor, Department of Molecular & Medical Genetics, Oregon Health & Science University)
- 2007 – 2009 Emily Turner, Ph.D. (current: Program Officer, Bill & Melinda Gates Foundation)

Graduate Students Trained (University of Washington)

- 2019 – Present Chengxiang (CX) Qiu (Genome Sciences)
- 2018 – Present Florence Chardon (Genome Sciences)
- 2018 – Present Sam Regalado (Genome Sciences)
- 2018 – Present Xingfang (Fanny) Huang (Computer Science & Engineering)
- 2017 – Present Anna Minkina (Genome Sciences)
- 2016 – Present Wei Chen (Molecular Engineering)

- 2015 – 2019 Molly Gasperini (Genome Sciences; dissertation entitled “Efficiently searching for enhancers and their target genes in the human genome”; current: Visiting Scientist, Octant Bio)
- 2015 – 2019 Andrew Hill (Genome Sciences; dissertation entitled “Expanding the scope and utility of single-cell genomic technologies”; current: Computational Biologist, 10X Genomics)
- 2014 – 2019 Seungsoo Kim (Genome Sciences; dissertation entitled “Maps and mechanisms of three-dimensional genome organization”; current: Postdoctoral Fellow, Wysocka Lab)
- 2016 – 2019 Junyue Cao (Molecular & Cellular Biology; dissertation entitled “Cell state and fate characterization by high-throughput single cell genomics”; current: Postdoctoral Fellow, Shendure Lab)
- 2015 – 2019 Hannah Pliner (Genome Sciences; joint trainee with Cole Trapnell; dissertation entitled “Algorithms for modeling gene regulation and determining cell type using single-cell molecular profiles”; current: Lead Data Scientist for Single Cell Genomics, Brotman Baty Institute for Precision Medicine)
- 2015 – 2018 Jason Klein (Medical Scientist Training Program, Genome Sciences; dissertation entitled “Massively Parallel Characterization of Enhancers in Evolution and Disease”; current: completing MSTP program)
- 2015 – 2018 Greg Findlay (Medical Scientist Training Program, Genome Sciences; dissertation entitled “High-throughput interrogation of genome function and cellular lineage"; current: completing MSTP program)
- 2014 – 2017 Vijay Ramani (Genome Sciences; dissertation entitled “Massively parallel analysis of nucleic acid structure”; current: Sandler Fellow, University of California, San Francisco)
- 2013 – 2017 Aaron McKenna (Genome Sciences; dissertation entitled “Whole-organism lineage tracing by combinatorial and cumulative genome editing”; current: Assistant Professor, Dartmouth University)
- 2012 – 2016 Matthew Snyder (Genome Sciences; dissertation entitled “Expanding the accuracy, resolution, and breadth of cell-free DNA investigation”; current: Research Scientist, Guardant Health)
- 2011 – 2014 Joshua Burton (Genome Sciences; dissertation entitled “New methods for de novo assembly of genomes and metagenomes”; current: Associate Director of Computational Biology, Resolution Bioscience)
- 2010 – 2014 Akash Kumar (Medical Scientist Training Program, Genome Sciences; dissertation entitled “Mutational Heterogeneity in Cancer: Lessons from the Brain and Prostate”; current: Pediatrics Resident at Stanford University)
- 2010 – 2014 Andrew Adey (Molecular & Cellular Biology; dissertation entitled “Comprehensive, precision genomics”; current: Assistant Professor, Department of Molecular & Medical Genetics, Oregon Health & Science University)
- 2009 – 2013 Jacob Kitzman (Genome Sciences; dissertation entitled “New technologies for sequencing and interpreting genomes”; current: Assistant Professor, Department of Genetics, University of Michigan)
- 2009 – 2012 Joseph Hiatt (Medical Scientist Training Program, Genome Sciences; dissertation entitled “Molecular tagging to overcome limitations of massively parallel sequencing”; current: Hematology-Oncology Fellow, University of Washington)
- 2007 – 2012 Sarah Ng (Genome Sciences; dissertation entitled “Next Generation Mendelian Genetics”; current: Research Fellow, Institute of Molecular and Cell Biology, Singapore)
- 2007 – 2012 Rupali Patwardhan (Genome Sciences; dissertation entitled “Massively parallel functional

dissection of regulatory elements"; current: Software Engineer, Facebook)

Rotation Students Supervised (University of Washington)

• Andrew Mullen	MSTP program	Summer 2019
• Chase Suiter	Molecular & Cellular Biology	Summer 2019
• Shawn Fayer	Genome Sciences	Spring 2019
• Chengxiang Qiu	Genome Sciences	Winter 2019
• James Anderson	Molecular & Cellular Biology	Winter 2019
• Eliza Barkan	Molecular & Cellular Biology	Fall 2018
• Michael Goldberg	Genome Sciences	Spring 2018
• Florence Chardon	Genome Sciences	Spring 2018
• Phillip Dishuck	Genome Sciences	Winter 2018
• William DeWitt	Genome Sciences	Fall 2017
• Xingfang Huang	Computer Science & Engineering	Fall 2017
• Sam Regalado	Genome Sciences	Summer 2017
• Ian Smith	Genome Sciences	Spring 2017
• April Lo	Genome Sciences	Spring 2017
• Anna Minkina	Genome Sciences	Fall 2016
• Wei Chen	Molecular Engineering	Spring 2016
• Eliah Overbey	Genome Sciences	Spring 2016
• Junyue Cao	Molecular & Cellular Biology	Summer 2015
• Molly Gasperini	Genome Sciences	Spring 2015
• Serena Liu	Genome Sciences	Spring 2015
• Hannah Pliner	Genome Sciences	Winter 2015
• Damon May	Genome Sciences	Winter 2015
• Andrew Hill	Genome Sciences	Fall 2014
• Vijay Ramani	Genome Sciences	Winter 2014
• Seungsoo Kim	Genome Sciences	Winter 2014
• Jason Klein	MSTP program	Summer 2013
• Hugh Haddox	Molecular & Cellular Biology	Spring 2013
• Aaron McKenna	Genome Sciences	Winter 2013
• Greg Findlay	MSTP program	Summer 2012
• Matthew Snyder	Genome Sciences	Spring 2012
• Jorgen Nelson	Genome Sciences	Winter 2012
• Elyse Hope	Genome Sciences	Winter 2012
• Meara Davies	Molecular & Cellular Biology	Fall 2011
• Josh Burton	Genome Sciences	Winter 2011
• Jenny Wagner	Genome Sciences	Winter 2011
• Andrew Adey	Molecular & Cellular Biology	Fall 2009
• David Young	MSTP program	Summer 2009
• Akash Kumar	MSTP program	Summer 2009
• Jacob Kitzman	Genome Sciences	Spring 2009
• Keisha Carlson	Genome Sciences	Winter 2009
• Jarrett Egerston	Genome Sciences	Winter 2009
• Matthew Maurano	Genome Sciences	Fall 2008
• Joseph Hiatt	MSTP program	Summer 2008
• Sayer Herrin	Genome Sciences	Winter 2008

• Rupali Patwardhan	Genome Sciences	Winter 2008
• Sarah Ng	Genome Sciences	Fall 2007

Graduate Student Committees (in addition to own trainees)

• 2019 – Present	Gesine Cauer	U.W. Genome Sciences	Advisor: Bill Noble
• 2018 – Present	Sanjay Srivatsan	U.W. Genome Sciences	Advisor: Cole Trapnell
• 2018 – Present	Andria Ellis	U.W. Genome Sciences	Advisor: Cole Trapnell
• 2018 – Present	Ian Smith	U.W. Genome Sciences	Advisor: Judit Villen
• 2017 – Present	Robin Kirkpatrick	U.W. Genome Sciences	Advisor: Jesse Zalatan
• 2016 – Present	Wei Zhou	U.W. Molecular & Cellular Biology	Advisor: Stan Fields
• 2016 – Present	Clara Amorosi	U.W. Genome Sciences	Advisor: Maitreya Dunham
• 2016 – Present	Nuttada Panpradist	U.W. Bioengineering	Advisor: Barry Lutz
• 2015 – Present	Ian Nova	U.W. Molecular Engineering	Advisor: Jens Gundlach
• 2015 – Present	Melissa Chaisson	U.W. Genome Sciences	Advisor: Doug Fowler
• 2016 – 2019	Aaron Wolf	U.W. Genome Sciences	Advisor: Josh Akey
• 2014 – 2019	Piero Lamelza	U.W. Molecular & Cellular Biology	Advisor: Michael Ailion
• 2018 – 2019	Peter Ney	U.W. Comp. Science & Engineering	Advisor: Tadayoshi Kohno
• 2016 – 2019	Rebecca Zaunbrecher	U.W. Bioengineering	Advisor: Mike Regnier
• 2013 – 2018	Jorgen Nelson	U.W. Genome Sciences	Advisor: David Baker
• 2015 – 2018	John Crowl	U.W. Immunology	Advisor: Dan Stetson
• 2015 – 2017	Jocelynn Pearl	U.W. Molecular & Cellular Biology	Advisor: Lee Hood
• 2014 – 2017	Hugh Haddox	U.W. Molecular & Cellular Biology	Advisor: Jesse Bloom
• 2011 – 2017	Jennifer Andrie	U.W. Genome Sciences	Advisor: Josh Akey
• 2015 – 2016	Alexander Rosenberg	U.W. Electrical Engineering	Advisor: Georg Seelig
• 2013 – 2016	David Young	U.W. Genome Sciences	Advisor: Stan Fields
• 2011 – 2015	Vaughn Iverson	U.W. Oceanography	Advisor: Virginia Armbrust
• 2012 – 2015	Benjamin Vernot	U.W. Genome Sciences	Advisor: Josh Akey
• 2012 – 2014	Andrew Laszlo	U.W. Physics	Advisor: Jens Gundlach
• 2012 – 2014	Niklas Krumm	U.W. Genome Sciences	Advisor: Evan Eichler
• 2010 – 2014	Russell Berg	U.W. Molecular & Cellular Biology	Advisor: Lalita Ramakrishnan
• 2010 – 2014	Keisha Carlson	U.W. Genome Sciences	Advisor: Christine Queitsch
• 2010 – 2014	Leslie Emery	U.W. Genome Sciences	Advisor: Josh Akey
• 2010 – 2013	Peter Sudmant	U.W. Genome Sciences	Advisor: Evan Eichler
• 2010 – 2013	Thomas White	U.W. Molecular & Cellular Biology	Advisor: Peter Nelson
• 2010 – 2013	Benjamin Whiddon	U.W. Genome Sciences	Advisor: Richard Palmiter
• 2009 – 2013	Cailyn Spurrell	U.W. Genome Sciences	Advisor: Mary-Claire King
• 2008 – 2013	Alan Rubin	U.W. Genome Sciences	Advisor: Phil Green
• 2011 – 2012	Lucas Gray	U.W. Biochemistry	Advisor: Alan Weiner
• 2009 – 2012	Joshua Bishop	U.W. Electrical Engineering	Advisor: Eric Klavins
• 2009 – 2012	Kyle Minch	U.W. Molecular & Cellular Biology	Advisor: David Sherman
• 2011	Sung Hang	U.W. Neurobiology and Behavior	Advisor: William Catterall
• 2010	Carlos Araya	U.W. Genome Sciences	Advisor: Stanley Fields
• 2008 – 2010	Steven Josefowicz	U.W. Immunology	Advisor: Sasha Rudensky
• 2008 – 2010	Kevin Schutz	U.W. Genome Sciences	Advisor: Stan Fields
• 2008 – 2010	Marcia Paddock	U.W. Immunology	Advisor: Andy Scharenberg

Courses Taught

- 2017, 2019 GENOME 373 – "Genomics & Proteomics" (University of Washington)
  Undergraduate lecture course; co-taught with Jim Bruce
- 2008 – present GENOME 550 – "Methods and Logic in Genetics" (University of Washington)
  Graduate seminar course; co-taught with Bob Waterston or Alejandro Wolf-Yadlin
- 2012 – 2016 HUBIO 554 – "Genetics" (University of Washington)
  Medical school 2nd year pre-clinical curriculum; co-chaired with Heather Mefford
- 2012 – 2015 CONJOINT 511 – "Genetic Anatomy" (University of Washington)
  Medical school 1st year elective; co-taught w/ Marshall Horwitz and John Clark
- 2010 – 2012 GENOME 373 – "Genome Informatics" (University of Washington)
  Undergraduate lecture course; co-taught with Jim Thomas or Elhanan Borenstein
- 2001 – 2003 "Principles of Pharmacology" (Harvard Medical School)
  Teaching assistant, 1st year medical school course

Other Teaching or Outreach Activities

- Apr 2019 Guest speaker, UW MSTP Dinner/Recruitment meeting
- Oct 2018 Panelist, "The Power of Personalized Medicine", Geekwire 2018 Summit (Seattle, WA)
- Dec 2016 Panelist, National Academy of Medicine (NAM) Rosenthal Symposium: "Precision Population Health" (UW)
- Nov 2016 Lecturer, Medical Genetics, "Introduction to Human & Medical Genetics" course (UW)
- Jul 2016 Speaker, "Wednesdays at the Genome", UW Genome Sciences Public Lecture Series
- May 2016 Guest session leader, BIOEN 498/599, "Genomics Era Sequencing Technologies and Analysis" (UW)
- Oct 2015 Guest session leader, MCB 517, "The Developmental Basis of Human Disease" (UW)
- Apr 2015 Guest session leader, CSE 590C, "Readings and Research in Computational Biology" (UW)
- Nov 2014 Guest session leader, EE 423, "Introduction to Synthetic Biology" (UW)
- Sep 2014 Lecturer, Medical Genetics, "Introduction to Human & Medical Genetics" (UW)
- Aug 2014 Co-organizer, UW Center for Mendelian Genomics (CMG) Data Analysis Workshop
- Jan 2014 Panelist, Edmonds Community College Brown Bag Lecture Series, "The Life and Cells of Henrietta Lacks: Science, Society, and Individual Perspectives"
- Dec 2013 Guest session leader, BIOL 485, "Senior Seminar in Cellular, Molecular and Developmental Biology" (UW)
- Nov 2013 Keynote speaker, UW Postdoc Association Symposium
- Nov 2013 Speaker, Pacific Science Center "Science Café" series
- Oct 2013 Guest session leader, MCB 517, "The Developmental Basis of Human Disease" (UW)
- Aug 2013 Co-organizer, UW Center for Mendelian Genomics (CMG) Data Analysis Workshop
- Jul 2013 Speaker, UW Genome Sciences summer research internship program
- Jun 2013 Guest session leader, MEBI 590, "Biomedical & Health Informatics Lecture Series" (UW)
- Apr 2013 Guest speaker, UW MSTP Dinner/Recruitment meeting
- Apr 2013 Guest session leader, EPI 590, "Introduction to Laboratory Methods in Population

Research" (UW)

- Oct 2012 Speaker, Seattle Sequencing Interest Group
- Oct 2012 Lecturer, Medical Genetics, “Introduction to Human & Medical Genetics” (UW)
- Jul 2012 Speaker, “Science on Tap” series
- Jul 2012 Speaker, UW Genome Sciences summer research internship program
- Apr 2012 Guest session leader, GENOME 580, “Ethics in Biomedical Research and Teaching” (UW)
- Apr 2011 Guest session leader, GENOME 580, “Ethics in Biomedical Research and Teaching” (UW)
- Apr 2011 Guest session leader, EPI 590 "Introduction to Laboratory Methods in Population Research" (UW)
- Oct 2010 Lecturer, Medical Genetics, “Introduction to Human & Medical Genetics” (UW)
- Nov 2009 Panelist, Lasker Foundation / UW Dept. of Genome Sciences Round Table: “Personal Genomes: Promise or Hype?”
- Sep 2009 Panelist, "The Two Body Question and Faculty with children" at HHMI Future Faculty Workshop
- Apr 2009 Guest session leader, GENOME 580, “Ethics in Biomedical Research and Teaching” (UW)
- Apr 2009 Guest speaker, UW MSTP Dinner/Recruitment meeting
- Apr 2009 Guest session leader, EPI 590, "Introduction to Laboratory Methods in Population Research" (UW)
- Feb 2009 Guest speaker, Rainier Scholars program (UW)
- Jul 2008 Speaker, StarNet 2008 Summer Workshop, UW Genome Sciences Education Outreach
- Jul 2008 Speaker, “Wednesdays at the Genome”, UW Genome Sciences Public Lecture Series
- Oct 2008 Speaker, Chalk Talk Workshop, UW Women in Genome Sciences (WiGS)
- May 2008 Guest session leader, GENOME 580, “Ethics in Biomedical Research and Teaching” (UW)

Active Patents & Published Patent Applications

- Polony fluorescent in situ sequencing beads (issued; 7,425,431)
- Error detection in sequence tag directed subassemblies of short sequencing reads (issued; 8,865,410)
- Sequence tag directed subassembly of short sequencing reads into long sequencing reads (issued; 8,846,347; 8,383,345; 10,227,585)
- Nanogrid rolling circle DNA sequencing (issued: 9,624,538)
- Methods for retrieval of sequence-verified DNA constructs (issued: 9,809,904)
- Massively parallel contiguity mapping (issued: 10,457,936)

- Multiplex pairwise assembly of DNA oligonucleotides (application; 20180320166)
- Methods of determining tissues and/or cell types giving rise to cell-free dna, and methods of identifying a

disease or disorder using same (application; PCT/US2015/042310)
- Methods and systems for large scale scaffolding of genome assemblies (application; PCT/US2014/057930)
- A framework for determining the relative effects of genetic mutations (application; 20160357903)
- Methods and systems for large scale scaffolding of genome assemblies (application; 20160239602)
- Multiplex homology-directed repair (application; 20160076093)
- Systems, Algorithms, and Software for Molecular Inversion Probe (MIP) Design (application; 20160055293)
- Highly multiplex single amino acid mutagenesis for massively parallel functional analysis (application; 20160017410)
- Whole genome sequencing of a human fetus (application; 20150105267)
- Multiplex decoding of sequence tags in barcodes (application; 20080269068)
- Wobble sequencing (application; 20070207482)
- Nucleic acid memory device (application; 20100099080)

Publications (* denotes equal contributors; # denotes corresponding/senior author(s); grey numbers denote primary publications, defined as those on which I and/or a member of my lab are a corresponding and/or a first author)

297. Srivatsan SR*, McFaline-Figueroa JL*, Ramani V*, Saunders L, Cao J, Packer J, Pliner HA, Jackson DL, Daza RM, Christiansen L, Zhang F, Steemers F, **Shendure J**#, Trapnell C#. Massively multiplex chemical transcriptomics at single cell resolution. *Science* 2019 Dec 5. pii: eaax6234. [Epub ahead of print]

296. Alexander J, Findlay GM, Kircher M, **Shendure J**#. Concurrent genome and epigenome editing by CRISPR-mediated sequence replacement. *BMC Biology* 2019 Nov 18;17(1):90.

295. Soza VL#, Lindsley D, Waalkes A, Ramage E, Patwardhan RP, Burton JN, Adey A, Kumar A, Qiu R, **Shendure J**, Hall B. The Rhododendron Genome and Chromosomal Organization Provide Insight into Shared Whole-Genome Duplications across the Heath Family (Ericaceae). *Genome Biology & Evolution* 2019 Dec 1;11(12):3353-3371.

294. Esposito D, Weile J, **Shendure J**, Starita LM, Papenfuss AT, Roth FP#, Fowler DM#, Rubin AF#. MaveDB: an open-source platform to distribute and interpret data from multiplexed assays of variant effect. *Genome Biology* 2019 Nov 4;20(1):223.

293. Ramani V#, Deng X, Qiu R, Lee C, Disteche CM, Noble WS, **Shendure J**#, Duan Z#. Sci-Hi-C: A single-cell Hi-C method for mapping 3D genome organization in large number of single cells. *Methods* 2019 Sep 16. pii: S1046-2023(18)30475-4. [Epub ahead of print]

292. Kim S, **Shendure J**#. Mechanisms of Interplay between Transcription Factors and the 3D Genome. *Molecular Cell* 2019 Oct 17;76(2):306-319.

291. Pliner HA, **Shendure J**#, Trapnell C#. Supervised classification enables rapid annotation of cell atlases. *Nature Methods* 2019 Oct;16(10):983-986.

290. Yin Y#, Jiang Y, Lam KG, Berletch JB, Disteche CM, Noble WS, Steemers FJ, Camerini-Otero RD, Adey AC, **Shendure J**#. High-Throughput Single-Cell Sequencing with Linear Amplification. *Molecular Cell* 2019 Nov 21;76(4):676-690.e10.

289. McFaline-Figueroa JL, Hill AJ, Qiu X, Jackson D, **Shendure J**, Trapnell C#. A pooled single-cell genetic screen identifies regulatory checkpoints in the continuum of the epithelial-to-mesenchymal transition. *Nature Genetics* 2019 Sep;51(9):1389-1398.

288. Kircher M*#, Xiong C*, Martin B*, Schubach M*, Inoue F, Bell RJA, Costello JF, **Shendure J**#, Ahituv N#. Saturation mutagenesis of twenty disease-associated regulatory elements at single base-pair resolution. *Nat Communications* 2019 Aug 8;10(1):3583.

287. Bertero A, Fields PA, Smith AST, Leonard A, Beussman K, Sniadecki NJ, Kim DH, Tse HF, Pabon L, **Shendure J**, Noble WS, Murry CE. Chromatin compartment dynamics in a haploinsufficient model of cardiac laminopathy. *Journal of Cell Biology* 2019 Sep 2;218(9):2919-2944.
286. Chen W, McKenna A*, Schreiber J*, Haeussler M, Yin Y, Agarwal V, Noble WS, **Shendure J**#. Massively parallel profiling and predictive modeling of the outcomes of CRISPR/Cas9-mediated double-strand break repair. *Nucleic Acids Research* 2019 Sep 5;47(15):7989-8003.
285. Klein JC, Keith A, Rice SJ, Shepherd C, Agarwal V, Loughlin J, **Shendure J**#. Functional testing of thousands of osteoarthritis-associated variants for regulatory activity. *Nature Communications* 2019 Jun 4;10(1):2434.
284. Minkina A, **Shendure J**#. Expanding the single-cell genomics toolkit. *Nature Genetics* 2019 Jun;51(6):931-932.
283. Shigaki D, Adato O, Adhikar AN, Dong S, Hawkins-Hooker A, Inoue F, Juven-Gershon T, Kenlay H, Martin B, Patra A, Penzar DP, Schubach M, Xiong C, Yan Z, Boyle AP, Kreimer A, Kulakovskiy IV, Reid J, Unger R, Yosef N, **Shendure J**, Ahituv N, Kircher M, Beer MA#. Integration of Multiple Epigenomic Marks Improves Prediction of Variant Impact in Saturation Mutagenesis Reporter Assay. *Human Mutation* 2019 Sep;40(9):1280-1291.
282. Farhat MR#, Freschi L, Calderon R, Ioerger T, Snyder M, Meehan CJ, de Jong B, Rigouts L, Sloutsky A, Kaur D, Sunyaev S, van Soolingen D, **Shendure J**, Sacchettini J, Murray M. GWAS for quantitative resistance phenotypes in Mycobacterium tuberculosis reveals resistance genes and regulatory regions. *Nature Communications* 2019 May 13;10(1):2128.
281. Kim S, Dunham MJ, **Shendure J**#. A combination of transcription factors mediates inducible interchromosomal contacts. *Elife* 2019 May 13;8.
280. Bertero A*, Fields PA*, Ramani V, Bonora G, Yardimci GG, Reinecke H, Pabon L, Noble WS, **Shendure J**, Murry CE#. Dynamics of genome reorganization during human cardiogenesis reveal an RBM20-dependent splicing factory. *Nature Communications* 2019 Apr 4;10(1):1538.
279. Ng BG, Lourenço CM, Losfeld ME, Buckingham KJ, Kircher M, Nickerson DA, **Shendure J**, Bamshad MJ; University of Washington Center for Mendelian Genomics, Freeze HH#. Mutations in the translocon-associated protein complex subunit SSR3 cause a novel congenital disorder of glycosylation. *Journal of Inherited Metabolic Disease* 2019 Sep;42(5):993-997.
278. **Shendure J**#, Findlay GM, Snyder MW. Genomic Medicine: Progress, Pitfalls, and Promise. *Cell* 2019 Mar 21;177(1):45-57.
277. Ramani V#, Qiu R, **Shendure J**#. High Sensitivity Profiling of Chromatin Structure by MNase-SSP. *Cell Reports* 2019 Feb 26;26(9):2465-2476.e4.
276. Swygert SG, Kim S, Wu X, Fu T, Hsieh TH, Rando OJ, Eisenman RN, **Shendure J**, McKnight JN, Tsukiyama T#. Condensin-Dependent Chromatin Compaction Represses Transcription Globally during Quiescence. *Molecular Cell* 2019 Feb 7;73(3):533-546.e4.
275. Cao J*, Spielmann M*, Qiu X, Huang X, Ibrahim DM, Hill AJ, Zhang F, Mundlos S, Christiansen L, Steemers FJ, Trapnell C#, **Shendure J**#. The single-cell transcriptional landscape of mammalian organogenesis. *Nature* 2019 Feb;566(7745):496-502.
274. Gasperini M#, Hill AJ, McFaline-Figueroa JL, Martin B, Kim S, Zhang MD, Jackson D, Leith A, Schreiber J, Noble WS, Trapnell C, Ahituv N, **Shendure J**#. A Genome-wide Framework for Mapping Gene Regulation via Cellular Genetic Screens. *Cell* 2019 Jan 10;176(1-2):377-390.e19.
273. Rentzsch P, Witten D, Cooper GM, **Shendure J**#, Kircher M#. CADD: predicting the deleteriousness of variants throughout the human genome. *Nucleic Acids Research* 2019 Jan 8;47(D1):D886-D894.
272. Ng BG, Underhill HR, Palm L, Bengtson P, Rozet JM, Gerber S, Munnich A, Zanlonghi X, Stevens CA, Kircher M, Nickerson DA, Buckingham KJ, Josephson KD, **Shendure J**, Bamshad MJ; University of

Washington Center for Mendelian Genomics, Freeze HH, Eklund EA[#]. DPAGT1 Deficiency with Encephalopathy (DPAGT1-CDG): Clinical and Genetic Description of 11 New Patients. *JIMD Reports* 2019;44:85-92.

271. Sanchez C, Snyder MW, Tanos R, **Shendure J**, Thierry AR[#]. New insights into structural features and optimal detection of circulating tumor DNA determined by single-strand DNA analysis. *NPJ Genomic Medicine* 2018 Nov 23;3:31.

270. CRyPTIC Consortium and the 100,000 Genomes Project, Allix-Béguec C, Arandjelovic I, Bi L, Beckert P, Bonnet M, Bradley P, Cabibbe AM, Cancino-Muñoz I, Caulfield MJ, Chaiprasert A, Cirillo DM, Clifton DA, Comas I, Crook DW, De Filippo MR, de Neeling H, Diel R, Drobniewski FA, Faksri K, Farhat MR, Fleming J, Fowler P, Fowler TA, Gao Q, Gardy J, Gascoyne-Binzi D, Gibertoni-Cruz AL, Gil-Brusola A, Golubchik T, Gonzalo X, Grandjean L, He G, Guthrie JL, Hoosdally S, Hunt M, Iqbal Z, Ismail N, Johnston J, Khanzada FM, Khor CC, Kohl TA, Kong C, Lipworth S, Liu Q, Maphalala G, Martinez E, Mathys V, Merker M, Miotto P, Mistry N, Moore DAJ, Murray M, Niemann S, Omar SV, Ong RT, Peto TEA, Posey JE, Prammananan T, Pym A, Rodrigues C, Rodrigues M, Rodwell T, Rossolini GM, Sánchez Padilla E, Schito M, Shen X, **Shendure J**, Sintchenko V, Sloutsky A, Smith EG, Snyder M, Soetaert K, Starks AM, Supply P, Suriyapol P, Tahseen S, Tang P, Teo YY, Thuong TNT, Thwaites G, Tortoli E, van Soolingen D, Walker AS, Walker TM, Wilcox M, Wilson DJ, Wyllie D, Yang Y, Zhang H, Zhao Y, Zhu B. Prediction of Susceptibility to First-Line Tuberculosis Drugs by DNA Sequencing. *New England Journal of Medicine* 2018 Oct 11;379(15):1403-1415.

269. Starita LM, Islam MM, Banerjee T, Adamovich AI, Gullingsrud J, Fields S, **Shendure J**[#], Parvin JD[#]. A Multiplex Homology-Directed DNA Repair Assay Reveals the Impact of More Than 1,000 BRCA1 Missense Substitution Variants on Protein Function. *American Journal of Human Genetics* 2018 Oct 4;103(4):498-508.

268. Liu S*, Huang S*, Chen F*, Zhao L*, Yuan Y*, Francis SS, Fang L, Li Z, Lin L, Liu R, Zhang Y, Xu H, Li S, Zhou Y, Davies RW, Liu Q, Walters RG, Lin K, Ju J, Korneliussen T, Yang MA, Fu Q, Wang J, Zhou L, Krogh A, Zhang H, Wang W, Chen Z, Cai Z, Yin Y, Yang H, Mao M, Shendure J, Wang J[#], Albrechtsen A[#], Jin X[#], Nielsen R[#], Xu X[#]. Genomic Analyses from Non-invasive Prenatal Testing Reveal Genetic Associations, Patterns of Viral Infections, and Chinese Population History. *Cell* 2018 Oct 4;175(2):347 359.e14.

267. Findlay GM, Daza RM, Martin B, Zhang MD, Leith AP, Gasperini M, Janizek JD, Huang X, Starita LM[#], **Shendure J**[#]. Accurate classification of BRCA1 variants with saturation genome editing. *Nature* 2018 Sep 12.

266. Cao J, Cusanovich DA*, Ramani V*, Aghamirzaie D, Pliner HA, Hill AJ, Daza RM, McFaline-Figueroa JL, Packer JS, Christiansen L, Steemers FJ, Adey AC, Trapnell C[#], **Shendure J**[#]. Joint profiling of chromatin accessibility and gene expression in thousands of single cells. *Science* 2018 Aug 30. pii: eaau0730.

265. Pliner HA, Packer JS, McFaline-Figueroa JL, Cusanovich DA, Daza RM, Aghamirzaie D, Srivatsan S, Qiu X, Jackson D, Minkina A, Adey AC, Steemers FJ, **Shendure J**[#], Trapnell C[#]. Cicero Predicts cis-Regulatory DNA Interactions from Single-Cell Chromatin Accessibility Data. *Molecular Cell* 2018 Sep 6;71(5):858-871.e8.

264. Cusanovich DA*, Hill AJ*, Aghamirzaie D, Daza RM, Pliner HA, Berletch JB, Filippova GN, Huang X, Christiansen L, DeWitt WS, Lee C, Regalado SG, Read DF, Steemers FJ, Disteche CM, Trapnell C[#], **Shendure J**[#]. A Single-Cell Atlas of In Vivo Mammalian Chromatin Accessibility. *Cell* 2018 Aug 23;174(5):1309-1324.e18.

263. Ng BG, Underhill HR, Palm L, Bengtson P, Rozet JM, Gerber S, Munnich A, Zanlonghi X, Stevens CA, Kircher M, Nickerson DA, Buckingham KJ, Josephson KD, **Shendure J**, Bamshad MJ; University of Washington Center for Mendelian Genomics, Freeze HH, Eklund EA[#]. DPAGT1 Deficiency with Encephalopathy (DPAGT1-CDG): Clinical and Genetic Description of 11 New Patients. *JIMD Reports* 2018 Aug 17.

262. Klein JC[#], Keith A, Agarwal V, Durham T, **Shendure J[#]**. Functional characterization of enhancer evolution in the primate lineage. *Genome Biology* 2018 Jul 25;19(1):99.

261. McKenna A[#], **Shendure J**. FlashFry: a fast and flexible tool for large-scale CRISPR target design. *BMC Biology* 2018 Jul 5;16(1):74.

260. Kronenberg ZN, Fiddes IT*, Gordon D, Murali S*, Cantsilieris S*, Meyerson OS*, Underwood JG*, Nelson BJ*, Chaisson MJP, Dougherty ML, Munson KM, Hastie AR, Diekhans M, Hormozdiari F, Lorusso N, Hoekzema K, Qiu R, Clark K, Raja A, Welch AE, Sorensen M, Baker C, Fulton RS, Armstrong J, Graves-Lindsay TA, Denli AM, Hoppe ER, Hsieh P, Hill CM, Pang AWC, Lee J, Lam ET, Dutcher SK, Gage FH, Warre WC, **Shendure J**, Haussler D, Schneider VA, Cao H, Ventura M, Wilson RK, Paten B, Pollen A, Eichler EE[#]. High-resolution comparative analysis of great ape genomes. *Science* 2018 Jun 8;360(6393).

259. Matreyek KA*, Starita LM*, Stephany JJ, Martin B, Chiasson MA, Gray VE, Kircher M, Khechaduri A, Dines JN, Hause RJ, Bhatia S, Evans WE, Relling MV, Yang W, **Shendure J[#]**, Fowler DM[#]. Multiplex assessment of protein variant abundance by massively parallel sequencing. *Nature Genetics* 2018 Jun;50(6):874-882.

258. Bonora G*, Deng X*, Fang H, Ramani V, Qiu R, Berletch JB, Filippova GN, Duan Z, **Shendure J**, Noble WS[#], Disteche CM[#]. Orientation-dependent Dxz4 contacts shape the 3D structure of the inactive X chromosome. *Nature Communications* 2018 Apr 13;9(1):1445.

257. Mulqueen RM, Pokholok D, Norberg SJ, Torkenczy KA, Fields AJ, Sun D, Sinnamon JR, **Shendure J**, Trapnell C, O'Roak BJ, Xia Z, Steemers FJ, Adey AC[#]. Highly scalable generation of DNA methylation profiles in single cells. *Nature Biotechnology* 2018 Jun;36(5):428-431.

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

255. Cusanovich DA*, Reddington JP*, Garfield DA*, Daza RM, Aghamirzaie D, Marco-Ferreres R, Pliner HA, Christiansen L, Qiu X, Steemers FJ, Trapnell C, **Shendure J[#]**, Furlong EEM[#]. The cis-regulatory dynamics of embryonic development at single-cell resolution. *Nature* 2018 Mar 22;555(7697):538-542.

254. Hill AJ*, McFaline-Figueroa JL*, Starita LM, Gasperini MJ, Matreyek KA, Packer J, Jackson D, **Shendure J[#]**, Trapnell C[#]. On the design of CRISPR-based single-cell molecular screens. *Nature Methods* 2018 Apr;15(4):271-274.

253. Ma W, Ay F, Lee C, Gulsoy G, Deng X, Cook S, Hesson J, Cavanaugh C, Ware CB, Krumm A, **Shendure J**, Blau CA, Disteche CM, Noble WS[#], Duan Z[#]. Using DNase Hi-C techniques to map global and local three-dimensional genome architecture at high resolution. *Methods* 2018 Jun 1;142:59-73.

252. Klein JC[#], Chen W, Gasperini M, **Shendure J[#]**. Identifying Novel Enhancer Elements with CRISPR-Based Screens. *ACS Chemical Biology* 2018 Feb 16;13(2):326-332.

251. Johnsen JM, Fletcher SN, Huston H, Roberge S, Martin BK, Kircher M, Josephson NC, **Shendure J**, Ruuska S, Koerper MA, Morales J, Pierce GF, Aschman DJ, Konkle BA[#]. Novel approach to genetic analysis and results in 3000 hemophilia patients enrolled in the My Life, Our Future initiative. *Blood Advances* 2017 May 18;1(13):824-834.

250. Gray VE, Hause RJ, Luebeck J, **Shendure J**, Fowler DM[#]. Quantitative Missense Variant Effect Prediction Using Large-Scale Mutagenesis Data. *Cell Systems* 2018 Jan 24;6(1):116-124.e3.

249. Weichenhan D[#], Wang Q, Adey A, Wolf S, **Shendure J**, Eils R, Plass C. Tagmentation-Based Library Preparation for Low DNA Input Whole Genome Bisulfite Sequencing. *Methods Molecular Biology* 2018;1708:105122.

248. Liu J, Halloran JT, Bilmes JA, Daza RM, Lee C, Mahen EM, Prunkard D, Song C, Blau S, Dorschner MO, Gadi VK, **Shendure J**, Blau CA[#], Noble WS[#]. Comprehensive statistical inference of the clonal structure of cancer from multiple biopsies. *Science Reports* 2017 Dec 5;7(1):16943.

247. Küry S*[#], van Woerden GM*, Besnard T*, Proietti Onori M, Latypova X, Towne MC, Cho MT, Prescott TE, Ploeg MA, Sanders S, Stessman HAF, Pujol A, Distel B, Robak LA, Bernstein JA, Denommé-Pichon AS, Lesca G, Sellars EA, Berg J, Carré W, Busk ØL, van Bon BWM, Waugh JL, Deardorff M, Hoganson GE, Bosanko KB, Johnson DS, Dabir T, Holla ØL, Sarkar A, Tveten K, de Bellescize J, Braathen GJ, Terhal PA, Grange DK, van Haeringen A, Lam C, Mirzaa G, Burton J, Bhoj EJ, Douglas J, Santani AB, Nesbitt AI, Helbig KL, Andrews MV, Begtrup A, Tang S, van Gassen KLI, Juusola J, Foss K, Enns GM, Moog U, Hinderhofer K, Paramasivam N, Lincoln S, Kusako BH, Lindenbaum P, Charpentier E, Nowak CB, Cherot E, Simonet T, Ruivenkamp CAL, Hahn S, Brownstein CA, Xia F, Schmitt S, Deb W, Bonneau D, Nizon M, Quinquis D, Chelly J, Rudolf G, Sanlaville D, Parent P, Gilbert-Dussardier B, Toutain A, Sutton VR, Thies J, Peart-Vissers LELM, Boisseau P, Vincent M, Grabrucker AM, Dubourg C; Undiagnosed Diseases Network, Tan WH, Verbeek NE, Granzow M, Santen GWE, **Shendure J**, Isidor B, Pasquier L, Redon R, Yang Y, State MW, Kleefstra T, Cogné B; GEM HUGO; Deciphering Developmental Disorders Study, Petrovski S, Retterer K, Eichler EE, Rosenfeld JA, Agrawal PB, Bézieau S*, Odent S*, Elgersma Y*[#], Mercier S*. De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. *American Journal of Human Genetics* 2017 Nov 2;101(5):768-788.

246. **Shendure J[#]**, Balasubramanian S, Church GM, Gilbert W, Rogers J, Schloss JA, Waterston RH. DNA sequencing at 40: past, present and future. *Nature* 2017 Oct 19;550(7676):345-353.

245. Dekker J[#], Belmont AS, Guttman M, Leshyk VO, Lis JT, Lomvardas S, Mirny LA, O'Shea CC, Park PJ, Ren B, Politz JCR, **Shendure J**, Zhong S; 4D Nucleome Network. The 4D nucleome project. *Nature* 2017 Sep 13;549(7671):219-226.

244. Smukowski Heil C, Burton JN, Liachko I, Friedrich A, Hanson NA, Morris CL, Schacherer J, **Shendure J**, Thomas JH, Dunham MJ[#]. Identification of a novel interspecific hybrid yeast from a metagenomic spontaneously inoculated beer sample using Hi-C. *Yeast* 2018 Jan;35(1):71-84.

243. Starita LM[#], Ahituv N, Dunham MJ, Kitzman JO, Roth FP, Seelig G, **Shendure J**, Fowler DM[#]. Variant Interpretation: Functional Assays to the Rescue. *American Journal of Human Genetics* 2017 Sep 7;101(3):315-325.

242. Ng BG, Asteggiano CG, Kircher M, Buckingham KJ, Raymond K, Nickerson DA, **Shendure J**, Bamshad MJ, University of Washington Center for Mendelian Genomics, Ensslen M[#], Freeze HH[#]. Encephalopathy caused by novel mutations in the CMP-sialic acid transporter, SLC35A1. *American Journal of Medical Genetics* 2017 Nov;173(11):2906-2911.

241. Cao J*, Packer JS*, Ramani V, Cusanovich DA, Huynh C, Daza R, Qiu X, Lee C, Furlan SN, Steemers FJ, Adey A, Waterston RH[#], Trapnell C[#], **Shendure J[#]**. Comprehensive single-cell transcriptional profiling of a multicellular organism. *Science* 2017 Aug 18;357(6352):661-667.

240. Gasperini M*[#], Findlay GM*, McKenna A, Milbank JH, Lee C, Zhang MD, Cusanovich DA, **Shendure J[#]**. CRISPR/Cas9-Mediated Scanning for Regulatory Elements Required for HPRT1 Expression via Thousands of Large, Programmed Genomic Deletions. *American Journal of Human Genetics* 2017 Aug 3;101(2):192-205.

239. Zhang F*, Christiansen L*, Thomas J, Pokholok D, Jackson R, Morrell N, Zhao Y, Wiley M, Welch E, Jaeger E, Granat A, Norberg SJ, Halpern A, C Rogert M, Ronaghi M, **Shendure** J, Gormley N, Gunderson KL, Steemers FJ[#]. Haplotype phasing of whole human genomes using bead-based barcode partitioning in a single tube. *Nature Biotechnology* 2017 Sep;35(9):852-857.

238. Kim S, Liachko I, Brickner DG, Cook K, Noble WS, Brickner JH, **Shendure J[#], Dunham MJ[#]**. The dynamic three-dimensional organization of the diploid yeast genome. *Elife* 2017 May 24;6.

237. Arts P*, van der Raadt J*, van Gestel SHC*, Steehouwer M, **Shendure J**, Hoischen A, Albers CA[#]. Quantification of differential gene expression by multiplexed targeted resequencing of cDNA. *Nature Communications* 2017 May 5;8:15190.

236. Bickhart DM, Rosen BD, Koren S, Sayre BL, Hastie AR, Chan S, Lee J, Lam ET, Liachko I, Sullivan ST, Burton JN, Huson HJ, Nystrom JC, Kelley CM, Hutchison JL, Zhou Y, Sun J, Crisa A, Ponce de Leon FA, Schwartz JC, Hammond JA, Waldbieser GC, Schroeder SG, Liu GE, Dunham MJ, **Shendure J**, Sonstegard TS, Phillippy AM[#], Van Tassell CP[#], Smith TP[#]. Single-molecule sequencing and chromatin conformation capture enable de novo reference assembly of the domestic goat genome. *Nature Genetics* 2017 Apr;49(4):643-650.

235. Simon MT*, NG BG*, Friederich MW, Wang RY, Boyer M, Kircher M, Collard R, Buckingham KJ, Chang R, **Shendure J**, Nickerson DA, Bamshad MJ, University of Washington Center for Mendelian Genomics, Van Hove JLK, Freeze HH, Abdenur JE[#]. Activation of a cryptic splice site in the mitochondrial elongation factor GFM1 causes combined OXPHOS deficiency. *Mitochondrion* 2017 May;34:84-90.

234. Ramani V, Deng X, Qiu R, Gunderson KL, Steemers FJ, Disteche CM, Noble WS, Duan Z[#], **Shendure J**[#]. Massivley multiplex single-cell Hi-C. *Nature Methods* 2017 Mar;14(3):263-266.

233. Neveling K*, Mensenkamp AR*, Derks R, Kwint M, Ouchene H, Steehouwer M, van Lier B, Bosgoed E, Rikken A, Tychon M, Zafeiropoulou D, Castelein S, Hehir-Kwa J, Tjwan Thung D, Hofste T, Lelieveld SH, Bertens SM, Adan IB, Eijkelenboom A, Tops BB, Tntema H, Stokowy T, Knappskog PM, Hoberg-Vetti H, Steen VM, Boyle E, Martin B, Ligtenberg MJ, **Shendure J**[#], Nelen MR[#], Hoischen A[#]. BRCA Testing by Single-Molecule Molecular Inversion Probes. *Clinical Chemistry* 2017 Jan;63(2):503-512.

232. Dougherty M*, Lazar J*, Klein JC*, Diaz K, Gobillot T, Grunblatt E, Hasle N, Lawrence D, Maurano M, Nelson M, Olson G, Srivatsan S, **Shendure J**, Keene CD, Bird T, Horwitz MS[#], Marshall DA[#]. Genome sequencing in a case of Niemann-Pick type C. *Cold Spring Harbor Molecular Case Studies* 2016 Nov;2(6):001222.

231. Inoue F*, Kircher M*, Martin B, Cooper GM, Witten DM, McManus MT, Ahituv N[#], **Shendure J**[#]. A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. *Genome Research* 2017 Jan;27(1):38-52.

230. Cantsilieris S[#], Stessman HA, **Shendure J**, Eichler EE. Targeted Capture and High-Throughput Sequencing Using Molecular Inversion Probes (MIPs). *Methods in Molecular Biology* 2017;1492:95-106.

229. Hause RJ, Pritchard CC, **Shendure J**[#], Salipante SJ[#]. Classification and characterization of microsatellite instability across 18 cancer types. *Nature Medicine* 2016 Nov;22(11):1342-1350.

228. Session AM*, Uno Y*, Kwon T*, Chapman JA, Toyoda A, Takahashi S, Fukui A, Hikosaka A, Suzuki A, Kondo M, van Heeringen SJ, Quigley I, Heinz S, Ogino H, Ochi H, Hellsten U, Lyons JB, Simakov O, Putnam N, Stites J, Kuroki Y, Tanaka T, Michiue T, Watanabe M, Bogdanovic O, Lister R, Georgiou G, Paranjpe SS, van Kruijsbergen I, Shu S, Carlson J, Kinoshita T, Ohta Y, Mawaribuchi S, Jenkins J, Grimwood J, Schmutz J, Mitros T, Mozaffari SV, Suzuki Y, Haramoto Y, Yamamoto TS, Takagi C, Heald R, Miller K, Haudenschild C, Kitzman J, Nakayama T, Izutsu Y, Robert J, Fortriede J, Burns K, Lotay V, Karimi K, Yasuoka Y, Dichmann DS, Flajnik MF, Houston DW, **Shendure J**, DuPasquier L, Vize PD, Zorn AM, Ito M, Marcotte EM, Wallingford JB, Ito Y, Asashima M, Ueno N, Matsuda Y, Veenstra GJ, Fujiyama A, Harland RM[#], Taira M[#], Rokhsar DS[#]. Genome evolution in the allotetraploid frog Xenopus laevis. *Nature* 2016 Oct 19;538(7625):336-343.

227. Ramani V, Cusanovich DA, Hause RJ, Ma W, Qiu R, Deng X, Blau CA, Disteche CM, Noble WS, **Shendure J**[#], Duan Z[#]. Mapping 3D genome architecture through in situ DNase Hi-C. *Nature Protocols* 2016 Nov;11(11):2104-21.

226. Mirzaa G[#], Timms AE, Conti V, Boyle EA, Girisha KM, Martin B, Kircher M, Olds C, Juusola J, Collins S, Park K, Carter M, Glass I, Krägeloh-Mann I, Chitayat D, Parikh AS, Bradshaw R, Torti E, Braddock S, Burke L, Ghedia S, Stephan M, Stewart F, Prasad C, Napier M, Saitta S, Straussberg R, Gabbett M, O'Connor BC, Keegan CE, Yin LJ, Lai AH, Martin N, McKinnon M, Addor MC, Boccuto L, Schwartz CE, Lanoel A, Conway RL, Devriendt K, Tatton-Brown K, Pierpont ME, Painter M, Worgan L, Reggin J, Hennekam R, Tsuchiya K, Pritchard CC, Aracena M, Gripp KW, Cordisco M, Esch HV, Garavelli L, Curry C, Goriely A, Kayserilli H, **Shendure J**, Graham J Jr, Guerrini R, Dobyns WB. PIK3CA-associated

developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. *JCI Insight* 2016 Jun 16;1(9). pii: e87623.

225. Gasperini M, Starita L, **Shendure J**[#]. The power of multiplexed functional analysis of genetic variants. *Nature Protocols* 2016 Oct;11(10):1782-7.

224. **Shendure J**. Human genomics: A deep dive into genetic variation. *Nature* 2016 Aug 17;536(7616):277-8.

223. Wheeler PG[#], Ng BG, Sanford L, Sutton VR, Bartholomew DW, Pastore MT, Bamshad MJ, Kircher M, Buckingham KJ, Nickerson DA, **Shendure J**, Freeze HH. SRD5A3-CDG: Expanding the phenotype of a congenital disorder of glycosylation with emphasis on adult onset features. *American Journal of Medical Genetics A* 2016 Dec;170(12):3165-3171.

222. Underhill HR[#], Kitzman JO, Hellwig S, Welker NC, Daza R, Baker DN, Gligorich KM, Rostomily RC, Bronner MP, **Shendure J**. Fragment Length of Circulating Tumor DNA. *PLoS Genetics* 2016 Jul 18;12(7):e1006162.

221. Burnicka-Turek O[#], Steimle JD, Huang W, Felker L, Kamp A, Kweon J, Peterson M, Reeves RH, Maslen CL, Gruber PJ, Yang XH, **Shendure J**, Moskowitz IP[#]. Cilia gene mutations cause atrioventricular septal defects by multiple mechanisms. *Human Molecular Genetics* 2016 Jul 15;25(14):3011-3028.

220. **Shendure J**[#], Fields S[#]. Massively Parallel Genetics. *Genetics* 2016 Jun;203(2):617-9.

219. McKenna A*, Findlay GM*, Gagnon JA*, Horwitz MS, Schier AF[#]**, Shendure J[#].** Whole organism lineage tracing by combinatorial and cumulative genome editing. *Science* 2016 May 26;pii: aaf7907. 2016 Jul 29;353(6298):aaf7907.

218. Mirzaa GM*[#], Campbell CD*, Solovieff N, Goold CP, Jansen LA, Menon S, Timms AE, Conti V, Biag JD, Olds C, Boyle EA, Collins S, Ishak G, Poliachik SL, Girisha KM, Yeung KS, Chung BH, Rahikkala E, Gunter SA, McDaniel SS, Macmurdo CF, Bernstein JA, Martin B, Leary RJ, Mahan S, Liu S, Weaver M, Dorschner MO, Jhangiani S, Muzny DM, Boerwinkle E, Gibbs RA, Lupski JR**, Shendure J**, Saneto RP, Novotny EJ, Wilson CJ, Sellers WR, Morrissey MP, Hevner RF, Ojemann JG, Guerrini R, Murphy LO, Winckler W, Dobyns WB. Association of MTOR Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. *JAMA Neurology* 2016 Jul 1;73(7):836-45.

217. Stone BC[#], Kas A, Billman ZP, Fuller DH, Fuller JT, **Shendure J**, Murphy SC[#]. Complex Minigene Library Vaccination for Discovery of Pre-Erythrocytic Plasmodium T Cell Antigens. *PLoS One* 2016 Apr 12;11(4):e0153449.

216. Gordon D*, Huddleston J*, Chaisson MJ*, Hill CM*, Kronenberg ZN*, Munson KM, Malig M, Raja A, Fiddes I, Hillier LW, Dunn C, Baker C, Armstrong J, Diekhans M, Paten B, **Shendure J**, Wilson RK, Haussler D, Chin CS, Eichler EE[#]. Long-read sequence assembly of the gorilla genome. *Science* 2016 Apr 1;352(6281):aae0344.

215. Alazami AM*, Al-Qattan SM*, Faqeih E, Alhashem A, Alshammari M, Alzahrani F, Al-Dosari MS, Patel N, Alsagheir A, Binabbas B, Alzaidan H, Alsiddiky A, Alharbi N, Alfadhel M, Kentab A, Daza RM, Kircher M, **Shendure J**, Hashem M, Alshahrani S, Rahbeeni Z, Khalifa O, Shaheen R, Alkuraya FS[#]. Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. *Human Genetics* 2016 May;135(5):525-40.

214. Ramani V[#], **Shendure J**[#]. Smash and DASH with Cas9. *Genome Biology* 2016 Mar 5;17:42.

213. Ng BG, Shiryaev SA, Rymen D, Eklund EA, Raymond K, Kircher M, Abdenur JE, Alehan F, Midro AT, Bamshad MJ, Barone R, Berry GT, Brumbaugh JE, Buckingham KJ, Clarkson K, Cole FS, O'Connor S, Cooper GM, Van Coster R, Demmer LA, Diogo L, Fay AJ, Ficicioglu C, Fiumara A, Gahl WA, Ganetzky R, Goel H, Harshman LA, He M, Jaeken J, James PM, Katz D, Keldermans L, Kibaek M, Kornberg AJ, Lachlan K, Lam C, Yaplito-Lee J, Nickerson DA, Peters HL, Race V, Régal L, Rush JS, Rutledge SL, **Shendure J**, Souche E, Sparks SE, Trapane P, Sanchez-Valle A, Vilain E, Vøllo A, Waechter CJ, Wang RY, Wolfe LA, Wong DA, Wood T, Yang AC; University of Washington Center for Mendelian

Genomics, Matthijs G, Freeze HH[#]. ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. *Human Mutation* 2016 Jul;37(7):653-60.

212. Kumar A*, Coleman I*, Morrissey C, Zhang X, True LD, Gulati R, Etzioni R, Bolouri H, Montgomery B, White T, Lucas JM, Brown LG, Dumpit RF, DeSarkar N, Higano C, Yu EY, Coleman R, Schultz N, Fang M, Lange PH, **Shendure J**, Vessella RL, Nelson PS[#]. Substantial interindividual and limited intraindividual genomic diversity among tumors from men with metastatic prostate cancer. *Nature Medicine* 2016 Apr;22(4):369-78.
211. Shah K*, Ali RH*, Ansar M*, Lee K, Chishti MS, Abbe I, Li B; University of Washington Center for Mendelian Genomics, Smith JD, Nickerson DA, **Shendure J**, Coucke PJ, Steyaert W, Bamshad MJ, Santos-Cortez RL, Leal SM[#], Ahmad W[#]. Mitral regurgitation as a phenotypic manifestation of nonphotosensitive trichothiodystrophy due to a splice variant in MPLKIP. *BMC Medical Genetics* 2016 Feb 16;17:13.
210. Ramani V, **Shendure J**, Duan Z[#]. Understanding Spatial Genome Organization: Methods and Insights. *Genomics Proteomics Bioinformatics* 2016 Feb;14(1):7-20.
209. Kuang SQ, Medina-Martinez O, Guo DC, Gong L, Regalado ES, Reynolds CL, Boileau C, Jondeau G, Prakash SK, Kwartler CS, Zhu LY, Peters AM, Duan XY, Bamshad MJ,**Shendure J**, Nickerson DA, Santos-Cortez RL, Dong X, Leal SM, Majesky MW, Swindell EC, Jamrich M, Milewicz DM[#]. FOXE3 mutations predispose to thoracic aortic aneurysms and dissections. *Journal of Clinical Investigation* 2016 Mar 1;126(3):948-61.
208. Guo DC, Regalado ES, Gong L, Duan X, Santos-Cortez RL, Arnaud P, Ren Z, Cai B, Hostetler EM, Moran R, Liang D, Estrera A, Safi HJ; University of Washington Center for Mendelian Genomics, Leal SM, Bamshad MJ, **Shendure J**, Nickerson DA, Jondeau G, Boileau C, Milewicz DM[#]. LOX Mutations Predispose to Thoracic Aortic Aneurysms and Dissections. *Circulation Research* 2016 Mar 18;118(6):928-34.
207. Snyder MW*, Kircher M*, Hill AJ, Daza RM**, Shendure J[#]**. Cell-free DNA Comprises an In Vivo Nucleosome Footprint that Informs Its Tissues-Of-Origin.Cell. *Cell* 2016 Jan 14;164(1-2):57-68.
206. Kan M*, Auer PL*, Wang GT*, Bucasas KL, Hooker S, Rodriguez A, Li B, Ellis J, Adrienne Cupples L, Ida Chen YD, Dupuis J, Fox CS, Gross MD, Smith JD, Heard-Costa N, Meigs JB, Pankow JS, Rotter JI, Siscovick D, Wilson JG, **Shendure J**, Jackson R, Peters U, Zhong H, Lin D, Hsu L, Franceschini N, Carlson C, Abecasis G, Gabriel S, Bamshad MJ, Altshuler D, Nickerson DA, North KE, Lange LA, Reiner AP, Leal SM[#]. Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. *European Journal of Human Genetics* 2016 Aug;24(8):1181-7.
205. Snyder MW, Gammill HS[#], **Shendure J**. Copy-Number Variation and False Positive Results of Prenatal Screening. *New England Journal of Medicine* 2015 Dec 24;373(26):2585.
204. Ansar M*, Jan A*, Santos-Cortez RL, Wang X, Suliman M, Acharya A, Habib R, Abbe I, Ali G, Lee K, Smith JD; University of Washington Center for Mendelian Genomics, Nickerson DA, **Shendure J**, Bamshad MJ, Ahmad W[#], Leal SM[#]. Expansion of the spectrum of ITGB6-related disorders to adolescent alopecia, dentogingival abnormalities and intellectual disability. *European Journal of Human Genetics* 2016 Aug;24(8):1223-7.
203. Berletch JB*, Ma W*, Yang F, **Shendure J**, Noble WS, Disteche CM[#], Deng X[#]. Identification of genes escaping X inactivation by allelic expression analysis in a novel hybrid mouse model. *Data Brief* 2015 Nov 3;5:761-9.
202. Ahmad F, Ansar M, Mehmood S, Izoduwa A, Lee K, Nasir A, Abrar M, Mehmood S, Ullah A, Aziz A; University of Washington Center for Mendelian Genomics, Smith JD, **Shendure J**, Bamshad MJ, Nicekrson DA, Santos-Cortez RL, Leal SM, Ahmad W[#]. A novel missense variant in the PNPLA1 gene

underlies congenital ichthyosis in three consanguineous families. *Journal of the European Academy of Dermatology and Venereology* 2016 Dec;30(12):e210-e213.

201. Phadnis N[#], Baker EP, Cooper JC, Frizzell KA, Hsieh E, de la Cruz AF, **Shendure J**, Kitzman JO, Malik HS[#]. An essential cell cycle regulation gene causes hybrid inviability in Drosophila. *Science* 2015 Dec 18;350(6267):1552-5.
200. Schueler M*, Halbritter J*, Phelps IG*, Braun DA, Otto EA, Porath JD, Gee HY, **Shendure J**, O'Roak BJ, Lawson JA, Nabhan MM, Soliman NA, Doherty D, Hildebrandt F[#]. Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. *Journal of Medical Genetics* 2016 Mar;53(3):208-14.
199. Salipante SJ*[#], Adey A*, Thomas A, Lee C, Liu YJ, Kumar A, Lewis AP, Wu D[#], Fromm JR[#], **Shendure J[#]**. Recurrent somatic loss of TNFRSF14 in classical Hodgkin lymphoma. *Genes Chromosomes Cancer* 2016 Mar;55(3):278-87.
198. Shaheen R, Patel N*, Shamseldin H*, Alzahrani F*, Al-Yamany R*, ALMoisheer A, Ewida N, Anazi S, Alnemer M, Elsheikh M, Alfaleh K, Alshammari M, Alhashem A, Alangari AA, Salih MA, Kircher M, Daza RM, Ibrahim N, Wakil SM, Alaqeel A, Altowaijri I, **Shendure J**, Al-Habib A, Faqieh E, Alkuraya FS[#]. Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. *Genetic Medicine* Jul;18(7):686-95.
197. Regalado ES, Guo DC, Santos-Cortez RL, Hostetler E, Bensend TA, Pannu H, Estrera A, Safi H, Mitchell AL, Evans JP, Leal SM, Bamshad M, **Shendure J**, Nickerson DA; University of Washington Center for Mendelian Genomics, Milewicz DM[#]. Pathogenic FBN1 variants in familial thoracic aortic aneurysms and dissections. *Clinical Genetics* 2016 Jun;89(6):719-23.
196. Ullah R, Ansar M, Durrani ZU, Lee K, Santos-Cortez RL, Muhammad D, Ali M, Zia M, Ayub M, Khan S, Smith JD, Nickerson DA, **Shendure J**, Bamshad M, Leal SM, Ahmad W[#]. Novel mutations in the genes TGM1 and ALOXE3 underlying autosomal recessive congenital ichthyosis. *International Journal of Dermatology* 2016 May;55(5):524-30.
195. Klein JC, Lajoie MJ, Schwartz JJ, Strauch EM, Nelson J, Baker D, **Shendure J[#]**. Multiplex pairwise assembly of array-derived DNA oligonucleotides. *Nucleic Acids Research* 2016 Mar 18;44(5):e43.
194. Mirzaa GM[#], Conti V, Timms AE, Smyser CD, Ahmed S, Carter M, Barnett S, Hufnagel RB, Goldstein A, Narumi-Kishimoto Y, Olds C, Collins S, Johnston K, Deleuze JF, Nitschké P, Friend K, Harris C, Goetsch A, Martin B, Boyle EA, Parrini E, Mei D, Tattini L, Slavotinek A, Blair E, Barnett C, **Shendure J,** Chelly J, Dobyns WB, Guerrini R[#]. Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. *Lancet Neurology* 2015 Dec;14(12):1182-95.
193. Rosenberg AB, Patwardhan RP, **Shendure J**, Seelig G[#]. Learning the sequence determinants of alternative splicing from millions of random sequences. *Cell* 2015 Oct 22;163(3):698-711.
192. Ng BG, Raymond K, Kircher M, Buckingham KJ, Wood T, **Shendure J**, Nickerson DA, Bamshad MJ; University of Washington Center for Mendelian Genomics, Wong JT, Monteiro FP, Graham BH, Jackson S, Sparkes R, Scheuerle AE, Cathey S, Kok F, Gibson JB, Freeze HH[#]. Expanding the Molecular and Clinical Phenotype of SSR4-CDG. *Human Mutation* 2015 Nov;36(11):1048-51.
191. Ansar M, Raza SI, Lee K, Irfanullah, Shahi S, Acharya A, Dai H, Smith JD, **Shendure J**, Bamshad MJ, Nickerson DA, Santos-Cortez RL, Ahmad W[#], Leal SM[#]. A homozygous missense variant in type I keratin KRT25 causes autosomal recessive woolly hair. *Journal of Medical Genetics* 2015 Oct;52(10):676-80.
190. Brennan G, Kitzman JO, **Shendure J**, Geballe AP[#]. Experimental Evolution Identifies Vaccinia Virus Mutations in A24R and A35R That Antagonize the Protein Kinase R Pathway and Accompany Collapse of an Extragenic Gene Amplification. *Journal of Virology* 2015 Oct 1;89(19):9986-97.
189. **Shendure J[#]**, Akey JM[#]. The origins, determinants, and consequences of human mutations. *Science* 2015 Sep 25;349(6255):1478-83.

188. Jorth P, Staudinger BJ, Wu X, Hisert KB, Hayden H, Garudathri J, Harding CL, Radey MC, Rezayat A, Bautista G, Berrington WR, Goddard AF, Zheng C, Angermeyer A, Brittnacher MJ, Kitzman J, **Shendure J**, Fligner CL, Mittler J, Aitken ML, Manoil C, Bruce JE, Yahr TL, Singh PK[#]. Regional Isolation Drives Bacterial Diversification within Cystic Fibrosis Lungs. *Cell Host & Microbe* 2015 Sep 9;18(3):307-19.

187. Rehman AU*, Santos-Cortez RL*, Drummond MC, Shahzad M, Lee K, Morell RJ, Ansar M, Jan A, Wang X, Aziz A, Riazuddin S, Smith JD, Wang GT, Ahmed ZM, Gul K, Shearer AE, Smith RJ, **Shendure J**, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics, Hinnant J, Khan SN, Fisher RA, Ahmad W, Friderici KH, Riazuddin S, Friedman TB, Wilch ES, Leal SM. Challenges and solutions for gene identification in the presence of familial locus heterogeneity. *European Journal of Human Genetics* 2015 Sep;23(9):1207-15.

186. Ansar M, Santos-Cortez RL, Saqib MA, Zulfiqar F, Lee K, Ashraf NM, Ullah E, Wang X, Sajid S, Khan FS, Amin-ud-Din M; University of Washington Center for Mendelian Genomics, Smith JD, **Shendure J**, Bamshad MJ, Nickerson DA, Hameed A, Riazuddin S, Ahmed ZM, Ahmad W, Leal SM[#]. Mutation of ATF6 causes autosomal recessive achromatopsia. *Human Genetics* 2015 Sep;134(9):941-50.

185. Bachmann-Gagescu R*, Phelps IG*, Dempsey JC, Sharma VA, Ishak GE, Boyle EA, Wilson M, Marques Lourenço C, Arslan M; University of Washington Center for Mendelian Genomics, **Shendure J**, Doherty D[#]. KIAA0586 is Mutated in Joubert Syndrome. *Human Mutation* 2015 Sep;36(9):831-5.

184. Ramani V, Qiu R, **Shendure J[#]**. High-throughput determination of RNA structure by proximity ligation. *Nature Biotechnology* 2015 Sep;33(9):980-4.

183. Deng X*, Ma W*, Ramani V, Hill A, Yang F, Ay F, Berletch JB, Blau CA, **Shendure J**, Duan Z[#], Noble WS[#], Disteche CM[#]. Bipartite structure of the inactive mouse X chromosome. *Genome Biology* 2015 Aug 7;16:152.

182. Chong JX, Buckingham KJ, Jhangiani SN, Boehm C, Sobreira N, Smith JD, Harrell TM, McMillin MJ, Wiszniewski W, Gambin T, Coban Akdemir ZH, Doheny K, Scott AF, Avramopoulos D, Chakravarti A, Hoover-Fong J, Mathews D, Witmer PD, Ling H, Hetrick K, Watkins L, Patterson KE, Reinier F, Blue E, Muzny D, Kircher M, Bilguvar K, López-Giráldez F, Sutton VR, Tabor HK, Leal SM, Gunel M, Mane S, Gibbs RA, Boerwinkle E, Hamosh A, **Shendure J**, Lupski JR, Lifton RP, Valle D, Nickerson DA; Centers for Mendelian Genomics, Bamshad MJ[#]. The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. *American Journal of Human Genetics* 2015 Aug 6;97(2):199-215.

181. Bachmann-Gagescu R, Dempsey JC, Phelps IG, O'Roak BJ, Knutzen DM, Rue TC, Ishak GE, Isabella CR, Gorden N, Adkins J, Boyle EA, de Lacy N, O'Day D, Alswaid A, Ramadevi A R, Lingappa L, Lourenço C, Martorell L, Garcia-Cazorla À, Ozyürek H, Haliloğlu G, Tuysuz B, Topçu M; University of Washington Center for Mendelian Genomics, Chance P, Parisi MA, Glass IA, **Shendure J**, Doherty D[#]. Joubert syndrome: a model for untangling recessive disorders with extreme genetic heterogeneity. *Journal of Medical Genetics*. 2015 Aug;52(8):514-22.

180. Wheway G, Schmidts M, Mans DA, Szymanska K, Nguyen TM, Racher H, Phelps IG, Toedt G, Kennedy J, Wunderlich KA, Sorusch N, Abdelhamed ZA, Natarajan S, Herridge W, van Reeuwijk J, Horn N, Boldt K, Parry DA, Letteboer SJ, Roosing S, Adams M, Bell SM, Bond J, Higgins J, Morrison EE, Tomlinson DC, Slaats GG, van Dam TJ, Huang L, Kessler K, Giessl A, Logan CV, Boyle EA, **Shendure J**, Anazi S, Aldahmesh M, Al Hazzaa S, Hegele RA, Ober C, Frosk P, Mhanni AA, Chodirker BN, Chudley AE, Lamont R, Bernier FP, Beaulieu CL, Gordon P, Pon RT, Donahue C, Barkovich AJ, Wolf L, Toomes C, Thiel CT, Boycott KM, McKibbin M, Inglehearn CF; UK10K Consortium; University of Washington Center for Mendelian Genomics, Stewart F, Omran H, Huynen MA, Sergouniotis PI, Alkuraya FS, Parboosingh JS, Innes AM, Willoughby CE, Giles RH, Webster AR, Ueffing M, Blacque O, Gleeson JG, Wolfrum U, Beales PL, Gibson T, Doherty D[#], Mitchison HM[#], Roepman R[#], Johnson CA[#]. An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. *Nature Cell Biology* 2015 Aug;17(8):1074-87.

179. Kircher M, **Shendure J#**. Running spell-check to identify regulatory variants. *Nature Genetics* 2015 Aug;47(8):853-5.

178. Santos-Cortez RL#, Chiong CM, Reyes-Quintos MR, Tantoco ML, Wang X, Acharya A, Abbe I, Giese AP, Smith JD, Allen EK, Li B, Cutiongco-de la Paz EM, Garcia MC, Llanes EG, Labra PJ, Gloria-Cruz TL, Chan AL, Wang GT, Daly KA, **Shendure J**, Bamshad MJ, Nickerson DA, Patel JA, Riazuddin S, Sale MM; University of Washington Center for Mendelian Genomics, Chonmaitree T, Ahmed ZM, Abes GT, Leal SM. Rare A2ML1 variants confer susceptibility to otitis media. *Nature Genetics* 2015 Aug;47(8):917-20.

177. Roach DJ, Burton JN, Lee C, Stackhouse B, Butler-Wu SM, Cookson BT, **Shendure J#**, Salipante SJ#. A Year of Infection in the Intensive Care Unit: Prospective Whole Genome Sequencing of Bacterial Clinical Isolates Reveals Cryptic Transmissions and Novel Microbiota. *PLoS Genetics* 2015 Jul 31;11(7):e1005413.

176. Kumar A*, Ryan A*, Kitzman JO, Wemmer N, Snyder MW, Sigurjonsson S, Lee C, Banjevic M, Zarutskie PW, Lewis AP, **Shendure J#**, Rabinowitz M#. Whole genome prediction for preimplantation genetic diagnosis. *Genome Medicine* 2015 Apr 8;7(1):35.

175. Chong JX, Burrage LC, Beck AE, Marvin CT, McMillin MJ, Shively KM, Harrell TM, Buckingham KJ, Bacino CA, Jain M, Alanay Y, Berry SA, Carey JC, Gibbs RA, Lee BH, Krakow D, **Shendure J**, Nickerson DA; University of Washington Center for Mendelian Genomics, Bamshad MJ#. Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. *American Journal of Human Genetics* 2015 May 7;96(5):841-9.

174. Cusanovich DA, Daza R, Adey A, Pliner H, Christiansen L, Gunderson KL, Steemers FJ, Trapnell C, **Shendure J#.** Multiplex single-cell profiling of chromatin accessibility by combinatorial cellular indexing. *Science* 2015 May 22;348(6237):910-4.

173. Snyder MW#, Adey A, Kitzman JO, **Shendure J#**. Haplotype-resolved genome sequencing: experimental methods and applications. *Nature Reviews Genetics* 2015 Jun;16(6):344-58.

172. Varoquaux N, Liachko I, Ay F, Burton JN, **Shendure J**, Dunham MJ, Vert JP, Noble WS#. Accurate identification of centromere locations in yeast genomes using Hi-C. *Nucleic Acids Research* 2015 Jun 23;43(11):5331-9.

171. Weren RD, Ligtenberg MJ, Kets CM, de Voer RM, Verwiel ET, Spruijt L, van Zelst-Stams WA, Jongmans MC, Gilissen C, Hehir-Kwa JY, Hoischen A, **Shendure J**, Boyle EA, Kamping EJ, Nagtegaal ID, Tops BB, Nagengast FM, Geurts van Kessel A, van Krieken JH, Kuiper RP*#, Hoogerbrugge N*. A germline homozygous mutation in the base-excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer. *Nature Genetics* 2015 Jun;47(6):668-71.

170. Fairfield H, Srivastava A*, Ananda G*, Liu R*, Kircher M, Lakshminarayana A, Harris B, Karst SY, Dionne LA, Kane CC, Curtain M, Berry ML, Ward-Bailey PF, Greenstein I, Byers C, Czechanski A, Sharp J, Palmer K, Gudis P, Martin W, Tadenev A, Bogdanik L, Pratt CH, Chang B, Schroeder DG, Cox GA, Cliften P, Milbrandt J, Murray S, Burgess R, Bergstrom DE, Donahue LR, Hamamy H, Masri A, Santoni FA, Makrythanasis P, Antonarakis SE, **Shendure J***, Reinholdt LG*#. Exome sequencing reveal pathogenic mutations in 91 strains of mice with Mendelian disorders. *Genome Research* 2015 Jul;25(7):948-57.

169. Yang F*, Deng X*, Ma W, Berletch JB, Rabaia N, Wei G, Moore JM, Filippova GN, Xu J, Liu Y, Noble WS, **Shendure J**, Disteche CM#. The lncRNA Firre anchors the inactive X chromosome to the nucleolus by binding CTCF and maintains H3K27me3 methylation. *Genome Biology* 2015 Mar 12;16(1):52.

168. Snyder MW*, Simmons LE*, Kitzman JO, Coe BP, Henson JM, Daza RM, Eichler EE, **Shendure J#**, Gammill HS#. Copy Number Variation and False-positives in Prenatal Aneuploidy Screening. *New England Journal of Medicine* 2015 Apr 23;372(17):1639-45.

167. Starita LM, Young DL, Islam M, Kitzman JO, Gullingsrud J, Hause RJ, Fowler DM, Parvin JD, **Shendure J***, Fields S*. Massively Parallel Functional Analysis of BRCA1 RING Domain Variants. *Genetics* 2015 Jun;200(2):413-22.

166. Berletch JB*, Ma W*, Yang F, **Shendure J**, Noble WS, Disteche CM#, Deng X#. Escape from X inactivation varies in mouse tissues. *PLoS Genetics* 2015 Mar 18;11(3):e1005079.

165. Jansen LA#, Mirzaa GM, Ishak GE, O'Roak BJ, Hiatt JB, Roden WH, Gunter SA, Christian SL, Collins S, Adams C, Rivière JB, St-Onge J, Ojemann JG, **Shendure J**, Hevner RF, Dobyns WB. PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. *Brain* 2015 Jun;138(Pt 6):1613-28.

164. Chong JX, McMillin MJ, Shively KM, Beck AE, Marvin CT, Armenteros JR, Buckingham KJ, Nkinsi NT, Boyle EA, Berry MN, Bocian M, Foulds N, Uzielli ML, Haldeman-Englert C, Hennekam RC, Kaplan P, Kline AD, Mercer CL, Nowaczyk MJ, Klein Wassink-Ruiter JS, McPherson EW, Moreno RA, Scheuerle AE, Shashi V, Stevens CA, Carey JC, Monteil A, Lory P, Tabor HK, Smith JD, **Shendure J**, Nickerson DA; University of Washington Center for Mendelian Genomics, Bamshad MJ#. De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. *American Journal of Human Genetics* 2015 Mar 5;96(3):462-73.

163. Carlson KD, Sudmant PH, Press MO, Eichler EE, **Shendure J**, Queitsch C#. MIPSTR: a method for multiplex genotyping of germline and somatic STR variation across many individuals. *Genome Research* 2015 May;25(5):750-61.

162. Stitziel NO, Peloso GM, Abifadel M, Cefalù AB, Fouchier S, Motazacker MM, Tada H, Larach DB, Awan Z, Haller JF, Pullinger CR, Varret M, Rabès JP, Noto D, Tarugi P, Kawashiri MA, Nohara A, Yamagishi M, Risman M, Deo R, Ruel I, **Shendure J**, Nickerson DA, Wilson JG, Rich SS, Gupta N, Farlow DN; NHLBI Grand Opportunity Exome Sequencing Project Family Studies Project Team, Neale BM, Daly MJ, Kane JP, Freeman MW, Genest J, Rader DJ, Mabuchi H, Kastelein JJ, Hovingh GK, Averna MR, Gabriel S, Boileau C, Kathiresan S#. Exome Sequencing in Suspected Monogenic Dyslipidemias. *Circulation: Cardiovascular Genetics* 2015 Apr;8(2):343-50.

161. Kumar A, Boyle EA, Tokita M, Mikheev AM, Sanger MC, Girard E, Silber JR, Gonzalez-Cuyar LF, Hiatt JB, Adey A, Lee C, Kitzman JO, Born DE, Silbergeld DL, Olson JM, Rostomily RC#, **Shendure J#**. Deep sequencing of multiple regions of glial tumors reveals spatial heterogeneity for mutations in clinically relevant genes. *Genome Biology* 2014 Dec 3;15(12):530.

160. D'Gama AM, Geng Y, Couto JA, Martin B, Boyle EA, LaCoursiere CM, Hossain A, Hatem NE, Barry B, Kwiatkowski DJ, Vinters HV, Barkovich AJ, **Shendure J**, Mathern GW, Walsh CA#, Poduri A#. mTOR pathway mutations cause hemimegalencephaly and focal cortical dysplasia. *Annals of Neurology* 2015 Apr;77(4):720-5.

159. Kitzman JO*#, Starita LM#, Lo RS, Fields S, **Shendure J#**. Massively parallel single-amino-acid mutagenesis. *Nature Methods* 2015 Mar;12(3):203-6, 4 p following 206.

158. Guo DC, Gong L, Regalado ES, Santos-Cortez RL, Zhao R, Cai B, Veeraraghavan S, Prakash SK, Johnson RJ, Muilenburg A, Willing M, Jondeau G, Boileau C, Pannu H, Moran R, Debacker J; GenTAC Investigators, National Heart, Lung, and Blood Institute Go Exome Sequencing Project; Montalcino Aortic Consortium, Bamshad MJ, **Shendure J**, Nickerson DA, Leal SM, Raman CS, Swindell EC, Milewicz DM#. MAT2A Mutations Predispose Individuals to Thoracic Aortic Aneurysms. *American Journal of Human Genetics* 2015 Jan 8;96(1):170-7.

157. Rehman AU*, Santos-Cortez RL*, Drummond MC, Shahzad M, Lee K, Morell RJ, Ansar M, Jan A, Wang X, Aziz A, Riazuddin S, Smith JD, Wang GT, Ahmed ZM, Gul K, Shearer AE, Smith RJ, **Shendure J**, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics, Hinnant J, Khan SN, Fisher RA, Ahmad W, Friderici KH, Riazuddin S, Friedman TB, Wilch ES, Leal SM#. Challenges and solutions for gene identification in the presence of familial locus heterogeneity. *European Journal of Human Genetics* 2015 Sep;23(9):1207-15.

156. **Shendure J#**. Life after genetics. *Genome Medicine* 2014 Oct 29;6(10):86. eCollection 2014.
155. Ma W, Ay F, Lee C, Gulsoy G, Deng X, Cook S, Hesson J, Cavanaugh C, Ware CB, Krumm A, Shendure J, Blau CA, Disteche CM, Noble WS#, Duan Z#. Fine-scale chromatin interaction maps reveal the cis-regulatory landscape of human lincRNA genes. *Nature Methods* 2015 Jan;12(1):71-8.
154. O'Roak BJ*, Stessman HA*, Boyle EA, Witherspoon KT, Martin B, Lee C, Vives L, Baker C, Hiatt JB, Nickerson DA, Bernier R, **Shendure J#,** Eichler EE#. Recurrent de novo mutations implicate novel genes underlying simplex autism risk. *Nature Communications* 2014 Nov 24;5:5595.
153. Hause RJ, **Shendure J#**. Genetic variation meets replication origins. *Cell* 2014 Nov 20;159(5):973-4.
152. Salipante SJ*#, Roach DJ*, Kitzman JO, Snyder MW, Stackhouse B, Butler-Wu SM, Lee C, Cookson BT, Shendure J#. Large-scale genomic sequencing of extraintestinal pathogenic Escherichia coli strains. *Genome Research* 2015 Jan;25(1):119-28.
151. Iossifov I*, O'Roak BJ*, Sanders SJ*, Ronemus M*, Krumm N, Levy D, Stessman HA, Witherspoon KT, Vives L, Patterson KE, Smith JD, Paeper B, Nickerson DA, Dea J, Dong S, Gonzalez LE, Mandell JD, Mane SM, Murtha MT, Sullivan CA, Walker MF, Waqar Z, Wei L, Willsey AJ, Yamrom B, Lee YH, Grabowska E, Dalkic E, Wang Z, Marks S, Andrews P, Leotta A, Kendall J, Hakker I, Rosenbaum J, Ma B, Rodgers L, Troge J, Narzisi G, Yoon S, Schatz MC, Ye K, McCombie WR, Shendure J#, Eichler EE#, State MW#, Wigler M#. The contribution of de novo coding mutations to autism spectrum disorder. *Nature* 2014 Nov 13;515(7526):216-21.
150. Birnbaum RY, Patwardhan RP, Kim MJ, Findlay GM, Martin B, Zhao J, Bell RJ, Smith RP, Ku AA, **Shendure J**, Ahituv N#. Systematic dissection of coding exons at single nucleotide resolution supports an additional role in cell-specific transcriptional regulation. *PLoS Genetics* 2014 Oct 23;10(10):e1004592.
149. Adey A, Kitzman JO, Burton JN, Daza R, Kumar A, Christiansen L, Ronaghi M, Amini S, Gunderson KL, Steemers FJ, **Shendure J#**. In vitro, long-range sequence information for de novo genome assembly via transposase contiguity. *Genome Research* 2014 Dec;24(12):2041-9.
148. Amini S, Pushkarev D, Christiansen L, Kostem E, Royce T, Turk C, Pignatelli N, Adey A, Kitzman JO, Vijayan K, Ronaghi M, **Shendure J**, Gunderson KL, Steemers FJ#. Haplotype-resolved whole-genome sequencing by contiguity-preserving transposition and combinatorial indexing. *Nature Genetics* 2014 Oct 19.
147. Cecchi AC, Guo D, Ren Z, Flynn K, Santos-Cortez RL, Leal SM, Wang GT, Regalado ES, Steinberg GK, **Shendure J**, Bamshad MJ; University of Washington Center for Mendelian Genomics, Grotta JC, Nickerson DA, Pannu H, Milewicz DM#. RNF213 Rare Variants in an Ethnically Diverse Population With Moyamoya Disease. *Stroke* 2014 Nov;45(11):3200-7.
146. Pritchard CC#, Morrissey C, Kumar A, Zhang X, Smith C, Coleman I, Salipante SJ, Milbank J, Yu M, Grady WM, Tait JF, Corey E, Vessella RL, Walsh T, **Shendure J**, Nelson PS. Complex MSH2 and MSH6 mutations in hypermutated microsatellite unstable advanced prostate cancer. *Nature Communications* 2014 Sep 25;5:4988.
145. Deriziotis P, O'Roak BJ, Graham SA, Estruch SB, Dimitropoulou D, Bernier RA, Gerdts J, **Shendure J**, Eichler EE, Fisher SE#. De novo TBR1 mutations in sporadic autism disrupt protein functions. *Nature Communications* 2014 Sep 18;5:4954.
144. Coe BP, Witherspoon K, Rosenfeld JA, van Bon BW, Vulto-van Silfhout AT, Bosco P, Friend KL, Baker C, Buono S, Vissers LE, Schuurs-Hoeijmakers JH, Hoischen A, Pfundt R, Krumm N, Carvill GL, Li D, Amaral D, Brown N, Lockhart PJ, Scheffer IE, Alberti A, Shaw M, Pettinato R, Tervo R, de Leeuw N, Reijnders MR, Torchia BS, Peeters H, Thompson E, O'Roak BJ, Fichera M, Hehir-Kwa JY, **Shendure J**, Mefford HC, Haan E, Gécz J, de Vries BB, Romano C, Eichler EE#. Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nature Genetics* 2014 Oct;46(10):1063-71.

143. Kumar A, Dougherty M, Findlay GM, Geisheker M, Klein J, Lazar J, Machkovech H, Resnick J, Resnick R, Salter AI, Talebi-Liasi F, Arakawa C, Baudin J, Bogaard A, Salesky R, Zhou Q, Smith K, Clark JI[#], **Shendure J[#]**, Horwitz MS[#]. Genome sequencing of idiopathic pulmonary fibrosis in conjunction with a medical school human anatomy course. *PLoS One* 2014 Sep 5;9(9):e106744.

142. Findlay GM[*#], Boyle EA*, Hause RJ, Klein JC, Shendure J[#]. Saturation editing of genomic regions by multiplex homology-directed repair. *Nature* 2014 Sep 4;513(7516):120-3.

141. Jamuar SS, Lam AT, Kircher M, D'Gama AM, Wang J, Barry BJ, Zhang X, Hill RS, Partlow JN, Rozzo A, Servattalab S, Mehta BK, Topcu M, Amrom D, Andermann E, Dan B, Parrini E, Guerrini R, Scheffer IE, Berkovic SF, Leventer RJ, Shen Y, Wu BL, Barkovich AJ, Sahin M, Chang BS, Bamshad M, Nickerson DA, **Shendure J**, Poduri A, Yu TW, Walsh CA[#]. Somatic mutations in cerebral cortical malformations. *New England Journal of Medicine* 2014 Aug 21;371(8):733-43.

140. Campbell CD, Mohajeri K, Malig M, Hormozdiari F, Nelson B, Du G, Patterson KM, Eng C, Torgerson DG, Hu D, Herman C, Chong JX, Ko A, O'Roak BJ, Krumm N, Vives L, Lee C, Roth LA, Rodriguez-Cintron W, Rodriguez-Santana J, Brigino-Buenaventura E, Davis A, Meade K, LeNoir MA, Thyne S, Jackson DJ, Gern JE, Lemanske RF Jr, **Shendure J**, Abney M, Burchard EG, Ober C, Eichler EE[#]. Whole-genome sequencing of individuals from a founder population identifies candidate genes for asthma. *PLoS One* 2014 Aug 12;9(8):e104396.

139. Schwartz JJ*, Roach DJ*, Thomas JH, **Shendure J[#]**. Primate evolution of the recombination regulator PRDM9. *Nature Communications* 2014 Jul 8;5:4370.

138. Bernier R, Golzio C, Xiong B, Stessman HA, Coe BP, Penn O, Witherspoon K, Gerdts J, Baker C, Vulto-van Silfhout AT, Schuurs-Hoeijmakers JH, Fichera M, Bosco P, Buono S, Alberti A, Failla P, Peeters H, Steyaert J, Vissers LE, Francescatto L, Mefford HC, Rosenfeld JA, Bakken T, O'Roak BJ, Pawlus M, Moon R, **Shendure J**, Amaral DG, Lein E, Rankin J, Romano C, de Vries BB, Katsanis N, Eichler EE[#]. Disruptive CHD8 mutations define a subtype of autism early in development. *Cell* 2014 Jul 17;158(2):263-76.

137. Laszlo AH, Derrington IM, Ross BC, Brinkerhoff H, Adey A, Nova IC, Craig JM, Langford KW, Samson JM, Daza R, Doering K, **Shendure J**, Gundlach JH[#]. Decoding long nanopore sequencing reads of natural DNA. *Nature Biotechnology* 2014 Aug;32(8):829-33.

136. Nuttle X, Itsara A, **Shendure J**, Eichler EE[#]. Resolving genomic disorder-associated breakpoints within segmental DNA duplications using massively parallel sequencing. *Nature Protocols* 2014 Jun;9(6):1496-513.

135. Boyle EA[#], O'Roak BJ, Martin BK, Kumar A, **Shendure J[#]**. MIPgen: Optimized Modeling and Design of Molecular Inversion Probes for Targeted Resequencing. *Bioinformatics* 2014 Sep 15;30(18):2670-2.

134. Burton JN*, Liachko I*, Dunham MJ, **Shendure J[#]**. Species-Level Deconvolution of Metagenome Assemblies with Hi-C-Based Contact Probability Maps. *G3 (Bethesda)* 2014 May 22;4(7):1339-46.

133. SenGupta DJ, Cummings LA, Hoogestraat DR, Butler-Wu SM, **Shendure J**, Cookson BT, Salipante SJ[#]. Whole Genome Sequencing for High-Resolution Investigation of Methicillin Resistant Staphylococcus aureus Epidemiology and Genome Plasticity. *Journal of Clinical Microbiology* 2014 Aug;52(8):2787-96.

132. MacArthur DG[#], Manolio TA, Dimmock DP, Rehm HL, **Shendure J**, Abecasis GR, Adams DR, Altman RB, Antonarakis SE, Ashley EA, Barrett JC, Biesecker LG, Conrad DF, Cooper GM, Cox NJ, Daly MJ, Gerstein MB, Goldstein DB, Hirschhorn JN, Leal SM, Pennacchio LA, Stamatoyannopoulos JA, Sunyaev SR, Valle D, Voight BF, Winckler W, Gunter C[#]. Guidelines for investigating causality of sequence variants in human disease. *Nature* 2014 Apr 24;508(7497):469-76.

131. Salipante SJ, Fromm JR, **Shendure J**, Wood BL, Wu D[#]. Detection of minimal residual disease in NPM1-mutated acute myeloid leukemia by next-generation sequencing. *Modern Pathology* 2014 Nov; 27(11): 1438-46.

130. McMillin MJ*, Beck AE*, Chong JX, Shively KM, Buckingham KJ, Gildersleeve HI, Aracena MI, Aylsworth AS, Bitoun P, Carey JC, Clericuzio CL, Crow YJ, Curry CJ, Devriendt K, Everman DB, Fryer A, Gibson K, Giovannucci Uzielli ML, Graham JM Jr, Hall JG, Hecht JT, Heidenreich RA, Hurst JA, Irani S, Krapels IP, Leroy JG, Mowat D, Plant GT, Robertson SP, Schorry EK, Scott RH, Seaver LH, Sherr E, Splitt M, Stewart H, Stumpel C, Temel SG, Weaver DD, Whiteford M, Williams MS, Tabor HK, Smith JD, **Shendure J**, Nickerson DA; University of Washington Center for Mendelian Genomics, Bamshad MJ#. Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. *American Journal of Human Genetics* 2014 May 1;94(5):734-44.
129. Carvill GL, Weckhuysen S, McMahon JM, Hartmann C, Møller RS, Hjalgrim H, Cook J, Geraghty E, O'Roak BJ, Petrou S, Clarke A, Gill D, Sadleir LG, Muhle H, von Spiczak S, Nikanorova M, Hodgson BL, Gazina EV, Suls A, **Shendure J**, Dibbens LM, De Jonghe P, Helbig I, Berkovic SF, Scheffer IE#, Mefford HC#. GABRA1 and STXBP1: Novel genetic causes of Dravet syndrome. *Neurology* 2014 Apr 8;82(14):1245-53.
128. Shinsky SA, Hu M, Vought VE, Ng SB, Bamshad MJ, **Shendure J**, Cosgrove MS#. A non-active site SET domain surface crucial for the interaction of MLL1 and the RbBP5-ASH2L heterodimer within MLL family core complexes. *Journal of Molecular Biology* 2014 Jun 12;426(12):2283-99.
127. Knowles MR#, Ostrowski LE, Leigh MW, Sears PR, Davis SD, Wolf WE, Hazucha MJ, Carson JL, Olivier KN, Sagel SD, Rosenfeld M, Ferkol TW, Dell SD, Milla CE, Randell SH, Yin W, Sannuti A, Metjian HM, Noone PG, Noone PJ, Olson CA, Patrone MV, Dang H, Lee HS, Hurd TW, Gee HY, Otto EA, Halbritter J, Kohl S, Kircher M, Krischer J, Bamshad MJ, Nickerson DA, Hildebrandt F, **Shendure J**, Zariwala MA#. Mutations in RSPH1 Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. *American Journal of Respiratory and Critical Care Medicine* 2014 Mar 15;189(6):707-17.
126. Losfeld ME, Ng BG, Kircher M, Buckingham KJ, Turner EH, Eroshkin A, Smith JD, **Shendure J**, Nickerson DA, Bamshad MJ; University of Washington Center for Mendelian Genomics, Freeze HH#. A new congenital disorder of glycosylation caused by a mutation in SSR4, the signal sequence receptor 4 protein of the TRAP complex. *Human Molecular Genetics* 2014 Mar 15;23(6):1602-5.
125. Kircher M*, Witten DM*, Jain P, O'Roak BJ, Cooper GM#, **Shendure J#**. A general framework for estimating the relative pathogenicity of human genetic variants. *Nature Genetics* 2014 Mar;46(3):310-5.
124. Brennan G, Kitzman JO, Rothenburg S, **Shendure J**, Geballe AP#. Adaptive gene amplification as an intermediate step in the expansion of virus host range. *PLoS Pathology* 2014 Mar 13;10(3):e1004002.
123. Santos-Cortez RL, Lee K, Giese AP, Ansar M, Amin-Ud-Din M, Rehn K, Wang X, Aziz A, Chiu I, Hussain Ali R, Smith JD; University of Washington Center for Mendelian Genomics, **Shendure J**, Bamshad M, Nickerson DA, Ahmed ZM, Ahmad W, Riazuddin S, Leal SM#. Adenylate cyclase 1 (ADCY1) mutations cause recessive hearing impairment in humans and defects in hair cell function and hearing in zebrafish. *Human Molecular Genetics* 2014 Jun 15;23(12):3289-98.
122. Taylor SD, Ericson NG, Burton JN, Prolla TA, Silber JR, **Shendure J**, Bielas J#. Targeted Enrichment and High-Resolution Digital Profiling of Mitochondrial DNA Deletions in Human Brain. *Aging Cell* 2014 Feb;13(1):29-38.
121. Boissel S, Jarjour J, Astrakhan A, Adey A, Gouble A, Duchateau P, **Shendure J**, Stoddard BL, Certo MT, Baker D, Scharenberg AM#. megaTALs: a rare-cleaving nuclease architecture for therapeutic genome engineering. *Nucleic Acids Research* 2014 Feb;42(4):2591-601.
120. Milewicz DM#, Regalado ES, **Shendure J**, Nickerson DA, Guo DC. Successes and challenges of using whole exome sequencing to identify novel genes underlying an inherited predisposition for thoracic aortic aneurysms and acute aortic dissections. *Trends in Cardiovascular Medicine* 2014 Feb;24(2):53-60.
119. Krumm N, O'Roak BJ, **Shendure J**, Eichler EE. A de novo convergence of autism genetics and molecular neuroscience. *Trends in Neuroscience* 2014 Feb;37(2):95-105.

118. Rehman AU*, Santos-Cortez RL*, Morell RJ*, Drummond MC, Ito T, Lee K, Khan AA, Basra MA, Wasif N, Ayub M, Ali RA, Raza SI; University of Washington Center for Mendelian Genomics, Nickerson DA, **Shendure J**, Bamshad M, Riazuddin S, Billington N, Khan SN, Friedman PL, Griffith AJ, Ahmad W, Riazuddin S, Leal SM[#], Friedman TB[#]. Mutations in TBC1D24, a Gene Associated With Epilepsy, Also Cause Nonsyndromic Deafness DFNB86. *American Journal of Human Genetics* 2014 Jan 2;94(1):144-52.
117. Tuz K*, Bachmann-Gagescu R*, O'Day DR*, Hua K, Isabella CR, Phelps IG, Stolarski AE, O'Roak BJ, Dempsey JC, Lourenco C, Alswaid A, Bönnemann CG, Medne L, Nampoothiri S, Stark Z, Leventer RJ, Topçu M, Cansu A, Jagadeesh S, Done S, Ishak GE, Glass IA, **Shendure J**, Neuhauss SC, Haldeman-Englert CR, Doherty D[#], Ferland RJ[#]. Mutations in CSPP1 Cause Primary Cilia Abnormalities and Joubert Syndrome with or without Jeune Asphyxiating Thoracic Dystrophy. *American Journal of Human Genetics* 2014 Jan 2;94(1):62-72.
116. He Z, O'Roak BJ, Smith JD, Wang G, Hooker S, Santos-Cortez RL, Li B, Kan M, Krumm N, Nickerson DA, **Shendure J**, Eichler EE, Leal SM[#]. Rare-variant extensions of the transmission disequilibrium test: application to autism exome sequence data. *American Journal of Human Genetics* 2014 Jan 2;94(1):33-46.
115. Prüfer K, Racimo F, Patterson N, Jay F, Sankararaman S, Sawyer S, Heinze A, Renaud G, Sudmant PH, de Filippo C, Li H, Mallick S, Dannemann M, Fu Q, Kircher M, Kuhlwilm M, Lachmann M, Meyer M, Ongyerth M, Siebauer M, Theunert C, Tandon A, Moorjani P, Pickrell J, Mullikin JC, Vohr SH, Green RE, Hellmann I, Johnson PL, Blanche H, Cann H, Kitzman JO, **Shendure J**, Eichler EE, Lein ES, Bakken TE, Golovanova LV, Doronichev VB, Shunkov MV, Derevianko AP, Viola B, Slatkin M[#], Reich D[#], Kelso J, Pääbo S[#]. The complete genome sequence of a Neanderthal from the Altai Mountains. *Nature* 2014 Jan 2;505(7481):43-9.
114. Burton JN[#], Adey A, Patwardhan RP, Qiu R, Kitzman JO, **Shendure J**[#]. Chromosome-scale scaffolding of de novo genome assemblies based on chromatin interactions. *Nature Biotechnology* 2013 Dec;31(12):1119-25.
113. Knowles MR[#], Ostrowski LE, Loges NT, Hurd T, Leigh MW, Huang L, Wolf WE, Carson JL, Hazucha MJ, Yin W, Davis SD, Dell SD, Ferkol TW, Sagel SD, Olivier KN, Jahnke C, Olbrich H, Werner C, Raidt J, Wallmeier J, Pennekamp P, Dougherty GW, Hjeij R, Gee HY, Otto EA, Halbritter J, Chaki M, Diaz KA, Braun DA, Porath JD, Schueler M, Baktai G, Griese M, Turner EH, Lewis AP, Bamshad MJ, Nickerson DA, Hildebrandt F, **Shendure J**, Omran H, Zariwala MA[#]. Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. *American Journal of Human Genetics* 2013 Oct 3;93(4):711-20.
112. Thompson O, Edgley M, Strasbourger P, Flibotte S, Ewing B, Adair R, Au V, Chaudry I, Fernando L, Hutter H, Kieffer A, Lau J, Lee N, Miller A, Raymant G, Shen B, **Shendure J**, Taylor J, Turner E, Hillier L, Moerman DG[#], Waterston RH[#]. The Million Mutation Project: A new approach to genetics in Caenorhabditis elegans. *Genome Research* 2013 Oct;23(10):1749-62.
111. Wang Q, Gu L, Adey A, Radlwimmer B, Wang W, Hovestadt V, Bähr M, Wolf S, **Shendure J**, Eils R, Plass C, Weichenhan D[#]. Tagmentation-based whole-genome bisulfite sequencing. *Nature Protocols* 2013 Oct;8(10):2022-32.
110. Fitzgerald LM*, Kumar A*, Boyle EA, Zhang Y, McIntosh LM, Kolb S, Stott-Miller M, Smith T, Karyadi D, Ostrander EA, Hsu L, **Shendure J**[#], Stanford JL[#]. Germline Missense Variants in the BTNL2 Gene Are Associated with Prostate Cancer Susceptibility. *Cancer Epidemiology, Biomarkers & Prevention* 2013 Sep;22(9):1520-8.
109. Carvill GL, Regan BM, Yendle SC, O'Roak BJ, Lozovaya N, Bruneau N, Burnashev N, Khan A, Cook J, Geraghty E, Sadleir LG, Turner SJ, Tsai MH, Webster R, Ouvrier R, Damiano JA, Berkovic SF, **Shendure J**, Hildebrand MS, Szepetowski P, Scheffer IE[#], Mefford HC[#]. GRIN2A mutations cause epilepsy-aphasia spectrum disorders. *Nature Genetics* 2013 Sep;45(9):1073-6.

108. Smith RP*, Taher L*, Patwardhan RP, Kim MJ, Inoue F, **Shendure J**[#], Ovcharenko I[#], Ahituv N[#]. Massively parallel decoding of mammalian regulatory sequences supports a flexible organizational model. *Nature Genetics* 2013 Sep;45(9):1021-8.
107. Buchovecky CM, Turley SD, Brown HM, Kyle SM, McDonald JG, Liu B, Pieper AA, Huang W, Katz DM, Russell DW, **Shendure J**, Justice MJ[#]. A suppressor screen in Mecp2 mutant mice implicates cholesterol metabolism in Rett syndrome. *Nature Genetics* 2013 Sep;45(9):1013-20.
106. Nuttle X, Huddleston J, O'Roak BJ, Antonacci F, Fichera M, Romano C, **Shendure J**, Eichler EE[#]. Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. *Nature Methods* 2013 Sep;10(9):903-9.
105. Guo DC, Regalado E, Casteel DE, Santos-Cortez RL, Gong L, Kim JJ, Dyack S, Horne SG, Chang G, Jondeau G, Boileau C, Coselli JS, Li Z, Leal SM, **Shendure J**, Rieder MJ, Bamshad MJ, Nickerson DA; GenTAC Registry Consortium; National Heart, Lung, and Blood Institute Grand Opportunity Exome Sequencing Project, Kim C, Milewicz DM[#]. Recurrent Gain-of-Function Mutation in PRKG1 Causes Thoracic Aortic Aneurysms and Acute Aortic Dissections. *American Journal of Human Genetics* 2013 Aug 8;93(2):398-404.
104. Adey A[*#], Burton JN*, Kitzman JO*, Hiatt JB, Lewis AP, Martin BK, Qiu R, Lee C, **Shendure J**[#]. The haplotype-resolved genome and epigenome of the aneuploid HeLa cancer cell line. *Nature* 2013 Aug 8;500(7461):207-11.
103. Laurie MT, Bertout JA, Taylor SD, Burton JN, **Shendure JA**, Bielas JH[#]. Simultaneous digital quantification and fluorescence-based size characterization of massively parallel sequencing libraries. *Biotechniques* 2013 Aug;55(2):61-7.
102. Bradnam KR*[#], Fass JN*, Alexandrov A, Baranay P, Bechner M, Birol I, Boisvert S, Chapman JA, Chapuis G, Chikhi R, Chitsaz H, Chou WC, Corbeil J, Del Fabbro C, Docking TR, Durbin R, Earl D, Emrich S, Fedotov P, Fonseca NA, Ganapathy G, Gibbs RA, Gnerre S, Godzaridis E, Goldstein S, Haimel M, Hall G, Haussler D, Hiatt JB, Ho IY, Howard J, Hunt M, Jackman SD, Jaffe DB, Jarvis E, Jiang H, Kazakov S, Kersey PJ, Kitzman JO, Knight JR, Koren S, Lam TW, Lavenier D, Laviolette F, Li Y, Li Z, Liu B, Liu Y, Luo R, Maccallum I, Macmanes MD, Maillet N, Melnikov S, Naquin D, Ning Z, Otto TD, Paten B, Paulo OS, Phillippy AM, Pina-Martins F, Place M, Przybylski D, Qin X, Qu C, Ribeiro FJ, Richards S, Rokhsar DS, Ruby JG, Scalabrin S, Schatz MC, Schwartz DC, Sergushichev A, Sharpe T, Shaw TI, **Shendure J**, Shi Y, Simpson JT, Song H, Tsarev F, Vezzi F, Vicedomini R, Vieira BM, Wang J, Worley KC, Yin S, Yiu SM, Yuan J, Zhang G, Zhang H, Zhou S, Korf IF[#]. Assemblathon 2: evaluating de novo methods of genome assembly in three vertebrate species. *Gigascience* 2013 Jul 22;2(1):10.
101. Carvill GL, Heavin SB, Yendle SC, McMahon JM, O'Roak BJ, Cook J, Khan A, Dorschner MO, Weaver M, Calvert S, Malone S, Wallace G, Stanley T, Bye AM, Bleasel A, Howell KB, Kivity S, Mackay MT, Rodriguez-Casero V, Webster R, Korczyn A, Afawi Z, Zelnick N, Lerman-Sagie T, Lev D, Møller RS, Gill D, Andrade DM, Freeman JL, Sadleir LG, **Shendure J**, Berkovic SF, Scheffer IE[#], Mefford HC[#]. Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. *Nature Genetics* 2013 Jul;45(7):825-30.
100. Snyder MW, Simmons LE, Kitzman JO, Santillan DA, Santillan MK, Gammill HS[#], **Shendure J**[#]. Noninvasive fetal genome sequencing: a primer. *Prenatal Diagnosis* 2013 Jun;33(6):547-54.
99. Salipante SJ[#], Sengupta DJ, Rosenthal C, Costa G, Spangler J, Sims EH, Jacobs MA, Miller SI, Hoogestraat DR, Cookson BT, McCoy C, Matsen FA, **Shendure J**, Lee CC, Harkins TT, Hoffman NG[#]. Rapid 16S rRNA Next-Generation Sequencing of Polymicrobial Clinical Samples for Diagnosis of Complex Bacterial Infections. *PLoS One* 2013 May 29;8(5):e65226.
98. Hiatt JB[#], Pritchard CC, Salipante SJ, O'Roak BJ, **Shendure J**[#]. Single molecule molecular inversion probes for targeted, high-accuracy detection of low-frequency variation. *Genome Research* 2013 May;23(5):843-54.

97. Deng X, Berletch JB, Ma W, Nguyen DK, Hiatt JB, Noble WS, **Shendure J**, Disteche CM[#]. Mammalian X upregulation is associated with enhanced transcription initiation, RNA half-life, and MOF-mediated H4K16 acetylation. *Developmental Cell* 2013 Apr 15;25(1):55-68.

96. Ng BG, Buckingham KJ, Raymond K, Kircher M, Turner EH, He M, Smith JD, Eroshkin A, Szybowska M, Losfeld ME, Chong JX, Kozenko M, Li C, Patterson MC, Gilbert RD, Nickerson DA, **Shendure J**, Bamshad MJ; University of Washington Center for Mendelian Genomics, Freeze HH[#]. Mosaicism of the UDP-galactose transporter SLC35A2 causes a congenital disorder of glycosylation. *American Journal of Human Genetics* 2013 Apr 4;92(4):632-6.

95. Marneros AG*, Beck AE*, Turner EH*, McMillin MJ, Edwards MJ, Field M, de Macena Sobreira NL, Perez AB, Fortes JA, Lampe AK, Giovannucci Uzielli ML, Gordon CT, Plessis G, Le Merrer M, Amiel J, Reichenberger E, Shively KM, Cerrato F, Labow BI, Tabor HK, Smith JD, **Shendure J**, Nickerson DA, Bamshad MJ[#]; University of Washington Center for Mendelian Genomics. Mutations in KCTD1 cause scalp-ear-nipple syndrome. *American Journal of Human Genetics* 2013 Apr 4;92(4):621-6.

94. Starita LM*, Pruneda JN*, Lo RS, Fowler DM, Kim HJ, Hiatt JB, **Shendure J**, Brzovic PS, Fields S[#], Klevit RE[#]. Activity-enhancing mutations in an E3 ubiquitin ligase identified by high-throughput mutagenesis. *Proceedings of the National Academy of Sciences* 2013 Apr 2;110(14):E1263-72.

93. **Shendure J[#]**. 2012 Curt Stern Award address. *American Journal of Human Genetics* 2013 Mar 7;92(3):340-4.

92. Below JE, Earl DL, Shively KM, McMillin MJ, Smith JD, Turner EH, Stephan MJ, Al-Gazali LI, Hertecant JL, Chitayat D, Unger S, Cohn DH, Krakow D, Swanson JM, Faustman EM, **Shendure J**, Nickerson DA, Bamshad MJ[#]; University of Washington Center for Mendelian Genomics. Whole-genome analysis reveals that mutations in inositol polyphosphate phosphatase-like 1 cause opsismodysplasia. *American Journal of Human Genetics* 2013 Jan 10;92(1):137-43.

91. Knowles MR[#], Leigh MW, Ostrowski LE, Huang L, Carson JL, Hazucha MJ, Yin W, Berg JS, Davis SD, Dell SD, Ferkol TW, Rosenfeld M, Sagel SD, Milla CE, Olivier KN, Turner EH, Lewis AP, Bamshad MJ, Nickerson DA, **Shendure J**, Zariwala MA[#]; Genetic Disorders of Mucociliary Clearance Consortium. Exome sequencing identifies mutations in CCDC114 as a cause of primary ciliary dyskinesia. *American Journal of Human Genetics* 2013 Jan 10;92(1):99-106.

90. McMillin MJ, Below JE, Shively KM, Beck AE, Gildersleeve HI, Pinner J, Gogola GR, Hecht JT, Grange DK, Harris DJ, Earl DL, Jagadeesh S, Mehta SG, Robertson SP, Swanson JM, Faustman EM, Mefford HC, **Shendure J**, Nickerson DA, Bamshad MJ[#]; University of Washington Center for Mendelian Genomics. Mutations in ECEL1 cause distal arthrogryposis type 5D. *American Journal of Human Genetics* 2013 Jan 10;92(1):150-6.

89. Fu W[#], O'Connor TD, Jun G, Kang HM, Abecasis G, Leal SM, Gabriel S, Altshuler D, **Shendure J**, Nickerson DA, Bamshad MJ; NHLBI Exome Sequencing Project, Akey JM[#]. Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. *Nature* 2013 Jan 10;493(7431):216-20.

88. O'Roak BJ, Vives L, Fu W, Egertson JD, Stanaway IB, Phelps IG, Carvill G, Kumar A, Lee C, Ankenman K, Munson J, Hiatt JB, Turner EH, Levy R, O'Day DR, Krumm N, Coe BP, Martin BK, Borenstein E, Nickerson DA, Mefford HC, Doherty D, Akey JM, Bernier R, Eichler EE[#], **Shendure J[#]**. Multiplex Targeted Sequencing Identifies Recurrently Mutated Genes in Autism Spectrum Disorders. *Science* 2012 Dec 21;338(6114):1619-22.

87. Loeb GB*, Khan AA*, Canner D, Hiatt JB, **Shendure J**, Darnell RB, Leslie CS, Rudensky AY[#]. Transcriptome-wide miR-155 Binding Map Reveals Widespread Noncanonical MicroRNA Targeting. *Molecular Cell* 2012 Nov 6.

86. Schwartz JJ, Lee C, Hiatt JB, Adey A, **Shendure J[#]**. Capturing native long-range contiguity by in situ library construction and optical sequencing. *Proceedings of the National Academy of Sciences* 2012 Nov 13;109(46):18749-54.

85. Stone B[#], Rieck M, Rawlings CA, Kas A, **Shendure J**, Jones H, Buckner JH. Identification of novel HLA class II target epitopes for generation of donor-specific T regulatory cells. *Clinical Immunology* 2012 Nov;145(2):153-60.
84. Duan Z[#], Andronescu M, Schutz K, Lee C, **Shendure J**, Fields S, Noble WS, Blau CA. A genome-wide 3C-method for characterizing the three-dimensional architectures of genomes. *Methods* 2012 Nov;58(3):277-88.
83. **Shendure J[#]**, Aiden EL[#]. The expanding scope of DNA sequencing. *Nature Biotechnology* 2012 Nov;30(11):1084-94.
82. Campbell CD, Chong JX, Malig M, Ko A, Dumont BL, Han L, Vives L, O'Roak BJ, Sudmant PH, **Shendure J**, Abney M, Ober C, Eichler EE[#]. Estimating the human mutation rate using autozygosity in a founder population. *Nature Genetics* 2012 Nov;44(11):1277-81.
81. Meyer M*[#], Kircher M*, Gansauge MT, Li H, Racimo F, Mallick S, Schraiber JG, Jay F, Prüfer K, de Filippo C, Sudmant PH, Alkan C, Fu Q, Do R, Rohland N, Tandon A, Siebauer M, Green RE, Bryc K, Briggs AW, Stenzel U, Dabney J, **Shendure J**, Kitzman J, Hammer MF, Shunkov MV, Derevianko AP, Patterson N, Andrés AM, Eichler EE, Slatkin M, Reich D[#], Kelso J, Pääbo S[#]. A high-coverage genome sequence from an archaic Denisovan individual. *Science* 2012 Oct 12;338(6104):222-6.
80. Horani A*, Druley TE*, Zariwala MA, Patel AC, Levinson BT, Van Arendonk LG, Thornton KC, Giacalone JC, Albee AJ, Wilson KS, Turner EH, Nickerson DA, **Shendure J**, Bayly PV, Leigh MW, Knowles MR, Brody SL, Dutcher SK, Ferkol TW[#]. Whole-Exome Capture and Sequencing Identifies HEATR2 Mutation as a Cause of Primary Ciliary Dyskinesia. *American Journal of Human Genetics* 2012 Oct 5;91(4):685-93.
79. Tabor HK[#], Murray JC, Gammill HS, Kitzman JO, Snyder MW, Ventura M, Lewis AP, Qiu R, Simmons LE, Rubens CE, Santillan MK, Eichler EE, Cheng EY, Bamshad MJ, **Shendure J**. Non-invasive fetal genome sequencing: opportunities and challenges. *American Journal of Medical Genetics (A)* 2012 Oct;158A(10):2382-4.
78. Peifer M, Fernández-Cuesta L, Sos ML, George J, Seidel D, Kasper LH, Plenker D, Leenders F, Sun R, Zander T, Menon R, Koker M, Dahmen I, Müller C, Di Cerbo V, Schildhaus HU, Altmüller J, Baessmann I, Becker C, de Wilde B, Vandesompele J, Böhm D, Ansén S, Gabler F, Wilkening I, Heynck S, Heuckmann JM, Lu X, Carter SL, Cibulskis K, Banerji S, Getz G, Park KS, Rauh D, Grütter C, Fischer M, Pasqualucci L, Wright G, Wainer Z, Russell P, Petersen I, Chen Y, Stoelben E, Ludwig C, Schnabel P, Hoffmann H, Muley T, Brockmann M, Engel-Riedel W, Muscarella LA, Fazio VM, Groen H, Timens W, Sietsma H, Thunnissen E, Smit E, Heideman DA, Snijders PJ, Cappuzzo F, Ligorio C, Damiani S, Field J, Solberg S, Brustugun OT, Lund-Iversen M, Sänger J, Clement JH, Soltermann A, Moch H, Weder W, Solomon B, Soria JC, Validire P, Besse B, Brambilla E, Brambilla C, Lantuejoul S, Lorimier P, Schneider PM, Hallek M, Pao W, Meyerson M, Sage J, **Shendure J**, Schneider R, Büttner R, Wolf J, Nürnberg P, Perner S, Heukamp LC, Brindle PK, Haas S, Thomas RK[#]. Integrative genome analyses identify key somatic driver mutations of small-cell lung cancer. *Nature Genetics* 2012 Oct;44(10):1104-10.
77. Schwartz JJ[#], Lee C, **Shendure J[#]**. Accurate gene synthesis with tag-directed retrieval of sequence-verified DNA molecules. *Nature Methods* 2012 Sep;9(9):913-5.
76. Elde NC[#], Child SJ, Eickbush MT, Kitzman JO, Rogers KS, **Shendure J**, Geballe AP, Malik HS[#]. Poxviruses deploy genomic accordions to adapt rapidly against host antiviral defenses. *Cell* 2012 Aug 17;150(4):831-41.
75. Boileau C[#], Guo DC, Hanna N, Regalado ES, Detaint D, Gong L, Varret M, Prakash SK, Li AH, d'Indy H, Braverman AC, Grandchamp B, Kwartler CS, Gouya L, Santos-Cortez RL, Abifadel M, Leal SM, Muti C, **Shendure J**, Gross MS, Rieder MJ, Vahanian A, Nickerson DA, Michel JB; National Heart, Lung, and Blood Institute (NHLBI) Go Exome Sequencing Project, Jondeau G, Milewicz DM[#]. TGFB2 mutations cause familial thoracic aortic aneurysms and dissections associated with mild systemic features of Marfan syndrome. *Nature Genetics* 2012 Jul 8;44(8):916-921.

76. Bamshad MJ[#], **Shendure JA**, Valle D, Hamosh A, Lupski JR, Gibbs RA, Boerwinkle E, Lifton RP, Gerstein M, Gunel M, Mane S, Nickerson DA; on behalf of the Centers for Mendelian Genomics. The Centers for Mendelian Genomics: A new large-scale initiative to identify the genes underlying rare Mendelian conditions. *American Journal of Medical Genetics (A)* 2012 Jul;158A(7):1523-1525.

73. Rivière JB, Mirzaa GM, O'Roak BJ, Beddaoui M, Alcantara D, Conway RL, St-Onge J, Schwartzentruber JA, Gripp KW, Nikkel SM, Worthylake T, Sullivan CT, Ward TR, Butler HE, Kramer NA, Albrecht B, Armour CM, Armstrong L, Caluseriu O, Cytrynbaum C, Drolet BA, Innes AM, Lauzon JL, Lin AE, Mancini GM, Meschino WS, Reggin JD, Saggar AK, Lerman-Sagie T, Uyanik G, Weksberg R, Zirn B, Beaulieu CL; Finding of Rare Disease Genes (FORGE) Canada Consortium, Majewski J, Bulman DE, O'Driscoll M, **Shendure J**, Graham JM Jr, Boycott KM, Dobyns WB[#]. De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. *Nature Genetics* 2012 Jun 24;44(8):934-40.

72. Kitzman JO[#], Snyder MW, Ventura M, Lewis AP, Qiu R, Simmons LE, Gammill HS, Rubens CE, Santillan DA, Murray JC, Tabor HK, Bamshad MJ, Eichler EE, **Shendure J[#]**. Noninvasive whole-genome sequencing of a human fetus. *Science Translational Medicine* 2012 Jun 6;4(137):137ra76.

71. Tabor HK[#], Stock J, Brazg T, McMillin MJ, Dent KM, Yu JH, **Shendure J**, Bamshad MJ. Informed consent for whole genome sequencing: A qualitative analysis of participant expectations and perceptions of risks, benefits, and harms. *American Journal of Medical Genetics (A)* 2012 Jun;158A(6):1310-9.

70. Adey A, **Shendure J[#]**. Ultra-low-input, tagmentation-based whole-genome bisulfite sequencing. *Genome Research* 2012 Jun;22(6):1139-43.

69. Kohane IS[#], **Shendure J[#]**. What's a Genome Worth? *Science Translational Medicine* 2012 May 9;4(133):133fs13.

68. Bernier FP, Caluseriu O, Ng S, Schwartzentruber J, Buckingham KJ, Innes AM, Jabs EW, Innis JW, Schuette JL, Gorski JL, Byers PH, Andelfinger G, Siu V, Lauzon J, Fernandez BA, McMillin M, Scott RH, Racher H; FORGE Canada Consortium, Majewski J, Nickerson DA, **Shendure J**, Bamshad MJ[#], Parboosingh JS[#]. Haploinsufficiency of SF3B4, a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. *American Journal of Human Genetics* 2012 May 4;90(5):925-33.

67. O'Roak BJ, Vives L, Girirajan S, Karakoc E, Krumm N, Coe BP, Levy R, Ko A, Lee C, Smith JD, Turner EH, Stanaway IB, Vernot B, Malig M, Baker C, Reilly B, Akey JM, Borenstein E, Rieder MJ, Nickerson DA, Bernier R, **Shendure J[#]**, Eichler EE[#]. Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. *Nature* 2012 Apr 4;485(7397):246-50.

66. Patwardhan RP*, Hiatt JB*, Witten DM, Kim MJ, Smith RP, May D, Lee C, Andrie JM, Lee SI, Cooper GM, Ahituv N[#], Pennacchio LA[#], **Shendure J[#]**. Massively parallel functional dissection of mammalian enhancers in vivo. *Nature Biotechnology* 2012 Feb 26;30(3):265-70.

65. Rivière JB*, van Bon BW*, Hoischen A, Kholmanskikh SS, O'Roak BJ, Gilissen C, Gijsen S, Sullivan CT, Christian SL, Abdul-Rahman OA, Atkin JF, Chassaing N, Drouin-Garraud V, Fry AE, Fryns JP, Gripp KW, Kempers M, Kleefstra T, Mancini GM, Nowaczyk MJ, van Ravenswaaij-Arts CM, Roscioli T, Marble M, Rosenfeld JA, Siu VM, de Vries BB, **Shendure J**, Verloes A, Veltman JA, Brunner HG, Ross ME, Pilz DT[#], Dobyns WB[#]. De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. *Nature Genetics* 2012 Feb 26;44(4):440-4, S1-2.

64. Hondowicz BD, Schwedhelm KV, Kas A, Tasch MA, Rawlings C, Ramchurren N, McIntosh M, D'Amico LA, Sanda S, Standifer NE, **Shendure J**, Stone B[#]. Discovery of T cell antigens by high-throughput screening of synthetic minigene libraries. *PLoS One* 2012;7(1):e29949.

63. Deng X, Hiatt JB, Nguyen DK, Ercan S, Sturgill D, Hillier LW, Schlesinger F, Davis CA, Reinke VJ, Gingeras TR, **Shendure J**, Waterston RH, Oliver B, Lieb JD, Disteche CM[#]. Evidence for compensatory upregulation of expressed X-linked genes in mammals, Caenorhabditis elegans and Drosophila melanogaster. *Nature Genetics* 2011 Oct 23;43(12):1179-85.

62. Kumar A, White TA, Mackenzie AP, Clegg N, Lee C, Dumpit RF, Coleman I, Ng SB, Salipante SJ, Rieder MJ, Nickerson DA, Corey E, Lange PH, Morrissey C, Vessella RL, Nelson PS[#], **Shendure J**[#]. Exome sequencing identifies a spectrum of mutation frequencies in advanced and lethal prostate cancers. *Proceedings of the National Academy of Sciences* 2011 Oct 11;108(41):17087-92.
61. George RD[#], McVicker G, Diederich R, Ng SB, Mackenzie AP, Swanson WJ, **Shendure J**[#], Thomas JH[#]. Trans genomic capture and sequencing of primate exomes reveals new targets of positive selection. *Genome Research* 2011 Oct;21(10):1686-94.
60. Ventura M, Catacchio CR, Alkan C, Marques-Bonet T, Sajjadian S, Graves TA, Hormozdiari F, Navarro A, Malig M, Baker C, Lee C, Turner EH, Chen L, Kidd JM, Archidiacono N, **Shendure J**, Wilson RK, Eichler EE[#]. Gorilla genome structural variation reveals evolutionary parallelisms with chimpanzee. *Genome Research* 2011 Oct;21(10):1640-9.
59. **Shendure JA**, Porreca GJ, Church GM, Gardner AF, Hendrickson CL, Kieleczawa J, Slatko BE. Overview of DNA sequencing strategies. *Current Protocols in Molecular Biology* 2011 Oct; Chapter 7: Unit7.1.
58. Nelson FK, Snyder M, Gardner AF, Hendrickson CL, **Shendure JA,** Porreca GJ, Church GM, Ausubel FM, Ju J, Kieleczawa K, Slatko BE. Introduction and Historical Overview of DNA Sequencing. *Current Protocols in Molecular Biology* 2011 Oct; Chapter 7: Unit7.0.
57. Bamshad MJ[#], Ng SB, Bigham AW, Tabor HK, Emond MJ, Nickerson DA, **Shendure J**[#]. Exome sequencing as a tool for Mendelian disease gene discovery. *Nature Reviews Genetics* 2011 Sep 27;12(11):745-55.
56. **Shendure J**[#]**.** Next-generation human genetics. *Genome Biology* 2011 Sep 14;12(9):408.
55. Fairfield H, Gilbert GJ, Barter M, Corrigan RR, Curtain M, Ding Y, D'Ascenzo M, Gerhardt DJ, He C, Huang W, Richmond T, Rowe L, Probst FJ, Bergstrom DE, Murray SA, Bult C, Richardson J, Kile BT, Gut I, Hager J, Sigurdsson S, Mauceli E, Di Palma F, Lindblad-Toh K, Cunningham ML, Cox TC, Justice MJ, Spector MS, Lowe SW, Albert T, Donahue LR, Jeddeloh J, **Shendure J**, Reinholdt LG[#]. Mutation discovery in mice by whole exome sequencing. *Genome Biology* 2011 Sep 14;12(9):R86.
54. Regalado ES, Guo DC, Villamizar C, Avidan N, Gilchrist D, McGillivray B, Clarke L, Bernier F, Santos-Cortez RL, Leal SM, Bertoli-Avella AM, **Shendure J**, Rieder MJ, Nickerson DA; NHLBI GO Exome Sequencing Project, Milewicz DM[#]. Exome Sequencing Identifies SMAD3 Mutations as a Cause of Familial Thoracic Aortic Aneurysm and Dissection With Intracranial and Other Arterial Aneurysms. *Circulation Research* 2011 Sep 2;109(6):680-6.
53. Cooper GM[#], **Shendure J**[#]**.** Needles in stacks of needles: finding disease-causal variants in a wealth of genomic data. *Nature Reviews Genetics* 2011 Aug 18;12(9):628-40.
52. Cosart T[#], Beja-Pereira A[#], Chen S, Ng SB, **Shendure J**, Luikart G. Exome-wide DNA capture and next generation sequencing in domestic and wild species. *BMC Genomics* 2011 Jul 5;12:347.
51. Hannibal MC*, Buckingham KJ*, Ng SB*, Ming JE, Beck AE, McMillin MJ, Gildersleeve HI, Bigham AW, Tabor HK, Mefford HC, Cook J, Yoshiura K, Matsumoto T, Matsumoto N, Miyake N, Tonoki H, Naritomi K, Kaname T, Nagai T, Ohashi H, Kurosawa K, Hou JW, Ohta T, Liang D, Sudo A, Morris CA, Banka S, Black GC, Clayton-Smith J, Nickerson DA, Zackai EH, Shaikh TH, Donnai D, Niikawa N, **Shendure J**, Bamshad MJ[#]. Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. *American Journal of Medical Genetics* 2011 Jul;155A(7):1511-6.
50. O'Roak BJ, Deriziotis P, Lee C, Vives L, Schwartz JJ, Girirajan S, Karakoc E, Mackenzie AP, Ng SB, Baker C, Rieder MJ, Nickerson DA, Bernier R, Fisher SE, **Shendure J**[#], Eichler EE[#]. Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. *Nature Genetics* 2011 Jun;43(6):585-9.
49. Kumar A, **Shendure J**, Nelson PS[#]. Genome interrupted: sequencing of prostate cancer reveals the importance of chromosomal rearrangements. *Genome Medicine* 2011 Apr 19;3(4):23.

48. Muthappan V*, Lee A*, Lamprecht T, Akileswaran L, Dintzis S, Lee C, Magrini V, Mardis E, **Shendure J**, Van Gelder R#. Biome representational in silico karyotyping. *Genome Research* 2011 Apr;21(4):626-33.
47. Gallagher LA, **Shendure J**, Manoil C#. Genome-Scale Identification of Resistance Functions in Pseudomonas aeruginosa Using Tn-seq. *MBio* 2011 Jan 18;2(1). pii: e00315-10.
46. Kitzman JO#, Mackenzie AP, Adey A, Hiatt JB, Patwardhan RP, Sudmant PH, Ng SB, Alkan C, Qiu R, Eichler EE, **Shendure J#**. Haplotype-resolved genome sequencing of a Gujarati Indian individual. *Nature Biotechnology* 2011 Jan;29(1):59-63.
45. Rios J, Stein E, **Shendure J**, Hobbs HH#, Cohen JC#. Identification by whole-genome resequencing of gene defect responsible for severe hypercholesterolemia. *Human Molecular Genetics* 2010 Nov 15;19(22):4313-8.
44. Sudmant PH*, Kitzman JO*, Antonacci F, Alkan C, Malig M, Tsalenko A, Sampas N, Bruhn L, **Shendure J**; 1000 Genomes Project, Eichler EE#. Diversity of human copy number variation and multicopy genes. *Science* 2010 Oct 29;330(6004):641-6.
43. Ng SB#, Nickerson DA, Bamshad MJ, **Shendure J**. Massively parallel sequencing and rare disease. *Human Molecular Genetics* 2010 Oct 15;19(R2):R119-24.
42. Ng SB*, Bigham AW*, Buckingham KJ, Hannibal MC, McMillin MJ, Gildersleeve HI, Beck AE, Tabor HK, Cooper GM, Mefford HC, Lee C, Turner EH, Smith JD, Rieder MJ, Yoshiura KI, Matsumoto N, Ohta T, Niikawa N, Nickerson DA, Bamshad MJ#, **Shendure J#**. Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. *Nature Genetics* 2010 Sep;42(9):790-3.
41. Igartua C, Turner EH, Ng SB, Hodges E, Hannon GJ, Bhattacharjee A, Rieder MJ, Nickerson DA, **Shendure J#**. Targeted enrichment of specific regions in the human genome by array hybridization. *Current Protocols in Human Genetics* 2010 Jul; Chapter 18: Unit 18.3.
40. Duan Z*, Andronescu M*, Schutz K, McIlwain S, Kim YJ, Lee C, **Shendure J**, Fields S, Blau CA#, Noble WS#. A three-dimensional model of the yeast genome. *Nature* 2010 May 20;465(7296):363-7.
39. Yang F, Babak T, **Shendure J**, Disteche CM#. Global survey of escape from X inactivation by RNA-sequencing in mouse. *Genome Research* 2010 May;20(5):614-22.
38. Roach JC*, Glusman G*, Smit AF*, Huff CD*, Hubley R, Shannon PT, Rowen L, Pant KP, Goodman N, Bamshad M, **Shendure J**, Drmanac R, Jorde LB#, Hood L#, Galas DJ. Analysis of Genetic Inheritance in a Family Quartet by Whole-Genome Sequencing. *Science* 2010 Apr 30;328(5978):636-9.
37. Cooper GM#, Goode DL, Ng SB, Sidow A, Bamshad MJ, **Shendure J**, Nickerson DA. Single-nucleotide evolutionary constraint scores highlight disease-causing mutations. *Nature Methods* 2010 Apr;7(4):250-1.
36. Mamanova L, Coffey AJ, Scott CE, Kozarewa I, Turner EH, Kumar A, Howard E, **Shendure J#**, Turner DJ#. Target-enrichment strategies for next-generation sequencing. *Nature Methods* 2010 Feb;7(2):111-8.
35. Hiatt JB*#, Patwardhan RP*, Turner EH, Lee C, **Shendure J#**. Parallel, tag-directed assembly of locally derived short sequence reads. *Nature Methods* 2010 Feb;7(2):119-22.
34. Adey A* Morrison HG*, Asan*, Xun X*, Kitzman JO, Turner EH, Stackhouse B, Mackenzie AP, Caruccio NC, Zhang X#, **Shendure J#**. Rapid, low-input, low-bias construction of shotgun fragment libraries by high-density in vitro transposition. *Genome Biology* 2010;11(12):R119.
33. Ng SB*, Buckingham KJ*, Lee C, Bigham AW, Tabor HK, Dent KM, Huff CD, Shannon PT, Jabs EW, Nickerson DA, **Shendure J#,** Bamshad MJ#. Exome sequencing identifies the cause of a mendelian disorder. *Nature Genetics* 2010 Jan;42(1):30-5.
32. Thomas JH#, Emerson RO, **Shendure J**. Extraordinary molecular evolution in the PRDM9 fertility gene. *PLoS One* 2009 Dec 30;4(12):e8505.
31. Patwardhan RP#, Lee C, Litvin O, Young DL, Pe'er D, **Shendure J#.** High-resolution analysis of DNA regulatory elements by synthetic saturation mutagenesis. *Nature Biotechnology* 2009 Dec;27(12):1173-5.

30. Vasta V, Ng SB, Turner EH, **Shendure J**[#]**,** Hahn SH[#]. Next generation sequence analysis for mitochondrial disorders. *Genome Medicine* 2009 Oct 23;1(10):100.

29. Ng SB[#], Turner EH, Robertson PD, Flygare SD, Bigham AW, Lee C, Shaffer T, Wong M, Bhattacharjee A, Eichler EE, Bamshad M, Nickerson DA, **Shendure J**[#]. Targeted capture and massively parallel sequencing of 12 human exomes. *Nature* 2009 Sep 10;461(7261):272-6.

28. **Shendure J,** Stewart CJ. Cancer genomes on a shoestring budget. *New England Journal of Medicine* 2009 Jun 25;360(26):2781-3.

27. Brkanac Z[#], Spencer D, **Shendure J**, Robertson PD, Matsushita M, Vu T, Bird TD, Olson MV, Raskind WH. IFRD1 is a candidate gene for SMNA on chromosome 7q22-q23. *American Journal of Human Genetics* 2009 May;84(5):692-7.

26. Turner EH, Lee C, Ng SB, Nickerson DA, **Shendure J**[#]**.** Massively parallel exon capture and library-free resequencing across 16 genomes. *Nature Methods* 2009 May;6(5):315-6.

25. Turner EH, Ng SB, Nickerson DA, **Shendure J**. Methods for genomic partitioning. *Annual Reviews in Genomics and Human Genetics* 2009;10:263-84.

24. **Shendure J**[#], Ji H[#]. Next generation DNA sequencing. *Nature Biotechnology* 2008 Oct;26(10):1135-45.

23. **Shendure J**[#]. The beginning of the end for microarrays? *Nature Methods* 2008 Jul;5(7):585-7.

22. Higgins AW, Alkuraya FS, Bosco AF, Brown KK, Bruns GA, Donovan DJ, Eisenman R, Fan Y, Farra CG, Ferguson HL, Gusella JF, Harris DJ, Herrick SR, Kelly C, Kim HG, Kishikawa S, Korf BR, Kulkarni S, Lally E, Leach NT, Lemyre E, Lewis J, Ligon AH, Lu W, Maas RL, MacDonald ME, Moore SD, Peters RE, Quade BJ, Quintero-Rivera F, Saadi I, Shen Y, **Shendure J**, Williamson RE, Morton CC[#]. Characterization of apparently balanced chromosomal rearrangements from the developmental genome anatomy project. *American Journal of Human Genetics* 2008 Mar;82(3):712-22.

21. **Shendure JA**, Porreca GJ, Church GM. Overview of DNA sequencing strategies. *Current Protocols in Molecular Biology* 2008 Jan; Chapter 7: Unit 7.1.

20. **Shendure J,** Porreca GJ, Church GM. Polony Sequencing, in Kim S, Tang H, Mardis ER, eds. *Genome Sequencing Technology and Algorithms*, 2008.

19. Porreca GJ*, Zhang K*, Li JB, Xie B, Austin D, Vassallo SL, LeProust EM, Peck BJ, Emig CJ, Dahl F, Gao Y, Church GM*[#], **Shendure J***[#]. Multiplex Amplification of Large Sets of Human Exons. *Nature Methods* 2007 Nov;4(11):931-6.

18. Moskowitz I, Kim JB, Moore M, Wolf C, Peterson MA, **Shendure J**, Norbrega M, Yokota Y, Berul C, Izumo S, Seidman JG*, Seidman CE*[#]. A Genetic Pathway Including Id2, Tbx5, and Nkx2-5 Required for Cardiac Conduction System Development. *Cell* 2007 Jun 29;129(7):1365-76.

17. Turner DJ, **Shendure J**, Porreca G, Church G, Green P, Tyler-Smith C, Hurles ME[#]. Assaying chromosomal inversions by single-molecule haplotyping. *Nature Methods* 2006 Jun;3(6):439-45.

16. Zhang K[#], Zhu J, **Shendure J**, Porreca GJ, Aach JD, Mitra RD, Church GM[#]. Polony haplotyping of individual human chromosome molecules. *Nature Genetics* 2006 Mar;38(3):382-7.

15. Porreca GJ, **Shendure J**, Church GM. Polony DNA Sequencing. *Current Protocols in Molecular Biology* 2006. 7.8.1-7.8.22.

14. **Shendure J***[#], Porreca GJ*[#], Reppas NB, Lin X, McCutcheon JP, Rosenbaum AM, Wang MD, Zhang K, Mitra RD, Church GM. Accurate Multiplex Polony Sequencing of an Evolved Bacterial Genome. *Science* 2005 Sep 9;309(5741):1728-32.

13. Zhu Z[#], **Shendure J**, Church GM[#]. Discovering functional transcription-factor combinations in the human cell cycle. *Genome Research* 2005 Jun;15(6):848-55.

12. **Shendure J**, Mitra RD, Varma C, Church GM[#]. Advanced sequencing technologies: methods and goals. *Nature Reviews Genetics* 2004 May;5(5):335-44.

11. Mitra RD, **Shendure J**, Olejnik J, Edyta-Krzymanska-Olejnik, Church GM[#]. Fluorescent in situ sequencing on polymerase colonies. *Analytical Biochemistry* 2003 Sep 1;320(1):55-65.
10. Zhu J*, **Shendure J***, Mitra RD, Church GM[#]. Single molecule profiling of alternative pre-mRNA splicing. *Science* 2003 Aug 8;301(5634):836-8.
9. Mitra RD, Butty VL, **Shendure J**, Williams BR, Housman DE, Church GM[#]. Digital genotyping and haplotyping with polymerase colonies. *Proceedings of the National Academy of Sciences* 2003 May 13;100(10):5926-31.
8. **Shendure J**, Church GM[#]. Computational discovery of sense-antisense transcription in the human and mouse genomes. *Genome Biology* 2002 Aug 22;3(9):RESEARCH0044.
7. Weber G*, **Shendure J***, Tanenbaum DM, Church GM, Meyerson M[#]. Identification of foreign gene sequences by transcript filtering against the human genome. *Nature Genetics* 2002 Feb;30(2):141-2.
6. Badarinarayana V, Estep PW 3rd, **Shendure J**, Edwards J, Tavazoie S, Lam F, Church GM[#]. Selection analyses of insertional mutants using subgenic-resolution arrays. *Nature Biotechnology* 2001 Nov;19(11):1060-5.
5. Aach J*, Bulyk ML, Church GM[#], Comander J, Derti A, **Shendure J***. Computational comparison of two draft sequences of the human genome. *Nature* 2001 Feb 15;409(6822):856-9.
4. Liang X[#], Munshi S, **Shendure J**, Mark G 3rd, Davies ME, Freed DC, Montefiori DC, Shiver JW. Epitope insertion into variable loops of HIV-1 gp120 as a potential means to improve immunogenicity of viral envelope protein. *Vaccine* 1999 Jul 16;17(22):2862-72.
3. Peirce JL*, Derr R*, **Shendure J**, Kolata T, Silver LM[#]. A major influence of sex-specific loci on alcohol preference in C57Bl/6 and DBA/2 inbred mice. *Mammalian Genome* 1998 Dec;9(12):942-8.
2. **Shendure J***, Melo JA*, Pociask K, Derr R, Silver LM[#]. Sex-restricted non-Mendelian inheritance of mouse chromosome 11 in the offspring of crosses between C57BL/6J and (C57BL/6J x DBA/2J)F1 mice. *Mammalian Genome* 1998 Oct;9(10):812-5.
1. Melo JA, **Shendure J**, Pociask K, Silver LM[#]. Identification of sex-specific quantitative trait loci controlling alcohol preference in C57BL/ 6 mice. *Nature Genetics* 1996 Jun;13(2):147-53.

Active Research Support

Howard Hughes Medical Institute 09/01/15 – 08/31/20
Investigator Award
Role: PI

Allen Frontiers Foundation 09/01/17 – 08/31/21
Allen Discovery Center for Cell Lineage (MPI: Shendure, Elowitz, Schier *et al.*)
Role: PI, Director

1R01HG010632 (NIH/NHGRI) 08/01/19 – 07/31/23
Versatile, exponentially scalable methods for single cell molecular profiling (MPI: Shendure, Trapnell)
Role: PI (MPI award)

1U54HL145611 (NIH/OD) 09/20/18 – 08/31/22
A spatially resolved molecular atlas of human endothelium (MPI: Shendure, Tapnell, Cai)
Role: PI (MPI award)

1R01GH009136 (NIH/NHGRI) 04/01/17 – 01/31/21
Predictive Modeling of Alternative Splicing and Polyadenylation from Millions of Random Sequences (MPI: Seelig, Shendure)

Role: PI (MPI award)

1UM1HG009408 (NIH/NHGRI) 02/01/17 – 01/31/21
Massively parallel reporter assays and genome editing of ENCODE predicted regulatory elements (MPI: Ahituv, Shendure)
Role: PI (MPI award)

1U54DK107979 (NIH/OD) 09/30/15 – 07/31/20
University of Washington Center for Nuclear Organization and Function (MPI: Noble, Shendure)
Role: PI (MPI award)

1R01HD089679 (NIH/NICHHD) 09/20/16 – 07/31/21
Placentomics using a novel method to isolate circulating placental derivatives (MPI: Stayton, Chiu, Seglin Gammil)
Role: Co-Investigator

TCPA-2017-04 (NIH/NIDDK) 05/01/17 – 04/30/19
Tethered nuclease strategies for in situ mapping of 3D nuclear organization (MPI: Henikoff, Shendure, Noble)
Role: PI (MPI award)

Completed Research Support

1R01HL130996 (NIH/NHLBI) 02/08/16 – 01/31/18
Mosaic: post-zygotic mutations in vascular and related developmental disorders (Dobyns)
Role: Co-Investigator

1R01DK103667 (NIH/NIDDK) 08/01/15 – 06/30/18
Functional assessment of distal regulatory SNPs associated with type 1 diabetes (Hawkins)
Role: Co-Investigator

1DP1HG007811 (NIH/OD) 09/23/13 – 07/31/18
Interpreting genetic variants of uncertain significance (Shendure)
Role: PI

5R01CA197139 (NIH/NHGRI) 03/01/15 – 02/28/18
Integrative interpretation of the organismal consequences of non-coding variation (MPI: Cooper, Shendure)
Role: PI (MPI award)

Paul G. Allen Family Foundation 04/01/14 – 04/01/18
Cell lineage defined by mitotic recombination (MPI: Horwitz, Shendure)
Role: PI (MPI award)

ETOP2013 (DOE/JGI) 10/01/13 – 09/30/15
Accurate gene synthesis with tag-directed retrieval of sequence-verified DNA molecules (Shendure)
Role: PI

1R01MH101221 (NIH/NIMH) 08/01/13 – 06/30/17
Sporadic Mutations and Autism Spectrum Disorders (Eichler)
Role: Co-Investigator

1R01CA160674 (NIH/NCI) 06/06/12 – 03/31/17
Clonally Expanded Mutations Identify Cancer Precursors in Chronic Inflammation (MPI: Loeb, Brentnall)
Role: Co-Investigator

1R01HG006768 (NIH/NHGRI) 04/01/12 – 03/31/15
Massively parallel, in vivo functional testing of regulatory elements (Ahituv, Shendure)
Role: PI (MPI award)

SFARI 191889EE (Simons Foundation) 01/01/12 – 12/31/13
Whole Exome Sequencing of Simons Simplex Collection Quads (Eichler)
Role: Co-investigator

1U54HG006493 (NIH/NHGRI) 12/05/11 – 11/30/15
UW Center for Mendelian Genomics (MPI: Bamshad, Nickerson, Shendure)
Role: PI (MPI award)

University of Washington Cystic Fibrosis Foundation 10/01/11 – 09/30/13
Studying Cystic Fibrosis Infections Using Massively Parallel Sequencing Technology (Shendure)
Role: PI

1R01HL110879-01 (NIH/NHLBI) 09/01/11 – 05/31/15
Investigating bacterial-host interactions driving CF Pulmonary Exacerbations (MPI: Bruce, Singh)
Role: Co-investigator

1R01HG006283 (NIH/NHGRI) 08/15/11 – 05/31/17
Massively parallel contiguity mapping (Shendure)
Role: PI

1R21CA160080 (NCI/NIH) 07/01/11 – 08/31/14
Ultrasensitive identification and precise quantitation of low frequency somatic mutations by molecular counting (Shendure)
Role: PI

5R01HG004348 (NIH/NHGRI) 07/01/11 – 06/30/12
Advances in Computational Gene Finding (Korf)
Role: Co-investigator

1R011AG039700 (NIH/NIMH) 05/01/11 – 04/30/16
Next Generation Mendelian Genetics in Familial Alzheimer Disease (Brkanac)
Role: Co-investigator

5U54AI057141-08REV (NIH/NIAID) 03/01/11 – 02/28/14
NW Research Center for Excellence in Biodefense and Emerging Infectious Diseases (Miller)
Role: PI of Developmental Project

SFARI 191889 (Simons Foundation) 12/01/10 – 11/30/11
Exome Sequencing of Simons Simplex Collection (SSC) Trios (Eichler)
Role: Co-investigator

1RC2HG005921 (NIH/NHGRI) 08/20/10 – 01/31/12
A Genome-wide Mutation Resource for C. elegans (Waterston)

Role: Co-investigator

W81XWH-10-1-0589 (Department of Defense) 07/01/10 – 08/14/13
Global Characterization of Protein Altering Mutations in Prostate Cancer (Shendure)
Role: PI (synergy award with Nelson at FHCRC)

Young Investigator Award (Prostate Cancer Foundation) 04/01/10 – 03/31/13
Methods & Tools for Next-Generation Analysis of Prostate Cancer Genomes (Shendure)
Role: PI

5R01NS069719 (NIH/NINDS) 04/01/10 – 03/31/14
Next Generation Gene Discovery in Neurogenetics (Raskind)
Role: Co-investigator

5R01NS069605 (NIH/NINDS) 02/15/10 – 02/14/14
A Genomic Approach to Epilepsy (Mefford)
Role: Co-investigator

1I01BX000531 (Department of Veterans Affairs) 10/01/09 – 09/30/13
Genetic Risk Factors for Parkinson's Disease (Zabetian)
Role: Consultant

5R01HD065285 (NIH/NICHD) 09/30/09 – 08/31/12
Genomic Identification of Autism Loci (Eichler)
Role: Co-investigator

1RC2CA148317 (NIH/NCI) 09/30/09 – 09/29/11
An infrastructure for cancer virus discovery from next-generation sequencing data (Meyerson)
Role: Co-investigator

1RC1AG035681 (NIH/NIA) 09/30/09 – 09/29/11
Mutational Cloning in Familial Dementia and Alzheimer's Disease (Raskind)
Role: Co-investigator

1RC2CA148232 (NIH/NCI) 09/30/09 – 09/29/11
Application of RiboTag-seq to Exploration of Tumor Microenvironments (Morris)
Role: Co-investigator

5RC2HG005608 (NIH/NHGRI) 09/30/09 – 08/31/12
Next Generation Mendelian Genetics (MPI: Bamshad, Nickerson, Raskind, Shendure)
Role: PI (MPI award)

5UC2HL102926 (NIH/NHLBI) 09/30/09 – 06/30/12
Northwest Genomics Center (MPI: Green, Nickerson, Rieder, Shendure)
Role: PI (MPI award)

3U54AI057141-06S1880509 (NIH/NIAID) 09/12/09 – 02/29/12
Massively parallel genome sequencing of antibiotic-resistant emerging pathogens (Shendure)
Role: PI

2P50HG003233 (NIH/NHGRI) 05/01/09 – 04/30/14

Center for the Epigenetics of Common Human Disease (Feinberg)
Role: Co-investigator

5P01CA078902 (NIH/NCI) 02/01/09 – 01/31/14
Identification of Canine Minor Histocompatability Antigens (Storb)
Role: PI of Project 1

5R01HL094976 (NIH/NHLBI) 09/30/08 – 06/30/12
SeattleSeq (MPI: Eichler, Green, Nickerson, Shendure)
Role: PI (MPI award)

1R21HG004749 (NIH/NHGRI) 07/23/08 – 06/30/10
Molecular Tools for Genome Partitioning (Shendure)
Role: PI

Invited Talks or Workshops

Dec 2019	*Invited speaker & session moderator,* 4DN Annual Meeting, NIH Common Fund, National Institutes of Health (Washington DC)
Nov 2019	*Invited speaker,* 2019 UW Genome Sciences Symposium, “Genomics from Worm to Human – in honor of Bob Waterston” (Seattle, WA)
Sep 2019	*Invited speaker,* International Common Disease Alliance, Scientific Plenary & Launch (Potomac, MD)
Sep 2019	*Keynote seminar,* Inaugural Integrative Biology Symposium, Salk Institute (San Diego, CA)
Oct 2019	*Invited speaker,* 2019 Welch Conference on Chemical Research, "The Chemistry of Genome Editing and Imaging" (Houston, TX)
Oct 2019	*Invited speaker,* Allen Frontiers Symposium, Allen Institute (Seattle, WA)
Jun 2019	*Jeffrey M. Trent Lectureship in Cancer Research*, National Human Genome Research Institute, National Institutes of Health (Bethesda, MD)
May 2019	*Grand rounds,* Clinical Research Division, Fred Hutchinson Cancer Research Center (Seattle, WA)
Apr 2019	*Invited speaker,* High-Throughput Dense Reconstruction of Cell Lineages, Janelia Research Campus/HHMI (Ashburn, VA)
Mar 2019	*Paul D. Gottlieb Distinguished Lectureship*, Univerity of Texas, Austin (Austin, TX)
Feb 2019	*Student invited speaker,* Program in Genetics & Genomics, Department of Genetics, Harvard Medical School (Boston, MA)
Jan 2019	*Planning committee member & invited speaker*, “From Genome to Phenotype: Genomic Variation Identification, Association and Function in Human Health and Disease” meeting, National Human Genome Research Institute (Bethesda, MD)
Dec 2018	*Invited speaker,* TOPMed Steering Committee/External Advisory Panel meeting (Washington DC)
Nov 2018	*Allan C. Wilson Memorial Lectureship*, University of California, Berkeley (Berkeley, CA)
Oct 2018	*Invited speaker,* Allen Frontiers Symposium, Allen Institute (Seattle, WA)
Oct 2018	*Invited speaker,* American Journal for Human Genetics CoLab, 68th Annual Meeting of American Society of Human Genetics (San Diego, CA)
Oct 2018	*Keynote Speaker,* 2018 Institute for Genomic Medicine Symposium, University of California, San Diego (San Diego, CA)
Oct 2018	*Invited Speaker,* Single Cell Genomics 2018, Broad Institute (Cambridge, MA)

Sep 2018	*Inaugural Speaker,* Frontiers in Genome Biology and Medicine Seminar, Center for Genomic Medicine, Massachusetts General Hospital (Boston, MA)
Sep 2018	*Invited Speaker,* Clinical Sequencing Evidence-Generating Research (CSER) consortium, Annual Meeting (Seattle, WA)
Jul 2018	*Plenary Speaker*, Society for Developmental Biology (Portland, OR)
Apr 2018	*Invited seminar,* Broad Institute, Cell Circuits & Epigenomics Program Seminar Series (Cambridge, MA)
Apr 2018	*Invited seminar,* Department of Biological Engineering, Massachusetts Institute of Technology (Cambridge, MA)
Mar 2018	*Keynote speaker,* The Engineering Biology Research Consortium (EBRC) Spring Retreat (Seattle, WA)
Mar 2018	*Invited seminar,* Stanford University, Frontiers in Biology Seminar Series (Palo Alto, CA)
Mar 2018	*Invited seminar,* Department of Pharmacology, University of Washington (Seattle, WA)
Feb 2018	*Plenary speaker,* Advances in Genome Biology and Technology (AGBT) (Marco Island, FL)
Jan 2018	*Plenary speaker,* PAG XXVI - Plant & Animal Genome Conference (San Diego, CA)
Jan 2018	*Inaugural Richard and Carol Hertzberg Prize for Technology Innovation,* University of California, San Diego (San Diego, CA)
Jan 2018	*Inaugural Dr. Nancy C. Andrews Physician-Scientist Lectureship*, Duke Physician-Scientist Symposium (Durham, NC)
Nov 2017	*Invited speaker,* HHMI Investigators Meeting (Chevy Chase, MD)
Oct 2017	*Invited speaker,* Allen Frontiers Symposium (San Francisco, CA)
Oct 2017	*British Society of Genetic Medicine Lectureship*, BSGM Annual Conference, Royal College of Physicians (London, UK)
Sep 2017	*Invited speaker,* Allen Institute Bioscience and Philanthropy Summit (Seattle, WA)
Sep 2017	*Keynote speaker,* 4th Human Genetics in NYC Conference (New York City, NY)
Aug 2017	*Plenary speaker,* 69th American Association for Clinical Chemistry (AACC) Annual Scientific Meeting (San Diego, CA)
Jul 2017	*Invited speaker,* The CRISPR-Cas9 Revolution, Cold Spring Harbor Laboratory (Cold Spring Harbor, NY)
May 2017	*Invited speaker,* Biology of Genomes, Cold Spring Harbor Laboratory (Cold Spring Harbor, NY)
May 2017	*Invited speaker,* Seattle-area HHMI Alumni Dinner (Seattle, WA)
Apr 2017	*Co-organizer & speaker,* Genomics of Rare Diseases, Wellcome Genome Campus (Hinxton, UK)
Mar 2017	*Invited speaker,* Future of Genomic Medicine X, Scripps Translational Science Institute (San Diego, CA)
Feb 2017	*Invited speaker*, Human Cell Atlas meeting, Stanford University (Palo Alto, CA)
Jan 2017	*Invited speaker,* 2017 Keystone Symposia on Precision Genome Engineering (Breckenridge, CO)
Dec 2016	*Grand rounds,* Department of Medicine, University of Washington (Seattle, WA)
Dec 2016	*Invited speaker*, Cardiovascular Center Breakfast Club, University of Washington (Seattle, WA)
Dec 2016	*Invited speaker*, Cell Science Symposium, Allen Institute for Cell Science (Seattle, WA)
Dec 2016	*Invited seminar,* Department of Molecular & Cellular Biology, Harvard University (Cambridge, MA)
Oct 2016	*Invited speaker*, Allen Frontiers Symposium (New York City, New York)
Oct 2016	*Invited speaker*, Immune Profiling in Health and Disease 2016 (Seattle, WA)
Oct 2016	*Invited seminar,* New York Genome Center (New York City, New York)

Sep 2016 *Keynote speaker,* Division of Human Biology Annual Retreat, Fred Hutchinson Cancer Research Center (Seattle, WA)

Sep 2016 *Keynote speaker,* Department of Pathology Annual Retreat, University of Washington (Bainbridge Island, WA)

May 2016 *Keynote panel,* The Association for Research in Vision and Ophthalmology (ARVO), 2016 Annual Meeting (Seattle, WA)

Feb 2016 *Invited speaker*, "Epigenomics 2016", Cold Spring Harbor Laboratories (San Juan, PR)

Jan 2016 *Invited seminar,* Donnelly Centre, University of Toronto (Toronto, ON)

Jan 2016 *Invited seminar,* University of Michigan Genome Science Training Program (Ann Arbor, MI)

Dec 2015 *Invited seminar,* UCSF Biomedical Sciences Seminar Series (San Francisco, CA)

Dec 2015 *Invited seminar,* UCLA Clinical and Translational Science Institute (Los Angeles, CA)

Oct 2015 *Invited seminar,* Single-Cell Genomics Interest Group, National Institutes of Health (Bethesda, MD)

Oct 2015 *Invited session moderator & speaker,* 65th Annual Meeting of American Society of Human Genetics, "Multiplexed and Multimodal Experimental Dissection of Genetic Variants" (Baltimore, MD)

Sep 2015 *Keynote Speaker*, Biological and Biomedical Sciences (BBS) graduate program, Harvard Medical School (Provincetown, MA)

Jul 2015 *Keynote speaker,* The Human Genetics & Genomics Gordon Research Conference, Regina Salve University (Newport, RI)

Jul 2015 *Invited speaker*, "The Evolution of Sequencing Technology: A Half-Century of Progress", Cold Spring Harbor Laboratories (Cold Spring Harbor, NY)

May 2015 *Invited speaker*, Cardiovascular Center Breakfast Club, University of Washington (Seattle, WA)

Apr 2015 *Co-organizer & speaker,* Genomics of Rare Disease: Beyond the Exome, Wellcome Trust Scientific Conferences (Hinxton, UK)

Mar 2015 *Invited speaker,* Third Annual Frontiers in Genomics Lecture, Institute for Genome Sciences, University of Maryland (Baltimore, MD)

Mar 2015 *Workshop participant,* NHGRI: From Genome Function to Biomedical Insight: ENCODE and Beyond (Bethesda, MD)

Jan 2015 *Invited speaker,* Public Health Genomics Symposium, Department of Biostatistics, University of Washington (Seattle, WA)

Jan 2015 *Invited seminar*, Five Point Lecture Series, New York Genome Center (New York, NY)

Jan 2015 *Invited seminar*, Department of Systems Biology, Columbia University (New York, NY)

Nov 2014 *Invited speaker,* 2014 PQG Conference, "Integrative Approaches to Understand Allelic Function", Harvard School of Public Health (Boston, MA)

Nov 2014 *Invited seminar*, Program in Medical & Population Genetics, Broad Institute of M.I.T. and Harvard (Cambridge, MA)

Oct 2014 *Keynote speaker,* RECOMB/ISCB Conference on Regulatory and Systems Genomics (San Diego, CA)

Oct 2014 *Invited session moderator & speaker,* 64th Annual Meeting of American Society of Human Genetics, "Viruses, Genomic Instability, and the Pathogenesis of Human Cancers" (San Diego, CA)

Sep 2014 *Invited speaker,* Nobel Forum Minisymposium: "Renaissance on the Diagnosis of Monogenic Diseases" (Stockholm, Sweden)

Aug 2014 *Invited Speaker,* Systems Biology of Infectious Disease: Pathogenesis to Personalized Medicine (Seattle, WA)

Jul 2014	*Workshop participant & speaker,* Future Opportunities for Genome Sequencing and Beyond: A Planning Workshop for the National Human Genome Research Institute (Washington DC)
Jul 2014	*Invited seminar*, HudsonAlpha Institute for Biotechnology (Huntsville, AL)
May 2014	*Invited seminar*, Quantitative Biology Seminar Series, Cold Spring Harbor Laboratories (Cold Spring Harbor, NY)
May 2014	*Workshop participant & speaker,* NIH Workshop on Scientific and Ethical Issues Related to Open Access HeLa Genomic Data (Bethesda, MD)
Apr 2014	*Invited speaker,* Institute for Systems Biology Annual Symposium: Systems Biology & Cancer (Seattle, WA)
Apr 2014	*Keynote speaker,* Stanford University Annual Genomics and Personalized Medicine 2014 (Palo Alto, CA)
Mar 2014	*Invited speaker,* The Future of Genomic Medicine VII, Scripps Health (La Jolla, CA)
Jan 2014	*Keynote speaker,* UCLA Center for Neurobehavioral Genetics Annual Retreat (Los Angeles, CA)
Dec 2013	*Participant,* NIH/NCI Center for Cancer Genomics Think Tank (Bethesda, MD)
Nov 2013	*Speake*r, NIH/NCI Innovative Molecular Analysis Technologies (IMAT) Grantee Meeting (Bethesda, MD)
Oct 2013	*Invited speaker,* FederaDAG: Next Generation DNA Sequencing: impact on clinical care and society (Utrecht, Netherlands)
Oct 2013	*Invited seminar,* Nijmegen Centre for Molecular Life Sciences (Nijmegen, Netherlands)
Oct 2013	*Participant & speaker,* NIH/NHGRI Sequencing Network Meeting (Washington DC)
July 2013	*Invited seminar,* Fred Hutchinson Cancer Research Center, Computational Biology Seminar Series (Seattle, WA)
July 2013	*Invited speaker,* The Human Genetics & Genomics Gordon Research Conference, Bryant University (Smithfield, RI)
Jun 2013	*Keynote speaker,* Functional Genomics Data Society (FGED) 15th International Conference (Seattle, WA)
May 2013	*Invited seminar*, Department of Cellular and Molecular Medicine, University of California, San Diego (San Diego, CA)
May 2013	*Invited seminar*, McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine (Baltimore, MD)
Apr 2013	*Invited seminar*, Institute for Genomics & Systems Biology, University of Chicago (Chicago, IL)
Apr 2013	*Speake*r, NIH / NHGRI Advanced Sequencing Technology Grantee Meeting (San Diego, CA)
Mar 2013	*Invited seminar*, HudsonAlpha Institute for Biotechnology (Huntsville, AL)
Mar 2013	*Invited seminar*, Seminars in Integrative Genomics, Vanderbilt University (Nashville, TN)
Mar 2013	*Plenary speaker,* 2013 Annual Meeting of the Association of Biomolecular Resource Facilities (Palm Springs, CA)
Feb 2013	*Plenary speaker,* Advances in Genome Biology and Technology (AGBT) (Marco Island, FL)
Jan 2013	*Keynote speaker,* The Eleventh Asia Pacific Bioinformatics Conference (Vancouver, BC)
Dec 2012	*Invited seminar*, Dept. of Molecular and Medical Genetics, Oregon Health & Science University (Portland, OR)
Nov 2012	*Invited speaker*, CSHL Personal Genomes meeting (Cold Spring Harbor, NY)
Nov 2012	*Invited participant in closing symposium,* 62th Annual Meeting of American Society of Human Genetics, “Human Genetics 2012 and Beyond: Present Progress and Future Frontiers” (San Francisco, CA)

Nov 2012	*Invited session moderator & speaker,* 62th Annual Meeting of American Society of Human Genetics, "Genomic Approaches to Mendelian Disorders" (San Francisco, CA)
Nov 2012	*Curt Stern Award: Presentation and Lecture,* 62th Annual Meeting of American Society of Human Genetics (San Francisco, CA)
Nov 2012	*Invited speaker,* Institute of Translational Health Sciences 'Omics Workshop - "Lessons Learned and the Path Forward" University of Washington, South Lake Union (Seattle, WA)
Oct 2012	*Participant & speaker,* NHGRI Sequencing Network Meeting (Houston, TX)
Sep 2012	*Invited speaker,* Nature Genetics "Genomics of Common Disease" meeting (Washington DC)
Sep 2012	*Workshop co-organizer & attendee,*" Implicating Sequence Variants in Human Disease" (Washington DC)
Aug 2012	*Invited speaker,* 43rd Annual Meeting of the Environmental Mutagen Society (Seattle, WA)
Jul 2012	*Invited speaker,* 1000 Genomes Community Meeting (Ann Arbor, MI)
Jun 2012	*Invited seminar*, Department of Pathology, University of Washington (Seattle, WA)
Jun 2012	*Invited speaker,* ESHG European Human Genetics Conference 2012 (Nürnberg, Germany)
Jun 2012	*Invited seminar,* UCLA Molecular Biology Institute (Los Angeles, CA)
May 2012	*Grand rounds,* Division of Hematology, University of Washington (Seattle, WA)
May 2012	*Invited seminar,* Institute for Systems Biology (Seattle, WA)
Apr 2012	*Invited speaker,* Chemical & Engineering News Webinar
Apr 2012	*Invited seminar*, NIH / NHGRI Division of Intramural Research (Bethesda, MD)
Apr 2012	*Speake*r, NIH / NHGRI Advanced Sequencing Technology Grantee Meeting (San Diego, CA)
Mar 2012	*Distinguished Lecture Series,* Duke University Program in Genetics and Genomics (Chapel Hill, NC)
Mar 2012	*Co-organizer & speaker,* NIH / NIDDK "Workshop on Rare Syndromic Body Fat Disorders-What Can They Teach Us?" (Bethesda, MD)
Feb 2012	*Invited seminar*, Program in Medical & Population Genetics, Broad Institute of M.I.T. and Harvard (Cambridge, MA)
Feb 2012	*Invited seminar*, Division of Genetics, Brigham and Women's Hospital, Harvard Medical School (Boston, MA)
Jan 2012	*Invited seminar*, Cystic Fibrosis Seminar Series, Seattle Children's Research Institute / University of Washington (Seattle, WA)
Jan 2012	*Grand rounds,* Department of Pathology, Brigham and Women's Hospital, Harvard Medical School (Boston, MA)
Dec 2011	*Invited seminar*, Department of Biology, University of Pennsylvania (Philadelphia, PA)
Oct 2011	*Guest speaker,* Fred Hutchinson Cancer Research Center, 8th Human Biology Division Retreat (Seattle, WA)
Oct 2011	*Keynote address,* "The Genome and Beyond", BioTechniques Virtual Symposium
Oct 2011	*Chair & organizer,* IPAM (Institute for Pure & Applied Mathematics): Mathematical and Computational Approaches in High-Throughput Genomics; Workshop I: Next-generation Sequencing Technology and Algorithms for Primary Data Analysis (Los Angeles, CA)
Sep 2011	*Invited speaker & session chair,* Beyond the Genome 2011 (Rockville, MD)
Sep 2011	*Invited speaker,* NHLBI Symposium: Genomics: Gene Discovery and Clinical Applications for Cardiovascular, Lung, and Blood Diseases (Bethesda, MD)
Jul 2011	*Workshop speaker*, Illumina Sequencing Expert Panel 2011 (Woodinville, WA)
Jul 2011	*Invited speaker, "*Revolution of Genome Science", 9th International Workshop on Advanced Genomics (Tokyo, Japan)
Jul 2011	*Invited speaker,* University of Tokyo, "Cutting Edge of Human Genome Science", 4th

	Symposium of the IMSUT & RCAST Global COE (Tokyo, Japan)
Apr 2011	*Invited seminar,* Princeton University and Lewis-Sigler Institute, Quantitative and Computational Biology seminar series (Princeton, NJ)
Mar 2011	*Invited speaker,* Genome 10K Workshop (Santa Cruz, CA)
Feb 2011	*Invited seminar,* Stanford University, Frontiers in Biology Seminar Series (Palo Alto, CA)
Jan 2011	*Invited seminar,* Institute for Molecular Medicine, UT Houston (Houston, TX)
Dec 2010	*Invited speaker,* Illumina Webinar
Dec 2010	*Invited seminar,* UCSF Biomedical Sciences Seminar Series (San Francisco, CA)
Dec 2010	*Invited seminar,* Amgen, Molecular and Computational Toxicology Seminar Series (Seattle, WA)
Nov 2010	*Invited speaker*, American Heart Association, Scientific Sessions 2010, "Whole Genome Sequencing and Integrative Genomics" session (Chicago, IL)
Nov 2010	*Invited speaker*, American Heart Association, Scientific Sessions 2010, "Whole Exome Resequencing: Methods and Early Findings" session (Chicago, IL)
Nov 2010	*Invited session moderator & speaker,* 60th Annual Meeting of American Society of Human Genetics, "Exome Sequencing and Human Genetics" (Washington DC)
Oct 2010	*Invited seminar,* Department of Global Health, University of Washington, Pathobiology Seminar Series (Seattle, WA)
Oct 2010	*Invited speaker,* Beyond the Genome 2010 (Boston, MA)
Sep 2010	*Invited speaker,* Prostate Cancer Foundation, 17th Annual Scientific Retreat (Washington DC)
Jul 2010	*Invited speaker,* Illumina PNW User Group Meeting (Seattle, WA)
Jul 2010	*Invited speaker,* BioC 2010 (Seattle, WA)
Jul 2010	*Workshop participant,* Planning the Future of Genomics: Foundational Research and Applications in Genomic Medicine, NHGRI (Warrenton, VA)
Jul 2010	*Invited speaker,* 13th International MGED Meeting (Boston, MA)
Jul 2010	*Invited speaker,* Merck (Boston, MA)
Jul 2010	*Evening lecture,* 51st Annual Short Course on Medical and Experimental Mammalian Genetics, The Jackson Laboratory (Bar Harbor, ME)
Jun 2010	*Invited seminar,* PNW Prostate Cancer SPORE Seminar Series (Seattle, WA)
May 2010	*Colloquium co-convener & speaker*, American Society for Microbiology 110th General Meeting, "Ultra-Deep Sequencing in Infectious Diseases" (San Diego, CA)
May 2010	*Invited speaker,* University of Washington, Computational Molecular Biology Spring Symposium (Seattle, WA)
May 2010	*Invited seminar,* University of Washington, Department of Medical Genetics Seminar Series (Seattle, WA)
May 2010	*Session co-chair & speaker,* The Biology of Genomes, Cold Spring Harbor Laboratories, "High Throughput Genomics & Genetics" (Cold Spring Harbor, NY)
May 2010	*Workshop participant,* NIH Director's "Big Think" Meeting (Bethesda, MD)
Apr 2010	*Invited speaker,* 4th International Conference on Primate Genomics (Seattle, WA)
Jan 2010	*Invited seminar,* Washington University in St. Louis, Department of Genetics (St. Louis, MO)
Jan 2010	*Invited seminar,* University of Chicago, Department of Human Genetics (Chicago, IL)
Dec 2009	*Invited speaker*, Simons Foundation, workshop on sequencing (New York City, NY)
Dec 2009	*Invited speaker*, Cardiovascular Center Breakfast Club, University of Washington (Seattle, WA)
Oct 2009	*Plenary speaker,* 59th Annual Meeting of American Society of Human Genetics (Honolulu, HI)
Sep 2009	*Grand rounds*, Department of Laboratory Medicine, University of Washington (Seattle, WA)
Sep 2009	*Invited speaker*, CSHL Personal Genomes meeting (Cold Spring Harbor, NY)

Aug 2009 *Invited speaker*, eMERGE Network Steering Committee meeting (Seattle, WA)
Aug 2009 *Invited seminar*, McDermott Center, Excellence in Human Genetics Lecture Series, UT Southwestern (Dallas, TX)
Jun 2009 *Invited speaker*, Genomic Tools and Technologies Summit, Cambridge Healthtech Institute (San Francisco, CA)
May 2009 *Invited speaker,* Northwest Institute of Genetic Medicine, 2009 Retreat (Seattle, WA)
Mar 2009 *Invited seminar*, University of Michigan, Center for Translational Pathology (Ann Arbor, MI)
Mar 2009 *Invited speaker,* Next-Generation Sequencing meeting, Cambridge Healthtech Institute (San Diego, CA)
Feb 2009 *Invited speaker,* Advances in Genome Biology and Technology (AGBT) (Marco Island, FL)
Feb 2009 *Invited speaker,* Advances in Genome Biology and Technology (AGBT), pre-meeting workshop (Marco Island, FL)
Dec 2008 *Invited seminar*, Puget Sound Blood Center Research (Seattle, WA)
Oct 2008 *Invited speaker,* Discovery2Diagnostics conference (San Diego, CA)
Sep 2008 *New Investigator Science in Medicine Lecture*, University of Washington (Seattle, WA)
Sep 2008 *Keynote address,* Institute for Systems Biology, Annual Retreat (Seabeck, WA)
Sep 2008 *Invited speaker,* Nature Genetics "Genomics of Common Disease" meeting (Cambridge, MA)
Aug 2008 *Invited seminar,* BC Cancer Agency, Genome Sciences Centre (Vancouver, BC)
Mar 2008 *Invited seminar,* Fred Hutchinson Cancer Research Center, Computational Biology Working Group Seminar Series (Seattle, WA)
Mar 2008 *Invited seminar,* University of Washington, Department of Medical Genetics Seminar Series (Seattle, WA)
Mar 2008 *Invited speaker,* Joint Genome Institute (JGI) User 3rd Annual Meeting (Walnut Creek, CA)
Feb 2008 *Invited speaker,* Association of Biomolecular Resource Facilities (ABRF) Annual Meeting (Salt Lake City, UT)
Feb 2008 *Plenary speaker,* Advances in Genome Biology and Technology (AGBT) (Marco Island, FL)
Nov 2007 *Invited seminar,* Stanford University, Frontiers in Biology Seminar Series (Palo Alto, CA)
Nov 2007 *Invited speaker,* 1st Annual Parallel Sequencing Genomics Meeting, Stanford Genome Technology Center, Stanford University (Palo Alto, CA)
Sep 2007 *Invited seminar,* Fred Hutchinson Cancer Research Center, Program in Prostate Cancer Research Seminar Series, (Seattle, WA)
May 2007 *Invited speaker,* Stanford Genome Technology Center, Stanford University (Palo Alto, CA)
Mar 2007 *Invited seminar*, Institute for Molecular Pediatric Sciences, University of Chicago (Chicago, IL)
Mar 2007 *Invited speaker,* Next Generation Sequencing: Applications and Case Studies, Cambridge Healthtech Institute (San Diego, CA)
Feb 2007 *Invited seminar*, Department of Genetics, University of Pennsylvania (Philadelphia, PA)
Feb 2007 *Invited seminar*, Department of Bioengineering, University of California, Berkeley (Berkeley, CA)
Feb 2007 *Invited seminar*, Division of Genetics, Brigham and Women's Hospital, Harvard Medical School (Boston, MA)
Feb 2007 *Invited seminar*, Department of Pathology, Massachusetts General Hospital, Harvard Medical School (Boston, MA)
Feb 2007 *Invited seminar*, Department of Genome Sciences, University of Washington (Seattle, WA)
Feb 2007 *Invited seminar*, Broad Institute of M.I.T. and Harvard (Cambridge, MA)
Jan 2007 *Invited seminar*, Department of Molecular & Cell Biology, University of California, Berkeley (Berkeley, CA)
Jan 2007 *Invited seminar*, National Human Genome Research Institute, National Institutes of Health

	(Bethesda, MD)
Jan 2007	*Workshop speaker,* Workshop on Systems Biology and Information Medicine in a Global Society, Princeton University (Princeton, NJ)
Jan 2007	*Invited seminar*, Institute for Systems Biology (Seattle, WA)
Mar 2006	*Invited seminar*, Biological Physics & Biophysical Chemistry Seminar, State University of New York, Stony Brook (Stony Brook, NY)