

CURRICULUM VITAE

Evan Eugene Eichler

Professor

Howard Hughes Medical Institute

University of Washington

Genome Sciences, Box 355065

Seattle, WA 98195-5065

(206) 543-9526

Date of birth: October 6, 1968, Cheyenne, WY
Personal status: Married, four children
WWW site: <http://www.gs.washington.edu/faculty/eichler.htm>
<http://eichlerlab.gs.washington.edu/>

EDUCATION

1995 Ph.D. Department of Human Molecular Genetics, Baylor College of Medicine, Houston, TX
Thesis (David L. Nelson, Supervisor): *AGG Interspersions within the FMR1 CGG Repeat: Models and Mechanisms of Triplet Repeat Instability*

1991 – Research Scholar, Deutscher Akademischer Austauschdienst
Ludwig-Maximilians Universität, Munich, Germany

1990 B.S. Department of Biology, University of Saskatchewan, Saskatoon, Canada
Honours Program in Biology

PROFESSIONAL EXPERIENCE

2008–Present Professor (with tenure)
Department of Genome Sciences, University of Washington (UW), Seattle, WA

2005–Present Howard Hughes Medical Institute Investigator (HHMI)

2015–Present Associate Member, New York Genome Center (NYGC), New York City, NY

2004–Present Affiliate Professor
Division of Human Biology, Fred Hutchinson Cancer Research Center, Seattle, WA

2004–2008 Associate Professor (with tenure)
Department of Genome Sciences, UW, Seattle, WA

2003–2004 Associate Professor (with tenure)
Department of Genetics, Case Western Reserve University (CWRU), Cleveland, OH

2003–2004 Appointed Faculty Member
Cancer Center, Division of Medical Sciences, CWRU, Cleveland, OH

2002–2004 Director of Bioinformatics Core Facility
Department of Genetics, CWRU, Cleveland, OH

1999–2004 Appointed Faculty Member
University Hospitals of Cleveland, Cleveland, OH

1997–2003 Assistant Professor
Department of Genetics, CWRU, Cleveland, OH

1997 Research Affiliate
Department of Human Genetics, Roswell Park Cancer Institute, Buffalo, NY

1995–1997 Postdoctoral Fellow, Biology and Biotechnology Research Program
Lawrence Livermore National Laboratory, Livermore, CA (Harvey Mohrenweiser, Supervisor)

HONORS AND AWARDS

2014–2016 Honorary Professor, Kunming University of Science and Technology (KUST), Kunming, China

2013 Allen Distinguished Investigator (ADI), The Paul G. Allen Foundation

2013 National Academy of Sciences (NAS; Elected 2012)

2012 Washington State Academy of Sciences (WSAS)

2012 Mendel Lecture (“Gilded Pea” Award): European Society of Human Genetics, Nuremberg, Germany

2010 AAAS (American Association for the Advancement of Science) Newcomb Cleveland Prize

2009 Distinguished Alumnus Award: Baylor College of Medicine, Graduate School

2008 Curt Stern Award: American Society of Human Genetics (ASHG), Philadelphia, PA

2006 AAAS Fellow

2005–Present Howard Hughes Medical Institute Investigator

1998–2001 Basil O’Connor Young Investigator Award: March of Dimes Birth Defects Foundation

1995–1997	Distinguished Human Genome Postdoctoral Fellowship: Department of Energy Genome Hollaender Fellowship
1994	Predoctoral Basic Research Award: American Society of Human Genetics
1993–1995	National Research Service Award/Human Genome Research: National Institutes of Health (NIH)
1990–1991	Research Scientist Award: Deutscher Akademischer Austauschdienst
1987	Canadian Summer Research Award: National Science and Engineering Research Council of Canada

ACADEMIC SERVICE

a) Editorial

2009–Present	Editorial Board of <i>Molecular Autism</i>
2004–2012	Academic Editor, <i>Public Library of Science (PLOS)</i>
2003	Section Editor, <i>Curr Opin Genet Dev</i> , Genomes and Evolution Special Issue
2002–Present	Editor of <i>Genome Research</i>
2002–2004	Editorial Board of <i>American Journal of Human Genetics</i>
2002–2007	Editorial Board of <i>DNA Sequence</i>
2002–2009	Editorial Board of <i>BMC Genomics</i>
1999–2002	Editorial Board of <i>Genome Research</i>

b) Scientific Advisory Boards (SABs)

2013–2015	New York Genome Center (NYGC) SAB
2012–Present	DNAnexus, Inc. SAB
2011–2013	SynapDx Corp. SAB
2009–2013	Pacific Biosciences of California, Inc. SAB
2009–2012	Simons Foundation Autism Research Initiative (SFARI) Structural Variation Project (SSVP) SAB
2008–2012	Yerkes National Primate Center SAB
2008–2012	International Cancer Genome Consortium SAB, Ontario Institute of Cancer Research
2005	Member, SAB (<i>ad hoc</i>), Genome Center North Carolina, UNC Chapel Hill
2004	Member, SAB (<i>ad hoc</i>), Regulatory Genetics and GRAND Project, Genome Center, McGill University, Montreal (Director: Tom Hudson)
2003	Member, SAB (<i>ad hoc</i>), Department of Evolutionary Genetics, Max Planck Institute for Molecular Anthropology, Leipzig (Director: Svante Pääbo)

c) International

2015–Present	Member, International Advisory Board, International Laboratory of Human Genome Research (LIIGH), National University of Mexico (UNAM), Queretaro, Mexico
2014–2016	Chinese 1000 Talents Program, Kunming University of Science and Technology, China
2012–2014	Board of Directors, American Society of Human Genetics (ASHG)
2012	Organizer, Personal Genomes and Medical Genomics Meeting, Cold Spring Harbor Laboratory, New York
2011	Reviewer, Molecular Cytogenetics, Wellcome Trust Centre for Human Genetics, Oxford
2011	Organizer, Keystone Symposium: Functional Consequences of Genome Structural Variation
2011–2014	Steering Committee, 1000 Genomes Project (1KG)
2009	Chair, Gordon Research Conference (GRC): Human Genetics & Genomics
2009	Chair, ASHG Nominating Committee
2009	Co-organizer, Banbury Conference, “Functional Consequences of Structural Variation”
2009–2014	Member, International Cytogenomic Standard Array (ISCA) Steering Committee
2008–Present	Co-Chair, 1000 Genomes Project Structural Variation Working Group
2007	Vice-Chair, Gordon Research Conference (GRC): Human Genetics & Genomics
2005–2007	Member, ASHG Awards Committee
2004–2005	Genome Study Section (GMX), Canadian Institutes of Health, permanent roster
2004–2005	Organizer, Symposium: Understanding Human Genome Evolution, Bertinoro, Italy
2002–2005	Member, HUGO (Human Genome Organization) Organizing Committee
2001–2004	Member, HUGO (Human Genome Organization) Annotation Committee
2001	Member, Human Genome Project, Sequence Analysis Group, International Human Sequencing Consortium
2001–2003	Member of Faculty of 1000, Genomics
2000	Workshop Organizer, ASHG, “Primate Origins and Evolution”
1999, 2002	Reviewer, Wellcome Trust, Genome Grants
1999, 2001	Reviewer, German Human Genome Project grant proposals

d) National Advisory

2015	Member, External Advisory Committee, MIND Institute IDDRC, UC Davis
2015	Chair, External Advisory Committee, Department of Human Genetics, University of Michigan

2014–Present NIH/NHGRI Study Section, Genome Research Review Committee, GNOM-G; term 2014–2018
 2014–2015 Member, IMFAR Program Committee
 2014 Reviewer, Paul G. Allen Foundation Grant
 2012 NCAB Working Group for the NCI Center for Cancer Genomics
 2012 NIH Workshop, Establishing a Central Resource of Data from Genome Sequencing Projects
 2012 Simons Foundation, SFARI 16p11.2 Workshop
 2011–2016 Autism Sequencing Consortium (ASC)
 2011 Reviewer, Department of Preventive Medicine, Keck School of Medicine, University of Southern California
 2010, 2011 Simons Foundation, Autism Next-generation Genome Sequencing Meeting
 2009 NIH Study Section, NIMH ARRA Stimulus GO Application Review Committee
 2009 NIH Study Section, NIMH P30 Study Section (*ad hoc*)
 2006–2007 CNS Foundation Young Scientist Faculty Advisory Committee
 2006 National Human Genome Research Institute (NHGRI), Genomic Structural Variation Steering Committee
 2005 NHGRI, External Advisory Board for Stanford University CEGS
 2005–2011 NHGRI, Medical Sequencing Working Group (MSWG) Member
 2004–2007 NIH Study Section, GCAT (formerly Genome), permanent roster
 2003–2004 NIH Study Section, Genome, permanent roster
 2003–2010 NHGRI, Annotating the Human Genome (AHG) Working Group, to identify species for large-scale whole-genome sequencing
 2003 FASEB Advisory Committee for FY2005 Federal Appropriations, DOE subcommittee
 2003–2004 NIH Study Section, Mammalian Genetics, *ad hoc* reviewer
 2003–2004 NSF Study Section, Hominid Review panel, Molecular Anthropology
 2002–2008 Member, BAC Resource Steering Panel (BRSP) Committee, NHGRI
 2001 Reviewer, Biotechnology Study Section, NIDDK
 2000–2001 NIH Study Section, Genome, *ad hoc* reviewer Camilla Day (SRA)
 2000 Reviewer, Board of Regents Millennium Trust Louisiana Health Excellence Fund Proposals (State)
 2000 Consultant, NIH trace data repository. Served as consultant on the creation of an archive for sequence trace data from the Human Genome Project
 1999, 2002 External Reviewer, Molecular Anthropology, NSF
 1999 Participant, NIH Summer Sequencing Project, part of group to assess utility of *Fugu rubripes* low-pass sequencing as a tool for human genome annotation
 1998 Participant, NIH Meeting "Summer Sequencing Experiment." Prepared NIH user report to assess usefulness of low-pass sequencing (minimal sequencing) as a new strategy for human genomic sequencing

e) University

2014–Present Co-PI, Interdisciplinary Training in Genome Sciences (Genome Training Grant: GTG; 2 T32 HG000035-21)
 2014–Present UW Medical School Training Program (MSTP) Admissions Committee
 2012–2013 Chair, Department of Genome Sciences Seminar Organizing Committee
 2011–Present Member, Genome Sciences Teaching Curriculum Committee
 2010–2011 Chair, Genome Sciences Faculty Search Committee
 2009 Member, Genome Sciences 371 Course Planning Committee
 2008–2009 Member, Department of Genome Sciences Seminar Organizing Committee
 2006–2008 Member, CFAR Genomics Core Steering Committee, UW
 2005–2008 Member, Human Variation and Medicine, Genome Sciences, UW
 2005–Present Member, Genome Training Grant Advisory Committee, UW
 2005–2008 Member, Genome Sciences Seminar Series Committee, UW
 2004–2007 Member, Genomics Faculty Search Committee, Genome Sciences, UW
 2004–2005 Member, UW Data Center Task Force, UW
 2004–2005 Organizer, Department of Genome Sciences Fourth Annual Symposium: Comparative Genome Analysis, UW
 2003–2004 Member, University Interdisciplinary Strategic Planning Committee, CWRU
 2003–2004 Member, Steering Committee, R25 Training in Computational Genomics and Epidemiology of Cancer
 2003–2004 Member, Committee Appointments, promotions and tenure, Department of Genetics
 2001–2003 Member, Chairman Search Committee for Department of Genetics
 2001–2004 Director of Bioinformatics Core Facility, Department of Genetics. Construction of LINUX high-capacity, multi-processor PC cluster farm, development of a graduate student computational laboratory to be used in conjunction with course offering (Gene 508, Spring 2001) and supervision of departmental systems administrator, programmer and database manager
 2000–2001 Chair, Bioinformatics Faculty Search Committee, Department of Genetics
 2000–2004 Executive Committee Member, Center for Computational Genomics. Joint collaboration between School of Medicine and School of Electrical Engineering and Computational Sciences

2000	CWRU “Bioinformatics/Genomics Technologies” Panel
1999	Graduate Student Poster Presentation Judge, BSTP Student Symposium
1999	Bioinformatics presentation on behalf of School of Medicine to Dr. Yutaka Kuwahara (Senior Corporate Executive, Leader of R & D Global Operation, Research and Development Group, Hitachi) for the purpose of establishing tera-flop supercomputing capacity at CWRU
1999	Departmental Bioinformatics Core Facility design
1998	Medical School Training Program (MSTP) NIH Site Visit
1998	Keck Foundation Equipment Grant

f) Membership Affiliations

2013–Present	National Academy of Sciences (NAS)
1997–Present	American Society of Human Genetics (ASHG)
1997–Present	American Association for the Advancement of Science (AAAS)

REVIEWER

Nature	Human Molecular Genetics
Science	Genetic Epidemiology
Cell	Chromosoma
New England Journal of Medicine	Journal of Medical Genetics
Nature Genetics	PLOS Computational Biology
Nature Biotechnology	American Journal of Psychiatry
Nature Medicine	Molecular Endocrinology
Genome Research	Genes Chromosomes and Cancer
Nature Review Genetics	BMC Genomics
Genes and Development	European Journal of Human Genetics
Nature Methods	Genomics
Neuron	Mammalian Genome
Trends in Genetics	Neurogenetics
American Journal of Human Genetics	Human Genetics
EMBO Journal	Journal of Molecular Evolution
Current Opinion Genetics and Development	Gene
PLOS Genetics	Molecular Phylogenetics and Evolution
Proceedings of the National Academy of Sciences	Mutation Research
Genome Biology	Molecular Autism
PLOS Biology	Cytogenetics and Cell Genetics
Science Translational Medicine	Journal of Molecular Genetics
Nucleic Acids Research	Somatic Cell and Molecular Genetics

TEACHING EXPERIENCE

2016–Current	GENOME 372 “Genomics and Proteomics” Lecturer (13 contact hours/5 weeks) Department of Genome Sciences, University of Washington (UW)
2009–Current	GENOME 465/565 “Advanced Human Genetics” Lecturer (13 contact hours/5 weeks) Department of Genome Sciences, University of Washington (UW)
2009–2014	GENOME 351 “Human Genetics - The Individual and Society” Lecturer (13 contact hours/5 weeks) Department of Genome Sciences, UW
2008	GENOME 371 “Introductory Genetics” Lecturer (25 contact hours/10 weeks) GENOME 465 “Advanced Human Genetics” Lecturer (13 contact hours/5 weeks) Department of Genome Sciences, UW
2007	GENOME 371 “Introductory Genetics” Faculty Shadow (50 contact hrs/10 weeks)

Department of Genome Sciences, UW

- 2006–2007 GENOME 465/565 “Advanced Human Genetics”
Lecturer: Genome Structure, Disease, Diversity and Evolution—a 10-week course co-taught with Mary-Claire King (13 contact hrs/5 weeks)
Department of Genome Sciences, UW
- 2006 PATHOLOGY 530 “Cytogenetics”
Lecturer: Recurrent Microdeletion and Microduplication Syndromes (1 contact hr)
Department of Genome Sciences, UW
- 2005–2006 GENOME 580 “Ethics in Biomedical Research”
Lecturer: Handling Data (1 contact hr)
Department of Genome Sciences, UW
- 2004 GENOME 511 “Genomics”
Lecturer: Genome Technology and Array Comparative Genomic Hybridization
Department of Genome Sciences, UW (2 contact hrs/year)
- 1997–2004 GENE 500/504 “Advanced Eukaryotic Genetics”
Lecturer and Section Leader of course module: Population, Quantitative and Evolutionary Genetics. Topics: Physical Mapping, Genome Organization, Human Molecular Evolution and Repeat Structure Introductory course for all 2nd year Genetics graduate students
Department of Genetics, Case Western Reserve University (CWRU) (8 contact hrs/year)
- 2000–2004 GENE 511 “Critical Analysis of Scientific Literature”
Discussion Leader
Department of Genetics, CWRU (2 contact hrs/year)
- 1998–2004 MED school Core Academic Program, Genetics core small group sessions
Discussion Leader: Mendelian Inheritance, Linkage, Cytogenetics, Triplet Repeat Diseases, Cancer Genetics
Genetics core small group sessions for medical students (4 contact hrs/year)
- 2001, 2003 GENE 508 “Bioinformatics and Computational Biology”
Course Organizer and Lecturer. Course designed to provide an understanding of the theory and application of computational methods for molecular biology research.
Twenty-two lectures covering DNA sequence, computational genomics, protein, gene expression and phylogenetic analysis. For every hour of lecture, there are 2-3 hours of problem solving exercises within the computational laboratory.
Advanced course for upper year Genetics graduate students.
Department of Genetics, CWRU (62 contact hrs/year)
- 2000–2001 GENE 458 “Introduction to Computational Biology”
Lecturer: Computational Genomics
Introductory course offering crossover training between Genetics and EECS
Department of Genetics, CWRU (2 contact hrs/year)
- 1999–2002 CBIO 453 “Correlated Curriculum in Cell and Molecular Biology” (C3MB)
Lecturer: Bioinformatics, Physical Mapping, Genomics
Introductory course for all incoming BSTP graduate students
Basic Science Training Research Program, CWRU (4 contact hrs/year)
- 1998, 2000 GENE 510 “Advanced Human Genetics”
Lecturer: Non-Mendelian Inheritance, Triplet Repeat Instability and Disease, Proteomic and Genomic Approaches, Single-Nucleotide Polymorphism and Phenotype Association
Advanced course for upper year Genetics graduate students
Department of Genetics, CWRU (6 contact hrs/year)

RESEARCH TRAINING**a) Doctoral Students**

- 2014–Present Madeleine Geisheker, MSTP, UW, doctoral candidate, advanced to candidacy September 2016.
- 2014–Present Max Dougherty, MSTP, UW, doctoral candidate, advanced to candidacy December 2016.
- 2011–2016 Michael Duyzend, MSTP, UW, advanced to candidacy August 2013, graduated June 2016. Thesis: Understanding the genetic basis of phenotype variability in individuals with neurocognitive disorders. Current: UW Medical School, Seattle
- 2011–2015 Xander Nuttle, UW, advanced to candidacy June 2012, graduated Nov 2015. Thesis: Human-specific duplicate genes: new frontiers for disease and evolution. Current: Postdoctoral Fellow, Massachusetts General Hospital & Harvard Medical School with Michael Talkowski, Boston
- 2010–2014 Nik Krumm, MSTP, UW, advanced to candidacy July 2012, graduated June 2014. Thesis: Discovery and convergence of inherited mutations in autism spectrum disorder. Current: UW Medical School, Seattle
- 2009–2013 Peter Sudmant, UW, advanced to candidacy August 2010, graduated September 2013. Thesis: Evolution and diversity of hominid genomes. Current: Postdoctoral Research Fellow, MIT with Chris Burge, Cambridge
- 2007–2011 Andrew Itsara, MSTP, UW, advanced to candidacy May 2009, graduated May 2011. Thesis: Detection and characterization of human copy-number variation. Current: Resident Physician, Internal Medicine Specialization, UW School of Medicine, Seattle
- 2006–2010 Jeffrey Kidd, UW, advanced to candidacy June 2007, graduated January 2010. Thesis: Mapping and sequencing human genomic structural variation. Current: Assistant Professor (tenure-track), Department of Human Genetics & Department of Computational Medicine and Biology, University of Michigan, Ann Arbor
- 2004–2008 Zhaoshi Jiang, UW, advanced to candidacy June 2005, graduated November 2008. Thesis: Evolutionary reconstruction of primate segmental duplications. Past: Research Scientist, Genentech, Inc.; Current: Associate Director, Bioinformatics, Gilead Sciences, San Francisco
- 2000–2007 Matthew E. Johnson, advanced to candidacy December 2001 (Genetics), graduated August 2007. Thesis: Low-copy repeat regions on chromosome 16 and rapid gene evolution. Current: Senior Research Associate, Children's Hospital of Philadelphia
- 2000–2004 Devin Locke, advanced to candidacy November 1998 (Genetics), joined laboratory April 2000 from Nicholls laboratory, graduated June 2004. Thesis: 15q11-q13 genomic instability. Past: Research Associate, Genome Center, Washington University School of Medicine & Lead Interpretation Scientist, Knome Inc.; Current: Director of Research and Development – Assembly, Seven Bridges Genomics, Cambridge
- 1999–2002 Jeffrey Bailey, advanced to candidacy December 1997 (Genetics), joined laboratory December 1999 from Chakravarti lab, graduated April 2002. Thesis: Genome-wide analysis and detection of segmental duplications. Current: Assistant Professor of Medicine and Physician (Transfusion Medicine), University of Massachusetts Medical School, Worcester
- 1998–2003 Juliann Horvath-Roth, advanced to candidacy November 1998 (Genetics), graduated November 2003. Thesis: Origin and mechanism of pericentromeric duplications. Current: Director, Genomics & Microbiology Research Laboratory, North Carolina Museum of Natural Sciences & Research Associate Professor, Biology, North Carolina Central University, Durham

b) Postdoctoral Fellows/Research Associates

- 2017–Present Hui Guo, Ph.D., postdoctoral research: Genetics of Autism.
- 2017–Present Davide Risso, Ph.D., postdoctoral research: Characterization of the function of *Homo sapiens*-specific gene families.
- 2016–Present PingHsun Hsieh, Ph.D., postdoctoral research: Paralogous copy number variation and disease association.
- 2015–Present Jason Underwood, Ph.D., postdoctoral research: Long-read transcript sequencing.

- 2015–2016 Chris Hill, Ph.D., postdoctoral research: Sequence and assembly of complex genomes using SMRT sequencing. Current: Software developer, Google Campus, Kirkland, WA
- 2015–Present Zev Kronenberg, Ph.D., postdoctoral research: Disease association and positive selection of structural variation.
- 2014–Present Stuart Cantsilieris, Ph.D., postdoctoral research: Structural diversity of duplicated immune response genes and disease association.
- 2014–Present Tychele Turner, Ph.D., postdoctoral research: Characterization of autism genetic risk factors.
- 2013–2015 Bo Xiong, Ph.D., postdoctoral research: Discovery and modeling of autism mutations. Current: Assistant Professor, Tongji Medical College of Huazhong University of Science and Technology at Wuhan, China
- 2013–2016 Holly Stessman, Ph.D., postdoctoral research: Intersection of genetic drivers in cancer and autism spectrum disorder. Current: Assistant Professor, Department of Pharmacology, Creighton University School of Medicine, Omaha, NE
- 2012–Present Mark Chaisson, Ph.D., postdoctoral research: *De novo* assembly of next-generation sequencing data and structural variation detection.
- 2012–2017 Osnat Penn, Ph.D., postdoctoral research: Gene expression analysis of recently duplicated genes. Current: Scientist II, Modeling, Analysis and Theory group, Allen Institute for Brain Science, Seattle, WA
- 2012–2013 Stuart Davidson, Ph.D., postdoctoral research: Investigations into the genetic basis of autism and Asperger phenotypes. (deceased)
- 2011–2015 Fereydoun Hormozdiari, Ph.D., postdoctoral research: Algorithm development for discovery and characterization of genome structural variation. Current: Assistant Professor, Biochemistry and Molecular Medicine; M.I.N.D. Institute, UC Davis Genome Center
- 2010–2015 Megan Dennis, Ph.D., National Research Service Award (NRSA) / K99/R00 Postdoctoral Fellow: Genetic and functional analysis of copy number variants associated with neurocognitive disease. Current: Assistant Professor, Department of Biochemistry and Molecular Medicine, University of California, Davis
- 2010–Present Bradley Coe, Ph.D., Canadian Institutes of Health Research (CIHR) Fellow: Development of a morbidity map for copy number variation in neurocognitive disorders. Current: Acting Instructor, Department of Genome Sciences, University of Washington
- 2010–2012 Beth Dumont, Ph.D., Genome Training Grant Fellow: Characterization of gene conversion within segmental duplications. Current: Assistant Professor, The Jackson Laboratory, Bar Harbor
- 2009–2012 Karyn Meltz Steinberg, Ph.D., National Research Service Award (NRSA) Fellow: Exploring regions of extreme diversity in the human genome. Current: Staff Scientist, The Genome Institute at Washington University, St. Louis
- 2009–2013 Emre Karakoc, Ph.D., postdoctoral research: Computational methods for characterization of genome and exome structural variation. Current: Assistant Professor, Department of Computer Engineering, Abdullah Gül University, Kayseri, Turkey
- 2009–2013 Brian O'Roak, Ph.D., postdoctoral research: Next-generation sequencing approaches to gene discovery in autism spectrum disorders. Current: Assistant Professor, Department of Molecular & Medical Genetics, Oregon Health & Sciences University, Portland
- 2008–2013 Catarina (Katie) Campbell, Ph.D., National Research Service Award (NRSA) fellow: High-throughput genotyping of structural variants. Current: Investigator II in Next Generation Diagnostics Group, Novartis Institutes for Biomedical Research, Boston
- 2008–2012 Santhosh Girirajan, Ph.D., postdoctoral research: Mechanisms and implications of large-scale genome rearrangements. Current: Assistant Professor (tenure-track), Department of Biochemistry and Molecular Biology & Department of Anthropology, Pennsylvania (Penn) State University, University Park

- 2007–2012 Francesca Antonacci, Ph.D., postdoctoral research: Discovery and characterization of chromosomal inversions as common variants in the human genome. Current: Assistant Professor, Department of Biology, University of Bari, Italy
- 2007–2011 Jeremiah J. Smith, Ph.D. (jointly supervised w/ Dr. Chris T. Amemiya), postdoctoral research: Developmentally programmed rearrangement of the lamprey genome. Current: Assistant Professor, University of Kentucky, Lexington
- 2007–2010 Tomas Marques-Bonet, Ph.D., Marie Curie Fellow: Evolution of human/great-ape segmental duplications. Current: Associate Professor & ICREA Researcher, Institut de Biologia Evolutiva, Universitat Pompeu Fabra, Barcelona, Spain
- 2007–2010 Gregory Cooper, Ph.D., Jane-Coffin Childs Fellow: High-throughput detection and genotyping of human copy number variation (Co-mentored w/ Debbie Nickerson). Current: Faculty Investigator, HudsonAlpha Institute for Biotechnology, Huntsville, & Adjunct Faculty, Department of Genetics, University of Alabama at Birmingham
- 2006–2009 Cemali Bekpen, Ph.D., HHMI Fellow: Functional characterization of Morpheus gene family. Current: Postdoc, Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Biology, Plön, Germany
- 2006–2008 Heather Mefford, M.D., Ph.D., Burroughs-Wellcome Scientist and Medical Genetics Fellow: Duplication-mediated rearrangement within fetal demise. Current: Associate Professor & Attending Physician, Department of Pediatrics, UW School of Medicine & Seattle Children's Hospital, Seattle
- 2005–2011 Can Alkan, Ph.D., HHMI Fellow: Development of mapping algorithms for next-generation sequence data. Current: Assistant Professor (tenure-track), Department of Computer Engineering, Bilkent University, Ankara, Turkey
- 2005–2007 Tera Newman-Eerkes, Ph.D., postdoctoral research: Structural variation and linkage disequilibrium within the human population. Past: CEO & Founder, iGenix, Inc.; Current: Business Owner/VP, Amplicon Consulting LLC & Director of R&D, Iverson Genetics, Seattle
- 2003–2007 Andrew Sharp, Ph.D., Rosetta Postdoctoral Fellow: Detection of segmental aneusomy in duplicated DNA. Current: Associate Professor/Senior Faculty, Genetics and Genomic Sciences, Mt. Sinai School of Medicine, New York City
- 2002–2006 Xinwei She, Ph.D., Rosetta Postdoctoral Fellow: Computational analysis of segmental duplications. Current: Senior Computational Scientist, Merck, Boston
- 2002–2004 Audrey Lynn, Ph.D. (jointly supervised with Dr. Terry Hassold), postdoctoral research: Genetic and physical correlation of recombination. Current: Project Coordinator, Department of Family Medicine, CWRU, Cleveland
- 2001–2002 Vicky Choi, Ph.D., PMMB Fellow: Computational methods for sequence assembly of duplicated regions within the human genome. Current: Assistant Professor, Department of Computer Science, Virginia Tech, Blacksburg
- 2001–2004 Rhea V. Samonte, Ph.D., postdoctoral research: Cytogenetic analysis of hominoid structural variation. Past: Laboratory Head and Assistant Professor, University of Philippines & GCCRD Project Manager, Manitoba Institute of Cell Biology; Current: Lab Director, PreventionGenetics, Marshfield
- 2001–2004 Ge Liu, Ph.D., postdoctoral research: Testing the model of the neutral theory of molecular evolution using comparative primate genomics. Current: Research Biologist, Bovine Functional Genomics Laboratory, USDA
- 1999–2002 Christine O'Keefe, Ph.D., postdoctoral research: Structural polymorphism within 16p11. Past: Research Associate, Cleveland Clinic Taussig Cancer Center; Current: Medical Writer, Cleveland HeartLab, Inc., Cleveland
- c) Masters**
- 2005–2006 Jonathan Bleyhl, M.S. (Genome Sciences), Detecting signatures of positive selection within recently duplicated genes (deceased)

- 2002–2004 Karen Hayden Miga, M.S. (Genetics), Structural variation between chimpanzee and human genomes, CWRU.
Current: Postdoctoral Scholar, Center for Biomolecular Science and Engineering, Univ of California, Santa Cruz
- 2003–2003 Tam Sneddon, M.S., Bioinformatics Diploma, External Placement, York University. Past: Research Scientist, NCBI, National Library of Medicine, NIH; Current: Senior Biocurator, Stanford University School of Medicine, San Francisco

d) Undergraduates

- 2016 Idara Akpandak, B.S., Biology, University of Maryland
- 2016–Present Naheed Arang, B.S., Microbiology & B.A., Integrated Science, UW
- 2015–2016 AnneMarie Welch, B.S., Microbiology, UW
- 2015–Present Vy Dang, B.S., Biochemistry and Microbiology, UW
- 2014 Ayorinde' Cooley, B.S., Biology, Morehouse College
- 2013 Claudia Espinoza, B.S., Biology, University of New Mexico
- 2013 Lana Harshman, B.S., Biology, UW
- 2011–2014 Kenneth M.K. Mark, B.S., Biochemistry, UW
- 2011, 2012 Daryl Dhanraj, B.S., Emory University
- 2011 Su Jen Khoo, B.S., Biotechnology, Penn State University
- 2011 Kian Hui Yeoh, B.S., Biotechnology, Penn State University
- 2010 Niels Hanson, B.S., Computer Science and Biology, University of British Columbia
- 2010 Farhad Hormozdiari, B.S., Computer Science, Simon Fraser University
- 2010 Iman Hajirasouliha, B.S., Computer Science, Simon Fraser University
- 2009, 2010 Eric Chiyembekeza, B.S., Emory University
- 2009–2011 Tiffany Vu, B.S., Biology, UW
- 2007 Neil Shafer, B.S., Biology, UW
- 2006–2007 Trisha Smith, B.S., Computer Science, UW
- 2006 Kerry Hall, B.S., Computer Science, UW
- 2005–2007 Maika Malig, B.S., Biology, UW, Morpheus Mouse Model
- 2002–2003 Samouil Lieberman, B.S., CWRU Electrical Engineering and Computer Sciences work study
- 2001–2002 Alexander Alekseyenko, B.S., CWRU Electrical Engineering and Computer Sciences, independent study, developing computational methods to incorporate sequence quality data into sequence alignments

e) Visiting Scientists/Scholars

- 2016 Yuan Liu, Kunming Institute of Zoology, The Chinese Academy of Sciences, China
- 2016–2017 Sultan Cingöz, Dokuz Eylül University School of Medicine, Izmir, Republic of Turkey
- 2015 Li-xin Yang, Kunming Institute of Zoology, The Chinese Academy of Sciences, China
- 2015 Yun-long Liu, Kunming Institute of Botany, The Chinese Academy of Sciences, China
- 2014–2015 Francesco Maria Calabrese, University of Bari, Italy
- 2013 Alexander Hoischen, Radboud University Medical Centre Nijmegen, The Netherlands
- 2012 Sebastien Jacquemont, University Hospital of Lausanne (CHUV), Switzerland
- 2012 Robert Barstead, University of Oklahoma & Oklahoma Medical Research Foundation
- 2011 Sònia Casillas, Institut de Biotecnologia i de Biomedicina Universitat Autònoma de Barcelona, Spain
- 2009–2010 Luis Alberto Pérez Jurado, Hospital Vall d'Hebron, Barcelona, Spain
- 2008–Present Mario Ventura, University of Bari, Italy
- 2008 Cenk Sahinalp, Simon Fraser University, Vancouver, BC, Canada
- 2008 Arcadi Navarro, Universitat Pompeu Fabra, Barcelona, Spain

f) Visiting Students/Interns

- 2017 Yuta Suzuki, University of Tokyo, Japan
- 2015 Fabio Anaclerio, University of Bari, Italy
- 2014–2016 Tianyun Wang, State Key Laboratory of Medical Genetics, Central South University, Changsha, China
- 2014 Navonil De Sarker, University of Calcutta, West Bengal, India
- 2014 Ahmed Mahfouz, Delft University of Technology, The Netherlands
- 2013 Giorgia Chiantante, University of Bari, Italy
- 2011, 2012 Javier Prado Martinez, Universitat Pompeu Fabra, Barcelona, Spain
- 2010 Niels Hanson, University of British Columbia, Vancouver, BC, Canada
- 2009–2010, 2011 Claudia Catacchio, University of Bari, Italy
- 2009, 2010 Belen Lorente, Universitat Pompeu Fabra, Barcelona, Spain
- 2009 Pietro D'Addabbo, University of Bari, Italy
- 2009 Iman Hajirasouliha, Simon Fraser University, Vancouver, BC, Canada

2008, 2009	Fereydoun Hormozdiari, Simon Fraser University, Vancouver, BC, Canada
2008	Giuliana Gianuzzi, University of Bari, Italy
2008	Karen Buysse, Ghent University Hospital, Belgium

g) Thesis Committees (*Chair)

2016–Present	Seung-been Steven Lee	Genome Sciences, UW	Advisor: Debbie Nickerson
2012–2016	P. Keolu O. Fox	Genome Sciences, UW	Advisor: Debbie Nickerson
2011–2015	Patrick Mitchell	Mol. Cell. Biol., UW	Advisor: Harmit Malik
2011–2014	Joshua Burton	Genome Sciences, UW	Advisor: Jay Shendure
2011–2014	Andrew Adey	Mol. Cell. Biol., UW	Advisor: Jay Shendure
2011–2014	Anna (Brosius) Sunshine	Genome Sciences, UW	Advisor: Maitreya Dunham
2011–2014	Rachel Diederich	Genome Sciences, UW	Advisor: James Thomas
2010–2013	Jacob Kitzman	Genome Sciences, UW	Advisor: Jay Shendure
2010–2014	Keisha Carlson	Genome Sciences, UW	Advisor: Christine Queitsch
2010–2013	Katrina Claw	Genome Sciences, UW	Advisor: Willie Swanson
2009–2012	Sarah Ng	Genome Sciences, UW	Advisor: Jay Shendure
2009–2011	Cailyn Spurrell	Genome Sciences, UW	Advisor: Mary-Claire King
2009–2013	Ray Malfavon-Borja	Genome Sciences, UW	Advisor: Harmit Malik
2009–2011	Alexander Nord	Genome Sciences, UW	Advisor: Mary-Claire King
2008–2012	Efrem Lim	Microbiology, UW/FHRC	Advisor: Michael Emerman
2007–2012	Kyle Siebenthall	Genome Sciences, UW/FHCRC	Advisor: Barb Trask
2007–2010	Thomas Nicholas	Genome Sciences, UW	Advisor: Joshua Akey
2007–2010	Diane Dickel	Genome Sciences, UW	Advisor: Mary-Claire King
2007–2010	Troy Zerr	Genome Sciences, UW	Advisor: Debbie Nickerson
2007–2009	Eithon Cadag	BHI, UW	Advisor: Peter Myler
2005–2009	Johanna Eddy	Mol. Cell. Biol., UW	Advisor: Nancy Maizels
2005–2007	Molly Orton	Mol. Cell. Biol., UW/FHCRC	Advisor: Harmit Malik
2005–2007	Jennifer Gogarten	Genome Sciences, UW/FHCRC	Advisor: Barb Trask
2004–2007	Nathan Clark	Genome Sciences, UW	Advisor: Willie Swanson
2003–2005	Liesel Brihn*	Genetics, CWRU	Advisor: Joe Nadeau
2002–2004	Cory Valley	Genetics, CWRU	Advisor: Hunt Willard
2001–2005	Toshimori Kitami	Genetics, CWRU	Advisor: Joe Nadeau
2001–2004	Michelle Holko	Genetics, CWRU	Advisor: Bryan Williams
2000–2004	Can Alkan	EECS, CWRU	Advisor: Cenk Sahinalp
2000–2003	Erica Burner	Genetics, CWRU	Advisor: Anne Matthews
1999–2003	Debra Matthews	Genetics, CWRU	Advisor: Aravinda Chakravarti
1998–2002	Mary Schueler*	Genetics, CWRU	Advisor: Hunt Willard
1998–2004	Jim Amos-Landgraf	Genetics, CWRU	Advisor: Hunt Willard
1998–2003	Minerva Carrasquillo	Genetics, CWRU	Advisor: Aravinda Chakravarti
1998–2003	David Satinover*	Genetics, CWRU	Advisor: Stuart Schwartz

h) External Examiner/Official Opponent of Ph.D. Dissertations/Defense

2010	Andrés Ingason	University of Copenhagen	Advisor: Thomas Werge
2010	Anna Wetterbom	Uppsala University	Advisor: Ulf Gyllensten
2007	Tomas Marques-Bonet	University of Pompeu-Fabra	Advisor: Arcadi Navarro
2006	Erik Arner	Karolinska Institutet	Advisor: Bjorn Andersson
2006	Louie van de Lagemaat	University of British Columbia	Advisor: Dixie Mager
2005	Ines Hellman	MPI, University of Leipzig	Advisor: Svante Pääbo
1999	Alyssa Barry	University of Melbourne	Advisor: Andy Choo

INVITED SEMINARS AND LECTURES

(1997–Present: 430 invited talks, seminars and keynote/plenary lectures)

1997

- Invited Seminar, Kaiser Permanente, Pediatrics Society, “Fragile X Syndrome: Mechanism and Clinical Implications,” Pleasanton, CA, January
- Invited Speaker, Chromosome 16 Workshop, Toronto, ON, Canada, March

1998

- Invited Speaker, EMBO Workshop, Hammersmith Hospital, “Trinucleotide Expansion Diseases in the Context of Mini- and Microsatellite Evolution,” London, UK, April

- Speaker, Cold Spring Harbor Laboratory (CSHL): Genome Mapping, Sequencing and Biology, Cold Spring Harbor, NY, May
- Invited Seminar, NIH: Genomic Alterations in Genetic Disease: Mechanism of Structural Rearrangements, Bethesda, MD, June
- Invited Speaker, Banbury Center Meeting: “Y Chromosome Disease and Evolution,” Lloyd Harbor, NY, July
- Invited Seminar, Genoplex (Biotechnology Company), Denver, CO, October
- Speaker, American Society of Human Genetics (ASHG), Denver, CO, October
- Invited Seminar, Department of Genetics, University of Pennsylvania Medical Center, (Host: Dr. Haig Kazazian, Jr.), Philadelphia, PA, November
- Invited Seminar, Molecular Biology and Biotechnology Departmental Seminar Series, Department of Molecular Biology, UW, Seattle, WA, December
- Invited Seminar, Computational Biology Seminar Series, UW, Seattle, WA, December

1999

- Seminar, Afternoon Series in Molecular Biology and Cell Biology, Cleveland, OH, January
- Invited Seminar, NIH Human Genome Lecture Series, Bethesda, MD, January
- Invited Seminar, Marshfield Clinic, Marshfield Clinic Wednesday Seminar, (Host: Dr. James Weber), Marshfield, WI, March

2000

- Invited Speaker, Reproductive Sciences 2000 “SNP Variation and Detection,” Salt Lake City, UT, February
- Invited Seminar, Department of Human Genetics, University of Chicago, Chicago, IL, March
- Invited Speaker, Banbury Center Meeting: Great Apes, Phenotypes and Genotypes, Lloyd Harbor, NY, March
- Invited Seminar, Department of Biological Sciences, University of Alberta, Edmonton, AB, Canada, April
- Invited Seminar, Department of Genetics, Ottawa General Hospital, Ottawa, ON, Canada, April
- Invited Speaker, Department of Energy “Exceptional Chromosomal Regions of the Human Genome,” Rockville, MD, May
- Invited Seminar, Celera Genomics, Rockville, MD, July
- Invited Speaker, Molecular Cytogenetics Gordon Conference, University of Oxford, Oxford, UK, July
- Invited Participant and Speaker, Whitehead MIT Genome Center, International Human Genome Sequencing Consortium: Genome Sequence Analysis, Boston, MA, August
- Invited Speaker, Workshop on Gene Order Dynamics, Montreal, PQ, Canada, September
- Organizer & Speaker, ASHG “Origins and Primate Evolution,” Philadelphia, PA, October
- Invited Participant, Children’s Hospital of Pennsylvania, HGP Sequence Analysis Group: International Human Sequencing Consortium, Philadelphia, PA, October
- Invited Speaker, NetGenics-Athersys Mini Symposium:, Computational Genetics Sequence Analysis and Annotation, Cleveland, OH, October

2001

- Invited Speaker, Advances in Genome Biology and Technology, Marco Island, FL, February
- Invited Lecture, NIH Lecture Series: Human Genome Sequence, Bethesda, MD, March
- Invited Speaker, Bioinformatics Policy Forum, CWRU, Cleveland, OH, March
- Invited Speaker, Banbury Center Meeting: Genomic Annotation Workshop, Lloyd Harbor, NY, March
- Guest Speaker, Advanced Genome Sequence Analysis Course, Cold Spring Harbor, NY, March
- Invited Speaker, Keystone Symposium: Human Genetics and Genomics, Breckenridge, CO, March
- Invited Speaker, American Genetic Association: Primate Evolutionary Genomics, San Diego, CA, May
- Invited Speaker, HHMI Joint Sequencing Workshop, Chevy Chase, MD, June
- Invited Speaker, Gordon Research Conference: Mutagenesis, Lewiston, ME, July
- Invited Speaker, Gordon Research Conference: Human Molecular Genetics, Newport, RI, August
- Invited Seminar, Baylor College of Medicine (Host: Juan Botas), Houston, TX, September
- Invited Speaker, Cold Spring Harbor Meeting on Computational Biology, Cold Spring Harbor, NY, September
- Invited Seminar, University of Michigan (Host: John Moran), Ann Arbor, MI, October
- Plenary Speaker, Genome and Sequence Analysis Conference (Host: Craig Venter), San Diego, CA, October
- Invited Seminar, CWRU Blood Group, (Host: Sandy Markowitz), Cleveland, OH, November
- Invited Seminar, Sick Children’s Hospital of Toronto (Host: Lap-Chee Tsui), Toronto, ON, Canada, November
- Invited Speaker, Salk Institute, Conference on Human Origins, La Jolla, CA, November
- Invited Seminar, Children’s Hospital of Pennsylvania (Host: Bev. Emanuel), Philadelphia, PA, November
- Invited Seminar, Department of Human Genetics, UCLA (Host: Nelson Freimer), Los Angeles, CA, December
- Invited Participant, NHGRI Genome Project Planning Session, Goals 2003–2008, Airlie, VA, December

2002

- Plenary Speaker, DOE Contractor Genome Meeting IX, Oakland, CA, January
- Invited Graduate Student Speaker, Department of Genomic Sciences, UW, Seattle, WA, February
- Invited Speaker, McDermott Center for Human Genetics, Southwestern Medical Center, Dallas, TX, March
- Invited Seminar, Department of Human Genetics, Emory University, Atlanta, GA, March
- Invited Seminar, Department of Biological Chemistry, University of California Irvine, Irvine, CA, March
- Invited Speaker, American Association of Anthropological Genetics, Buffalo, NY, April
- Plenary Speaker, Human Genome Meeting (HGM 2002), Shanghai, China, April
- Plenary Speaker, RECOMB 2002, Washington, DC, April
- Invited Participant, Eleventh International Strategy Meeting on Human Genome Sequencing, NY, May
- Invited Seminar, Washington University School of Medicine, Genetics, St. Louis, MO, May
- Invited Lecture, Frontiers of Genomics VI, University of Madison-Wisconsin, Madison, WI, May
- Invited Speaker, European Human Genetics Meeting, Strasbourg, France, May
- Catalyst Speaker, Chimpanzee Conference One, Yerkes Regional Primate Center, Atlanta, GA, June
- Invited Speaker, NSF “Genomics of Human Origins,” National Science Foundation, Arlington, VA, July
- Invited Lecture, European School of Genetic Medicine, Bertinoro, Italy, August
- Invited Seminar, Department of Zoology, Miami University, Oxford, OH, September
- Invited Speaker, 5th International Meeting on Single-Nucleotide Polymorphism and Complex Genome Analysis, Reykjavik, Iceland, October
- Invited Seminar, Decode Genetics, Reykjavik, Iceland, October
- Invited Symposium, ASHG Meeting, Baltimore, MD, October
- Invited Seminar, Institute of Genetic Medicine, University of Southern California, Los Angeles, CA, November
- Invited Participant, NHGRI meeting “Beyond the Beginning: The Future of Genomics II,” Airlie, VA, November
- Invited Seminar, Department of Pharmacology, Southwestern Medical Center, Dallas, TX, December
- Invited Seminar, Yale School of Medicine, New Haven, CT, December

2003

- Invited Seminar, Carolina Center for Genome Sciences, University North Carolina, Chapel Hill, NC, February
- Invited Seminar, Distinguished Lecture in Genome Sciences, Lawrence Berkeley National Laboratory, Berkeley, CA, Feb.
- Invited Seminar, Joint Genome Institute, DOE, Walnut Creek, CA, February
- Invited Seminar, Genome Sciences, UW, Seattle, WA, March
- Organizer, Human Genome Meeting 2003, Cancun, Mexico, April
- Invited Speaker, Bioinformatics 2003, SOCBIN, Helsinki, Finland, May
- Invited Symposium, Genome of Homo sapiens. 68th Annual Cold Spring Harbor Symposium, Cold Spring Harbor, NY, May
- Invited Seminar, Institute of Genetic Medicine, Johns Hopkins University, Baltimore, MD, June
- Invited Speaker, XIX International Congress of Genetics, Melbourne, Australia, July
- Plenary Speaker, 13th North American Colloquium on Animal Cytogenetics and Gene Mapping, Louisville, KY, July
- Invited Speaker, Gordon Research Conference: Human Genetics and Genomics, Waterville, ME, August
- Plenary Speaker, European Society of Cytogenetics, Bologna, Italy, September
- Invited Seminar, British Society of Human Genetics, York, UK, September
- Invited Seminar, Max Planck Institute for Molecular Anthropology, Leipzig, Germany, September
- Plenary Speaker, European Life Scientist Organization (ELSO 2003), Dresden, Germany, September
- Invited Speaker, RECOMB Satellite: Comparative Genomics IMA, Minneapolis, MN, October
- Invited Seminar, Department of Biological Sciences, Louisiana State University, Baton Rouge, LA, November
- Invited Seminar, Department of Human Genetics, McGill University, Montreal, PQ, Canada, November
- Invited Seminar, Department of Human Genetics, University of Chicago, Chicago, IL, November
- Invited Seminar, Department of Biological Sciences, Program in Molecular and Computational Biology, University of Southern California, Los Angeles, CA, December
- Invited Seminar, Department of Pathology, CWRU, Cleveland, OH, December

2004

- Invited Speaker, Keystone Symposium: Human Genome Sequence Variation, Breckenridge, CO, January
- Invited Speaker, Evolutionary Genomics, University of Arizona, Tucson, AZ, January
- Invited Seminar, Rutgers University, New Brunswick, NJ, February
- Invited Seminar, Fred Hutchinson Cancer Research Center, Seattle, WA, February
- Invited Participant, Concept Development/Planning Meeting: The Development of a Chimpanzee Molecular Biology Discovery Resource, Coriell Institute, Camden, NJ, February
- Invited Speaker, Sequencing the Chimpanzee Genome, UCSC, San Diego, CA, March

- Invited Speaker, La Jolla Origins of Humans, Salk Institute, San Diego, CA, March
- Plenary Speaker, HUGO Genome Meeting, Berlin, Germany, April
- Keynote Speaker, 2004 Genetics Symposium, Penn State University, State College, PA, May
- Invited Speaker, Genomes and Evolution, SMCBE, Penn State University, PA, June
- Invited Lecturer, The Jackson Laboratory: Experimental and Medical Genetics Short Course, Bar Harbor, ME, July
- Invited Participant, Workshop to Resequence the Human Genome, NIH, Bethesda, MD, July
- Co-Organizer & Speaker, Understanding Human Genome Evolution, Bertinoro, Italy, September
- Invited Speaker, ASHG, Toronto, ON, Canada, October
- Invited Speaker, Art Institute Seattle University, Seattle, WA, October
- Invited Seminar, Department of Pathology and Genetics, Uppsala University, Uppsala, Sweden, November
- Invited Seminar, Department of Genomics and Bioinformatics, Karolinska Institutet, Stockholm, Sweden, November
- Invited Participant, ISCN Meeting, Vancouver, BC, Canada, December

2005

- Invited Seminar, Institute for Genome Sciences and Policy, Duke University, NC, January
- Invited Seminar, UBC Genome Sequencing Center, Vancouver, BC, Canada, February
- Invited Seminar, Department of Computing Sciences, Simon Fraser University, Vancouver, BC, Canada, February
- Invited Speaker, International Conference on Primate Genomics, Seattle, WA, March
- Invited Lectures (2), Functional Genomics Neuroscience, Panum Institute, University of Copenhagen, Denmark, April
- Frontiers Lecture in Biological Research, Stanford University School of Medicine, CA, April
- Invited Speaker & Co-organizer, Biology of Genomes, Cold Spring Harbor, NY, May
- Invited Speaker, Genome Structural Variation Symposium, Toronto, ON, Canada, July
- Invited Speaker, Gordon Research Conference: Genomics and Genetics, Newport, RI, July
- Invited Speaker, Gordon Research Conference: Chromosome Dynamics, New London, NH, July
- Plenary Speaker & Moderator, David W. Smith Workshop, Iowa City, IA, August
- Invited Speaker, American Society of Primatologists, Portland, OR, August
- Distinguished Lecture Series, Wellcome Trust, Sanger Center, Hinxton, UK, September
- Invited Plenary, European Science Foundation: Functional Genomics and Disease, Oslo, Norway, September
- Invited Rudbeck Seminar, Uppsala University, Sweden, September
- Invited Lecture, British Society of Human Genetics, York University, York, UK, September
- Keynote Speaker, American Society of Plant Biologists, Snowbird, UT, October
- Invited Speaker, Marie Curie Conference on ArrayCGH and Molecular Cytogenetics, Monopoli, Bari, Italy, October
- Invited Speaker, ASHG, Salt Lake City, UT, October
- Invited Speaker, Center for Excellence in Genome Research, USC, Los Angeles, CA, November
- Invited Lecture, Population Biology, Evolution and Ecology, Emory University, Atlanta, GA, December

2006

- Keynote Speaker, Symposium: DNA Structure, Genomic Rearrangements and Human Disease, Houston, TX, March
- Invited Speaker, Banbury Center Meeting: Autism Genetics Meeting, Lloyd Harbor, NY, March
- Invited Seminar, Nemours Biomedical Research Center, Alfred I Dupont Hospital, Wilmington, DE, March
- Frontiers of Genomics Lecture, Center for Genome Research, National University of Mexico, Cuernavaca, Mexico, April
- Invited Speaker, 2nd International Meeting on Cryptic Chromosomal Rearrangements in Mental Retardation and Autism, Troina, Italy, April
- Invited Plenary, Annual European Society of Human Genetics Meeting, Amsterdam, The Netherlands, May
- Invited Speaker, 3rd Annual HapMap Analysis Meeting, Broad Institute, Boston, MA, May
- Invited Speaker, National Advisory Council for Human Genome Research, Bethesda, MD, May
- Invited Plenary, Human Genome Meeting (HUGO) 2006, Helsinki, Finland, May
- Invited Lecture, Dahlem Colloquium, Max Planck Institute for Human Molecular Genetics, Berlin, Germany, June
- Invited Lecture, 47th Short Course on Mammalian Genetics, Bar Harbor, ME, July
- Invited Speaker, International Congress of Human Genetics, Brisbane, Australia, August
- Invited Speaker, Chimpanzees in Research Conference, Yerkes National Primate Center, Atlanta, GA, October
- Invited Speaker, ASHG, New Orleans, LA, October
- Invited Speaker, NIAID Population Genetics Annual Meeting, Washington, DC, November
- Invited Speaker, NAS Sackler Colloquium, The New Comparative Biology of Human Nature, Orange County, CA, November
- Invited Lecture, Biosciences Series on Evolution, Universitat Autònoma de Barcelona, Barcelona, Spain, November

2007

- Invited Lecture, Pompeu-Fabra University, Department of Human Genetics, Barcelona, Spain, January
- Invited Student Seminar, Molecular Genetics Program, Emory University, Atlanta, GA, January
- Invited Speaker, Advances in Genome Biology and Technology, Marco Island, FL, February
- Invited Seminar, Evolving Genome Seminar Series, University of Michigan, Ann Arbor, MI, March
- Invited Seminar, Department of Genetics, University of Wisconsin, Madison, WI, May
- Invited Seminar, Waisman Center, University of Wisconsin, Madison, WI, May
- Invited Speaker, Scientific Breakthroughs of the Year Session, American Thoracic Society Meeting, San Francisco, CA, May
- Invited Speaker, FASEB Mobile Element Meeting, Tucson, AZ, June
- Invited Seminar, Department of Developmental Biology, Pasteur Institute, Paris, France, June
- Invited Speaker, The Jackson Laboratory: Annual Birkenmeier Lectureship, Bar Harbor, ME, June
- Invited Plenary Speaker, European Conference of Cytogenetics, Istanbul, Turkey, July
- Invited Speaker & Organizer, Gordon Research Conference: Human Genetics and Genomics, Newport, RI, July
- Invited Plenary Speaker, Brazilian Congress of Genetics, Aguas de Lindoia, Brazil, September
- Invited Faculty & Speaker, Young Neuroscientists' Workshop, Solvang, CA, September
- Invited Speaker, HUGO Mutation Detection, Xiamen, China, September
- Invited Plenary Speaker, World Congress of Psychiatric Genetics, New York, NY, October
- Invited Seminar, Mayo Clinic, Rochester, MN, October
- Invited Plenary Speaker, NIH Intramural Sequencing Center 10th Anniversary Symposium, Bethesda, MD, October
- Invited Speaker, Applied Biosystems Symposium, ASHG, San Diego, CA, October
- Invited Seminar, John Innes Centre, Norwich, UK, November
- Invited Speaker & Host, Nature Genome Structural Variation and Evolution Symposium, Seattle, WA, November
- Invited Speaker, Molecular Medicine Public Lecture Series, UW, Seattle, WA, December
- Invited Seminar, Department of Molecular Biology Seminar Series at Massachusetts General Hospital, Boston, MA, Dec.
- Invited Seminar, John Hopkins University, Department of Molecular Biology and Genetics, Baltimore, MD, December

2008

- Invited Seminar, St. Jude Children's Research Hospital, Danny Thomas Lecture Series, Memphis, TN, January
- Invited Seminar, University of California, San Francisco, Seminars in Biomedical Science Series, San Francisco, CA, January
- Invited Lecture, UW Cardiovascular Health Research Unit, Works-in-Progress Series, Seattle, WA, February
- Invited Speaker, 2008 American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting, Phoenix, AZ, Mar.
- Invited Speaker, Genomic Disorders, Wellcome Trust Conference Centre, Genomic Disorders, Hinxton, UK, March
- Invited Seminar, UC Davis Genome Center, Forefronts of Genomics Colloquium, Davis, CA, March
- Invited Speaker, 3rd International Conference on Primate Genomics & Human Disease Conference, Seattle, WA, April
- Invited Seminar, Genentech, San Francisco, CA, April
- Invited Speaker & Session Chair, 1000 Genomes Project & CSHL: Biology of Genomes Meeting, Cold Spring Harbor, NY, May
- Invited Speaker, IHG Symposium: Genomics and Personalized Medicine, University of Minnesota, Minneapolis, MN, June
- Invited Speaker, Molecular Genetics Consortium Workshop, Atlanta, GA, June
- Invited Seminar, Illumina, San Diego, CA, June
- Invited Speaker, XX International Congress of Genetics, Berlin, Germany, July
- Invited Speaker, Genomics of Common Disease, Broad Institute, Boston, MA, September
- Invited Speaker, AnEUploidy Workshop, University of Geneva, Geneva, Switzerland, September
- Invited Plenary, FISV Congress (Federation of Life Scientist Meeting), Riva del Garda, Italy, September
- Invited Plenary, Human Genome Meeting (HUGO, HGM2008), Hyderabad, India, September
- Invited Speaker, Human Variome Meeting, Hyderabad, India, September
- Invited Speaker, CSHL: Personalized Genomes Meeting, Cold Spring Harbor, NY, October
- Invited Speaker, PROUST Genes at Work on Time Conference, Torino, Italy, October
- Invited Lecture, Graduate Student Symposium, Baylor College of Medicine, Houston, TX, October
- Invited Seminar, Department of Genetics, Emory University, Atlanta, GA, November
- Invited Seminar, Molecular Cell, Biology and Bioinformatics Program, Virginia Tech, VA, November
- Invited Speaker, 1000 Genomes Meeting, ASHG Meeting, Philadelphia, PA, November
- Invited Speaker, Australian Health and Medical Research Congress, Brisbane, Australia, November
- Invited Participant, 2nd International Consortium Workshop on Clinical Cytogenetic Arrays, Bethesda, MD, December
- Invited Speaker, American College of Neuropsychopharmacology (ACNP) 47th Annual Meeting, Scottsdale, AZ, December

2009

- Invited Seminar, Indiana University Department of Biology, Bloomington, IN, January
- Invited Seminar, Miami 2009 Winter Symposium: Interpreting the Human Genome, Miami, FL, January

- Invited Participant & Discussion Leader, NHGRI workshop “Dark Matter of Genomic Associations with Complex Diseases,” Bethesda, MD, February
- Invited Seminar, Washington University Department of Genetics Spring Seminar Series, St. Louis, MO, February
- Invited Seminar, Comprehending Copy Number Variation Meeting, San Diego, CA, March
- Invited Seminar, University of California San Diego Genetics and Genomics Seminar Series, San Diego, CA, March
- Invited Seminar, Arizona Initiative for the Biology of Complex Diseases (ABCD) Colloquium: Problems in Complex Disease Biology, Tucson, AZ, March
- Invited Keynote Plenary, International Congress on Schizophrenia Research, San Diego, CA, March
- Invited Seminar, Washington University in St. Louis Symposium Celebrating the Darwin Bicentennial, St. Louis, MO, March
- Invited Seminar, Morehouse College Biology Seminar Series, Atlanta, GA, March
- Invited Seminar, 20th Annual Meeting of the German Society of Human Genetics, Aachen, Germany, April
- Invited Seminar, Pharmacogenetics Research Network (PGRN), Rochester, MN, April
- Invited Seminar, Cornell University Department of Molecular Biology and Genetics, Ithaca, NY, April
- Invited Seminar, The Institute of Genetics and Biophysics, Naples, Italy, April
- Invited Seminar, European Genetics Foundation Course in Medical Genetics, Bertinoro, Italy, April
- Invited Keynote, Sequencing, Finishing and Analysis in the Future, Santa Fe, NM, May
- Invited Seminar, Fred Hutchinson Cancer Research Center, Seattle, WA, May
- Invited Speaker, 74th Cold Spring Harbor Symposium: Evolution: The Molecular Landscape, Cold Spring Harbor, NY, May
- Invited Speaker, 8th International Workshop on Advanced Genomics, Tokyo, Japan, June
- Invited Speaker, "Wednesdays at the Genome" Public Lecture Series, UW, Seattle, WA, July
- Speaker & Session Chair, Gordon Research Conference: Human Genetics and Genomics, Biddeford, ME, July
- Invited Seminar, The Jackson Laboratory: 50th Annual Genetics Course, Bar Harbor, ME, July
- Invited Plenary Workshop, Association for the Advancement of Animal Breeding and Genetics: Comparative Genomics Workshop, Rowland Flat, Australia, September
- Invited Session & Presidential Symposium Speaker, ASHG Annual Meeting, Honolulu, HI, October
- Invited Speaker, American Society of Nephrology's (ASN) 42nd Annual Renal Week Meeting, San Diego, CA, October
- Invited Speaker, 2009 PQG Conference: Human Genetic Variation, Health and Disease: New Knowledge, New Quantitative Challenges, Boston, MA, November
- Invited Course Presenter, CSHL: Advanced Sequencing Technologies & Applications, Cold Spring Harbor, NY, November
- Invited Speaker, Banbury Center Meeting: Structural Variation in the Human Genome, Lloyd Harbor, NY, November
- Invited Speaker, Department of Human Genetics Seminar Series, University of Chicago, Chicago, IL, December
- Invited Distinguished Lecturer, American College of Neuropsychopharmacology (ACNP) Annual Meeting, Miami, FL, Dec.

2010

- Invited Plenary Speaker, Plant and Animal Genome (PAG) XVIII Meeting, San Diego, CA, January
- Invited Speaker, VanBUG, Vancouver, BC, Canada, January
- Invited Speaker, Symposium on Transformational Genomics Honoring Dan Pinkel, PhD, San Francisco, CA, February
- Invited Speaker & Co-Chair, CARTA Symposium: The Evolution of Human Biodiversity, UCSD, San Diego, CA, March
- Invited Speaker, Uppsala University, Uppsala, Sweden, March
- Invited Speaker, Genomic Disorders 2010: Copy Number and Sequence Variation in Mendelian and Complex Traits, Wellcome Trust Conference Centre, Hinxton, UK, March
- Invited Keynote Speaker, Stanford Genomics Symposium, Stanford, CA, April
- Invited Speaker, Genes, Genomes, and Pediatric Disease (GGPD) Seminar Series, Children's Hospital of Philadelphia, PA, April
- Invited Keynote Speaker, American Cytogenetics Conference (ACC), Niagara Falls, ON, Canada, May
- Invited Speaker, CINP (Collegium Internationale Neuro-Psychopharmacologicum) World Congress, Hong Kong, China, June
- Invited Speaker, Nobel Symposium: Genetics in Medicine, Stockholm, Sweden, June
- Invited Speaker, European Molecular Biology Laboratory (EMBL) Human Variation: Cause and Consequence, Heidelberg, Germany, June
- Invited Speaker, Berlin Summer Meeting: Quantitative Genomics, Berlin, Germany, June
- Invited Speaker, Third International Standard Cytogenomic Array (ISCA) Workshop, Bethesda, MD, June
- Invited Participant, NHGRI Planning for the Future of Genomics meeting “Foundational Research and Applications in Genomic Medicine,” Warrenton, VA, July
- Invited Speaker, European Science Foundation (ESF) Next Generation Sequencing Meeting, Leiden, The Netherlands, August
- Invited Speaker, Washington University School of Medicine, St. Louis, MO, September
- Invited Speaker, 2nd AnEUploidy Workshop, Split, Croatia, September
- Invited Speaker, Sig. K. Thoresen Foundation and The Norwegian Academy of Sciences “Genomic and Genetic Aspects for Human Health and Disease” Symposium, Oslo, Norway, September

- Invited Speaker, University of Adelaide, Adelaide, Australia, September
- Invited Speaker, Murdoch Children's Research Institute (MCRI), Melbourne, Australia, September
- Invited Plenary Speaker, OzBio2010: The molecules of life: From discovery to biotechnology, Melbourne, Australia, September
- Invited Course Lecturer, CSHL: Advanced Sequencing Technologies & Applications, Cold Spring Harbor, NY, October
- Invited Speaker, Boston University Genome Science Institute, Boston, MA, October
- Invited Speaker, UCLA Bioinformatics Seminar Series, Los Angeles, CA, November
- Invited Speaker, ASHG Annual Meeting, Washington, DC, November
- Invited Speaker, Scripps Translational Science Institute, La Jolla, CA, November
- Invited Public Symposium Session, Society for Neuroscience Annual Meeting, San Diego, CA, November
- Invited Lecturer, Utrecht University Cancer Genomics & Developmental Biology (CGDB) Masterclass, Doorwerth, The Netherlands, December
- Invited Speaker, University of Utah School of Medicine Seminar Series, Salt Lake City, UT, December

2011

- Speaker & Organizer, Keystone Symposium: Functional Consequences of Genome Structural Variation, Steamboat Springs, CO, January
- Invited Workshop Presenter, Workshop on Comparative Genomics, Český Krumlov, Czech Republic, January
- Invited Speaker, First Annual International Standards for Cytogenomic Arrays (ISCA) Consortium Conference, Atlanta, GA, Jan.
- Invited Speaker, Johns Hopkins University School of Medicine Institute of Genetic Medicine (IGM) Seminar Series, Baltimore, MD, February
- Invited Speaker, Human Genomics: The Next 10 Years (Scripps Seaside Forum), San Diego, CA, February
- Invited Speaker, HUGO's 15th Human Genome Meeting (HGM 2011): Genomics of Human Diversity and Hereditary Disorders, Dubai, United Arab Emirates, March
- Invited Speaker, EMBL Eminent Speaker Seminar Series, Rome, Italy, March
- Invited Keynote Speaker, RECOMB 2011 Conference: 15th Annual International Conference on Research in Computational Molecular Biology, Vancouver, BC, Canada, March
- Invited Speaker, 8th GeneMappers Conference, Hobart, Australia, April
- Invited Speaker, CARTA Symposium: The Genetics of Humanness, UCSD, San Diego, CA, April
- Invited Speaker, Duke University Program in Genetics & Genomics Seminar Series, Durham, NC, April
- Invited Speaker, HudsonAlpha Institute for Biotechnology Seminar Series, Huntsville, GA, April
- Invited Speaker, 2011 American Asthma Foundation (AAF) Annual Scientific Meeting, San Francisco, CA, May
- Invited Speaker, Frontiers in Biology Seminar, Stanford University, San Francisco, CA, May
- Invited Speaker, Department of Molecular and Medical Genetics (MMG) Seminar, Oregon Health and Science University, Portland, OR, June
- Invited Keynote Speaker, Signature Scientific Microarray Conference, Spokane, WA, June
- Invited Speaker, Mouse Lemur Genetics and Genomics: Emerging Opportunities, Janelia Farm Research Campus, DC, June
- Invited Speaker, NIH/NCI Frederick Campus, Frederick, MD, June
- Invited Speaker, UC Davis MIND Institute, Sacramento, CA, June
- Invited Keynote Speaker, 8th European Cytogenetics Conference (ECA), Porto, Portugal, July
- Invited Speaker, University of Porto, CIBIO, Porto, Portugal, July
- Invited Lecturer, Workshop on Comparative Genomics, North America 2011, Fort Collins, CO, July
- Invited Speaker, Gordon Research Conference: Human Genetics and Genomics, Newport, RI, July
- Invited Speaker, The Jackson Laboratory: 52nd Annual Genetics Course, Bar Harbor, ME, July
- Invited Speaker, Autism Sequencing Consortium Mtg, NIH, Bethesda, MD, September
- Invited Plenary, The 3rd EMBO Meeting: Advancing the Life Sciences, Vienna, Austria, September
- Invited Speaker, 2011 SFARI Annual Meeting (Simons Foundation), Washington, DC, September
- Invited Speaker, Symposium on the Emerging Genetics and Neurobiology of Severe Mental Illness, The Broad Institute, Boston, MA, September
- Invited Session Speaker, 12th International Congress of Human Genetics (ICHG) and the 61st ASHG Annual Meeting, Montreal, QB, Canada, October
- Invited Speaker, NIEHS CNV Meeting, Montreal, QB, Canada, October
- Invited Keynote, CSHL: Genome Informatics Meeting, Cold Spring Harbor, NY, November
- Invited Speaker, University of Lausanne BIG Seminar, Lausanne, Switzerland, November
- Invited Speaker, 25th Annual Roland D. Pinkham, M.D. Basic Science Lectureship Diversity and Evolution of the Human Genome: From "Origins" to Evo-Devo, Seattle, WA, November
- Invited Speaker, Banbury Center: Psychiatric Genomics, Cold Spring Harbor, NY, December

2012

- Invited Speaker, UC Davis MIND Institute, Sacramento, CA, January
- Invited Speaker, Baylor Genetics Anniversary Gala, Symposium & Retreat, Houston, TX, January
- Invited Speaker, Institute for Integrative Genome Biology (IIGB) Seminar Series, University of California, Riverside, CA, Feb.
- Invited Speaker, Leiden Genetic Colloquia (LGC) Lecture Series, Leiden, The Netherlands, February
- Invited Speaker, Scripps Translational Science Institute, The Future of Genomic Medicine V Conference, La Jolla, CA, March
- Invited Lectureship & Speaker, Eva Raik Lecture, RCPA: Pathology Update 2012, Sydney, Australia, March
- Invited Speaker, 16th Human Genome Meeting 2012 (HGM2012), Sydney, Australia, March
- Invited Speaker, Memorial Sloan-Kettering Cancer Center President's Research Seminar, New York, NY, March
- Invited Speaker, Department of Genetics Harvard Medical School, Boston, MA, April
- Invited Speaker, The Broad Institute, Boston, MA, April
- Invited Speaker, Lewis-Sigler Institute Princeton University, Princeton, NJ, April
- Invited Seminar, Roche-Nature Medicine Translational Neuroscience Symposium, Buonas, Switzerland, April
- Invited Speaker, 2012 American Asthma Foundation (AAF) Annual Scientific Meeting, San Francisco, CA, May
- Invited Plenary Speaker, International Dermatogenetics Workshop, Beijing, China, June
- Invited Plenary Speaker & Session Speaker, ESHG European Human Genetics Conference 2012, Nürnberg, Germany, June
- Invited Keynote Lecture, MMI Education & Training: Molecular Medicine Ireland, Dublin, Ireland, June
- Invited Speaker, Gordon Research Conference: Neural Development, Newport, RI, August
- Invited Speaker, International Workshop: Structural and Functional Diversity of Genomes, Brno, Czech Republic, September
- Invited Speaker, CIBERER 2012 International Symposium: Advances in the Biomedical Research of ASD, Barcelona, Spain, Sept.
- Invited Speaker, Ernst Klenk Symposium in Molecular Medicine: The Genomic Future of Medicine, Cologne, Germany, Sept.
- Invited Speaker, UNC Chapel Hill: Genome Sciences Building Opening Symposium, Chapel Hill, NC, October
- Invited Keynote Lecture, Department of Genetics at University of Alabama at Birmingham & HudsonAlpha Institute for Biotechnology: 7th Annual Genetics Scientific Retreat, Huntsville, GA, October
- Invited Speaker, Pharmacogenomics Research Network Meeting (PGRN), Seattle, WA, October
- Invited Speaker, Seattle Pacific University, Seattle, WA, November
- Invited Seminar, Center for Human Genetics KU Leuven, Leuven, Belgium, December
- Invited Speaker, Wellcome Trust Centre for Human Genetics Seminar, Oxford, UK, December

2013

- Invited Seminar, Albert Einstein College of Medicine Department of Genetics, New York, NY, January
- Invited Speaker, Keystone Symposium: New Frontiers in Cardiovascular Genetics Beyond GWAS, Tahoe City, CA, January
- Invited Speaker, SALK/IPSEN/NATURE Symposium on Biological Complexity: Molecular Biology of Psychiatric Disorders, San Diego, CA, January
- Invited Seminar, Stanford Institute for Neuro-Innovation and Translational Neurosciences (SINTN), Stanford, CA, February
- Invited Speaker, University of Texas MD Anderson Cancer Center John H. Blaffer Lecture Series, Houston, TX, February
- Invited Lectureship, First Harris Lewin Lecture, Institute for Genomic Biology at University of Illinois at Urbana-Champaign, Urbana, IL, February
- Invited Speaker, Scripps Translational Science Institute, The Future of Genomic Medicine VI Conference, La Jolla, CA, March
- Invited Lectureship, 9th Annual Evelyn Galman Spritz Endowed Lecture, Human Medical Genetics and Genomics Program, University of Colorado, Anschutz Medical Campus, Aurora, CO, March
- Invited Speaker, UCSF Biochemistry Seminar Series, San Francisco, CA, March
- Invited Speaker, The Rockefeller University Lecture Series, New York City, NY, March
- Invited Plenary, 8th International Meeting on CNVs & Genes in Intellectual Disability & Autism, Troina, Italy, April
- Invited Plenary, GENCODYS International Conference, Paphos, Cyprus, April
- Invited Speaker, University of Wisconsin: Genomics Seminar Series, Madison, WI, April
- Invited Speaker, SEBM Symposium Experimental Biology 2013: New Experimental Approaches to Human Brain Function in Health and Disease, Boston, MA, April
- Speaker & Inductee, National Academy of Sciences (NAS) 150th Annual Meeting, Washington, DC, April
- Invited Speaker, 2013 American Asthma Foundation (AAF) Annual Scientific Meeting, San Francisco, CA, May
- Invited Speaker, Center for Integrative Genomics (CIG) Symposium 2013: Genome, Disease and Evolution, Université de Lausanne, Lausanne, Switzerland, June
- Invited Speaker, 9th European Cytogenetics (ECA) Conference, Dublin, Ireland, June
- Invited Speaker, Gordon Research Conference: Human Genetics and Genomics, Smithfield, RI, July
- Invited Speaker, The Jackson Laboratory: 54th Annual Short Course on Medical and Experimental Mammalian Genetics, Bar Harbor, ME, July

- Invited Plenary, Human Genetics Society of Australasia (HGSA) 2013 Annual Scientific Meeting, Queenstown, New Zealand, August
- Invited Speaker & Session Co-chair, CSHL: Behavior & Neurogenetics of Nonhuman Primates, Cold Spring Harbor, NY, Sept.
- Invited Speaker, University of Liverpool Institute of Integrative Biology, Liverpool, UK, September
- Invited Plenary, British Society for Genetic Medicine/British Society of Human Genetics (BSGM/BSHG), Liverpool, UK, Sept.
- Invited Speaker, 2013 SFARI Annual Meeting (Simons Foundation), New York, NY, September
- Invited Speaker, Nijmegen Centre for Molecular Life Sciences, Nijmegen, The Netherlands, October
- Invited Speaker, Human Evolution Symposium, Swedish Society for Medical Genetics & Science, Uppsala, Sweden, October
- Invited Speaker, MIT's Simons Center for the Social Brain, Boston, MA, October
- Invited Session Speaker, 63rd ASHG Annual Meeting, Boston, MA, October
- Invited Lecture, Virginia Tech Carilion Research Institute (VTCRI): Distinguished Lecture Series, Roanoke, VA, November
- Invited Speaker, Simons Bioinformatics Symposium on Structural Variant Detection, New York, NY, November
- Invited Lecture, EMBL Distinguished Visitor Lecture, Heidelberg, Germany, December
- Invited Lecture, McGill University Distinguished Lectures in Human Genetics, Montreal, Canada, December
- Invited Speaker, Pacific Biosciences of California, Inc., Menlo Park, CA, December

2014

- Invited Speaker, Pacific Symposium on Biocomputing (PSB), Kona, HI, January
- Invited Faculty, Workshop on Comparative Genomics, Český Krumlov, Czech Republic, January
- Invited Public Keynote, New York University (NYU) Abu Dhabi Institute, Abu Dhabi, United Arab Emirates, February
- Invited Speaker, British Society for Cell Biology (BSCB) & British Society for Developmental Biology (BSDB) Joint Spring Meeting, University of Warwick, UK, March
- Invited Lecture, İhsan Doğramacı Lectureship, Bilkent University, Ankara, Turkey, April
- Invited Keynote, 9th International Meeting on CNVs & Genes in Intellectual Disability & Autism, Troina, Italy, April
- Invited Lecture and Grand Rounds, 8th Irene Uchida Lecture, University of Manitoba, Winnipeg, MB, Canada, April
- Invited Lecture, Penn State University Genomix Club, State College, PA, April
- Invited Lecture, Stanley Institute Lecture Series, Cold Spring Harbor, NY, April
- Invited Speaker, California Life Company (Calico), South San Francisco, CA, May
- Invited Speaker, Science+Fiction Dialogue 2014 "Brave New World", University of Basel, Switzerland, May
- Invited Plenary, I-CORE Spring Meeting: Gene Regulation in Complex Human Disease, Tel Aviv University, Israel, June
- Breakout Group Organizer and Speaker, Future Opportunities for Genome Sequencing and Beyond: A Planning Workshop for the NHGRI, Bethesda, MD, July
- Invited Speaker, Renaissance in diagnosis of monogenic diseases, Mini-symposium, Frontiers in Medicine, Nobel Forum, Karolinska Institutet, Stockholm, Sweden, September
- Invited Speaker, Kunming University of Science and Technology (KUST), Kunming, China, September
- Invited Speaker, Kunming Institute of Botany (KIB), Kunming, China, September
- Invited Speaker, Tufts University, School of Medicine, Boston, MA, October
- Invited Lecture, New York Genome Center (NYGC) Evening Lecture Series, New York City, NY, October
- Invited Seminar, University of Maryland Computational Biology, Bioinformatics, and Genomics (CBBG), College Park, MD, November
- Invited Speaker, Association for Molecular Pathology (AMP), Washington, DC, November
- Invited Seminar, Iowa Institute of Human Genetics, University of Iowa, Iowa City, IA, November
- Invited Speaker, Allen Institute for Brain Science Seminar Series, Seattle, WA, December
- Invited Lecture, UT Southwestern Medical Center Lecture Series, Dallas, TX, December

2015

- Invited Lecture, UCLA Department of Human Genetics, Los Angeles, CA, January
- Invited Speaker, Revolutionizing Next-Generation Sequencing: Tools And Technologies, Leuven, Belgium, January
- Invited Faculty, Workshop on Comparative Genomics, Český Krumlov, Czech Republic, January
- Invited Speaker, Allen Distinguished Investigator Life Science Symposium, San Diego, CA, February
- Invited Lecturer, NIH Wednesday Afternoon Lecture Series (WALS), Bethesda, MD, February
- Invited Speaker, University of Toledo Seminar Series, Toledo, OH, February
- Invited Plenary Session Speaker, Advances in Genome Biology and Technology (AGBT), 16th Annual Meeting, Marco Island, FL, February
- Invited Keynote, Human Genome Meeting (HGM) 2015, Kuala Lumpur, Malaysia, March
- Invited Speaker, American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting, Salt Lake City, UT, March

- Invited Lecture, Reed College Biology Department Seminar Series, Portland, OR, April
- Invited Speaker, 2015 SFARI Science Meeting (Simons Foundation), New York, NY, April
- Invited Speaker, 10th International Meeting on CNVs & Genes in Intellectual Disability & Autism, Troina, Italy, April
- Invited Speaker, iBG-izmir Genome Conference, Izmir, Turkey, April
- Invited Speaker, Kunming University of Science and Technology (KUST), Kunming, China, May
- Invited Seminar, Intellectual and Developmental Disabilities Research Center (IDDR) Seminar Series at The Children’s Hospital of Philadelphia (CHOP) and University of Pennsylvania, Philadelphia, PA, May
- Invited Seminar, Case Western Reserve University Department of Genetics Wilson Symposium, Cleveland, OH, May
- Invited Keynote, Natureconference “Genome Variation in Precision Medicine 2015”, Changsha, Hunan, China, May
- Invited Speaker, 11th International Workshop on Advanced Genomics (11AGW), Tokyo, Japan, May
- Invited Seminar, UCLA Integrative Center for Neurogenetics Seminar, Los Angeles, CA, May
- Invited Speaker, Future Perspectives in Computational Pan-Genomics, Leiden, The Netherlands, June
- Invited Keynote, Max Planck Institute (MPI) for Psycholinguistics Opening, Nijmegen, The Netherlands, June
- Invited Seminar, Radboud university medical center, Department of Human Genetics, Nijmegen, The Netherlands, June
- Invited Seminar, Gordon Research Conference Molecular Mechanisms in Evolution, Stonehill College, Easton, MA, June
- Invited Keynote, 15th Congress of the European Society for Evolutionary Biology (ESEB), Lausanne, Switzerland, August
- Invited Keynote, Long reads, single cells and cream teas, University of Exeter, UK, September
- Invited Seminar, Genomics of Common Diseases, Wellcome Trust Genome Campus, Hinxton, UK, September
- Invited Keynote, EMBO | EMBL Symposium: The Mobile Genome: Genetic and Physiological Impacts of Transposable Elements, Heidelberg, Germany, September
- Invited Lectureship, Barton Childs – Predoctoral Training Program in Human Genetics at Johns Hopkins University, Baltimore, MD, October
- Invited Speaker, Kunming University of Science and Technology (KUST), Kunming, China, November

2016

- Invited Faculty, Workshop on Genomics Application, Český Krumlov, Czech Republic, January
- Invited Plenary, Genetics & Society, Belgian Society of Human Genetics (BeSHG) & The Netherlands Society of Human Genetics (NVHG), Leuven, Belgium, February
- Invited Speaker, Centre for Human Genetics 50th Anniversary Celebration, University of Leuven, Leuven, Belgium, February
- Invited Speaker, New York University (NYU), New York, NY, March
- Invited Speaker, Rockefeller University: Crick Symposium, New York, NY, March
- Invited Speaker, Mount Sinai School of Medicine, New York, NY, March
- Invited Keynote, Xi’an Jiaotong University, Xi’an, China, April
- Invited Plenary, 8th European Conference on Comparative Neurobiology, Munich, Germany, April
- Invited Speaker, 2016 SFARI Science Meeting (Simons Foundation), New York, NY, April
- Invited Speaker, Oregon National Primate Research Center (ONPRC) Inaugural Primate Genetics Symposium, Beaverton, OR, April
- Invited Educational Session & Concurrent Symposia Speaker, European Human Genetics Conference (ESHG), Barcelona, Spain, May
- Invited Keynote, Keystone Symposia on Molecular & Cellular Biology: Understanding the Function of Human Genome Variation, Uppsala, Sweden, June
- Invited Keynote, Leiden University Medical Center: European SMRT Informatics Developers Meeting, Leiden, Netherlands, June
- Invited Plenary Speaker (videoconference), 3rd International Conference on Algorithms for Computational Biology, AICoB 2016, Trujillo, Spain, June
- Invited Speaker, PreventionGenetics, Marshfield, WI, June
- Invited Speaker, 7th International Symposium on Primate Research, Kunming, China, August
- Invited Speaker, International Colloquium “Perspectives in Genomics” 2016, Cancún, Mexico, October
- Invited Speaker, American Neurological Association (ANA) Meeting, Baltimore, MD, October
- Invited Scientific Session, Forbeck Forum on Chromosomal Instability/Aneuploidy, Hilton Head, SC, November
- Invited Seminar Speaker, The University of North Carolina (UNC) at Charlotte Department of Bioinformatics and Genomics, Charlotte, NC, November
- Invited Seminar Speaker, Telethon Institute of Genetics and Medicine (TIGEM), Naples, Italy, November
- Invited Speaker, XIX Italian Society of Human Genetics (SIGU) National Congress, Torino, Italy, November

2017

- Invited Faculty, Workshop on Genomics Application, Český Krumlov, Czech Republic, January

- Invited Seminar Speaker, Department of Genetics, Development and Cell Biology, Iowa State University, Ames, Iowa, February
- Invited Session Speaker, 38th Annual Lorne Genome Conference, Lorne, Australia, February
- Invited Speaker, Murdoch Children's Research Institute, Melbourne, Australia, February
- Invited Speaker, 2017 SFARI Science Meeting (Simons Foundation), New York, NY, April
- Invited Seminar Speaker, University of California, San Francisco Biomedical Sciences (BMS), San Francisco, CA, April
- Invited Seminar Speaker, University of Virginia Genome Sciences, Charlottesville, VA, April
- Invited Speaker, Advances in Precision Medicine: Genetics, Columbia University, New York, NY, April

RESEARCH GRANTS

a) Active

National Institutes of Health (R01 HG002385-13)

Title: Sequence and Assembly of Segmental Duplications

Goal: To provide a systematic approach for closing gaps within pericentromeric regions of human chromosomes.

PI: Evan E. Eichler

2007–2018

National Institutes of Health/ National Institutes of Mental Health (R01 MH101221)

Title: Sporadic Mutations and Autism Spectrum Disorders

Goal: To perform exome sequencing of 225 SSC autism quads to discover pathogenic SNPs and CNVs associated with disease and further validate these loci using targeted resequencing in 2000 probands.

PI: Evan E. Eichler

2013–2020

National Institutes of Health (1U01NS077275-01)

Title: 7 of 7 Epi4K: Copy Number Variants Project

Goal: Discover and genotype CNVs from genome and exome sequence data from patients with epilepsy.

PI: Evan E. Eichler/Heather C. Mefford

2011–2017

Simons Foundation (RFA 303241)

Title: Simons Autism Gene Characterization

Goal: Define high-impact genes and mutations associated with sporadic autism.

PI: Evan E. Eichler

2014–2017

Simons Foundation (RFA 385035)

Title: Structural Variation and the Genetic Architecture of Autism

Goal: characterize structural variation associated with sporadic autism in 500 autism quad families.

PI: Evan E. Eichler

2015–2017

National Human Genome Research Institute (NHGRI) (1U24HG009081)

Title: High Quality Human and Non-human Primate Genome Assemblies

Goal: Generate high quality reference genomes that better represent the complexity of human diversity and significantly improve the quality of index non-human primate (NHP) genomes, reaching a quality level more in line with the current human genome (GRCh38).

PI: Susan Dutcher, Evan Eichler

2016–2019

National Human Genome Research Institute (NHGRI) (1U54HG006493)

Title: UW Center for Mendelian Genomics

Goal: Establish the UW Center for Mendelian Genomics (UW-CMG) to apply exome sequencing and analysis to discover the candidate genes and sequence variants underlying rare Mendelian disorders and other human health-related Mendelian phenotypes.

PI: Debbie Nickerson; Michael Bamshad; Jay Shendure

2011–2019

National Institutes of Health (1U41HG007497)

Title: An Integrative Analysis of Structural Variation for the 1000 Genomes Project

Goal: Provide accurate methods for detection of structural variations, providing a comprehensive list of MEI and inversion events of the samples sequenced by the 1000 Genomes Project.

PI: Charles Lee

2013–2017

National Institutes of Health (5U41HG007635)

Title: Improving the Human Reference Genome Resource

Goal: Identify and resolve misassemblies in the current human reference genome, to generate alternate reference assemblies for structurally complex regions, and to generate community resources (both genomic and software) to improve assemblies.

PI: Rick Wilson

2014–2017

b) Previous

The Paul G. Allen Family Foundation (11631)

Title: Genetic Mutation of HARs and Human Neurocognition

Goal: To establish a genetic link between disruptive mutation of human accelerated regions (HARs) and specific neurodevelopmental phenotypes, restricting functional characterization to those with phenotypic effect.

PI: Evan E. Eichler

2013–2016

National Institutes of Health (1U01NS077303-01)

Title: 3 of 7 Epi4K: Sequencing, Biostatistics & Bioinformatics Core

Goal: Provide exome and genome sequence data as well as copy number variant data to investigators associated with the Epi4K consortium.

PI: David Goldstein

2011–2016

Simons Foundation (RFA 294112)

Title: Simons VIP: The genetic basis underlying the phenotypic variability of the 16p11.2 CNV

Goal: Investigate the genetics underlying the variability of disease associated with patients carrying the chromosome 16p11.2 deletion and duplication.

PI: Evan E. Eichler

2013–2015

National Institutes of Health (1R01MH092367)

Title: Next Generation Gene Discovery in Familial Autism

Goal: Perform massively parallel whole-exome sequencing and array comparative genomic hybridization to identify novel genes for familial autism.

PI: Zoran Brkanac

2011–2015

National Institutes of Health (1R24GM095471)

Title: Germline Sequence Resources and Analyses in a Vertebrate Model that Undergoes PGR

Goal: Provide genomics support for the assembly, analysis and characterization of the lamprey germline genome.

PI: Chris Amemiya

2011–2015

National Institutes of Health (P01 HG004120-06)

Title: Human Genome Structural Variation

Goal: Identify, sequence and genotype fine-scale structural variation.

PI: Evan E. Eichler

Co-PIs: Debbie Nickerson

2007–2014

Simons Foundation (RFA 191889EE)

Title: Whole Exome Sequencing of Simons Simplex Collection Quads

Goal: Perform exome sequencing of 225 SSC autism quads to discover pathogenic SNPs and CNVs associated with disease and further validate these loci using targeted resequencing in 2000 probands.

PI: Evan E. Eichler

2012–2014

National Institutes of Health (1U01MH100233)

Title: 1/4-The Autism Sequencing Consortium: Autism gene discovery in >20,000 exomes

Goal: Coordinate and benchmark copy number variant calling algorithms across a metanalysis of exome and genome sequencing projects.

PI: Joseph D. Buxbaum

2013–2014

National Institutes of Health (5R01NS069719)

Title: Next Generation Gene Discovery in Neurogenetics

Goal: Identify candidate genetic variants for neurogenetic disorders and to validate these variants/genes in families, across panels of subjects and/or by functional studies.

PI: Wendy Raskind

2010–2014

American Asthma Foundation (AAF) (10-0159)

Title: Comprehensive Analysis of the Effects of Copy Number Variation on Asthma

Goal: Assess the role of copy-number polymorphisms in contributing to asthma based on analyses of affected and unaffected individuals from the Hutterite population.

PI: Evan E. Eichler

2010–2013

Simons Foundation (SFARI 2009 RFA 137578)

Title: Genomic Hotspots of Autism

Goal: Examine ~1000 regions of the genome prone to recurrent rearrangements and assess their contribution to autism and related phenotypes in the Simons Simplex Collection of sporadic autists.

PI: Evan E. Eichler

2009–2013

NHLBI (1RC2 HL102926)

Title: Northwest Genomics Center

Goal: Apply next-generation exome sequencing to medically relevant DNA sample cohorts selected by NHLBI.

PI: Debbie Nickerson

2009–2012

National Institutes of Health (U01HG0052209)

Title: Structural Genomic Variation Analysis for the 1000 Genomes Project (1KG)

Goal: Develop computational methods to mine structural variation data from the 1KG. As part of the consortium, our lab specifically tested paired-end read approaches to detect insertions and deletions.

PI: Charles Lee

2009–2012

National Institutes of Health (5R01HL094976-02)

Title: SeattleSeq

Goal: Explore deep resequencing of human genes that can lead to the discovery of rare, nonsynonymous sequence variants that are robustly associated with complex human phenotypes.

PI: Debbie Nickerson

2008–2012

Simons Foundation (RFA 191889)

Title: Exome Sequencing of Simons Simplex Collection (SSC) Trios

Goal: Perform exome sequencing of 400 SSC autism trios in collaboration with Matt State at Yale University to discover pathogenic SNPs associated with disease.

PI: Evan E. Eichler

2010–2011

National Institutes of Health (R01 GM058815-13)

Title: Mechanism and Instability of Segmental Duplications (Competing Renewal)

Goal: Investigate evolution, mechanism and instability of low-copy repeats on chromosome 16.

PI: Evan E. Eichler
2007–2011

National Institutes of Health (1R01HD065285-02)

Title: Genomic Identification of Autism Loci

Goal: Explore the hypothesis that autism is caused by highly-penetrant, rare mutations using emerging technologies that screen regions for autism-specific copy-number variation (CNV) mutations and exonic point mutations.

PI: Evan E. Eichler
2009–2011

National Institutes of Health (3P01HG004120-03S1)

Title: Human Genome Structural Variation

Goal: Expand genotyping of structural variation to 2,000 genome samples being analyzed as part of the 1KG.

PI: Evan E. Eichler
2009–2010

National Institutes of Health (R01 HD043569-06)

Title: Segmental Aneusomy between Blocks of Duplicated DNA

Goal: Assess large-scale genomic rearrangements using microarray CGH in patients with idiopathic mental retardation.

PI: Evan E. Eichler
2003–2009

National Institutes of Health (R01 GM58815)

Title: Mechanism and Instability of Pericentromeric Duplications

Goal: Investigate molecular mechanism responsible for transposition of gene-containing segments to human chromosomes.

PI: Evan E. Eichler
1999–2007

National Institutes of Health (U54 HG02043)

Title: UW Genome Center Large-Scale Sequencing Program

Goal: Develop production sequencing capacity and systematic computational/experimental methodology to target problematic euchromatic regions of the human genome.

PI: Maynard Olson (UW)
Co-PI: Evan E. Eichler
2003–2006

National Institutes of Health (R01 ES10631)

Title: Genetic and Environmental Factors in Deletion Disorders

Goal: Examine the molecular mechanisms underlying rearrangement associated with Prader-Willi and Angelman syndromes.

PI: Robert Nicholls (University of Pennsylvania)
Co-PI: Evan E. Eichler
2001–2006

Department of Energy (R01 ER62862)

Title: Sequence-Ready Characterization of the Pericentromeric Region of 19p12

Goal: Develop and implement a sequence-anchor strategy to generate a contiguous BAC/cosmid map of the most proximal portion of 19p12.

PI: Evan E. Eichler
1999–2003

March of Dimes Birth Defects Foundation (FY99-0120)

Title: Chromosome Duplication and Instability

Goal: Characterize the pericentromeric region of 15q11-q13 and its involvement in supernumerary marker chromosome formation.

PI: Evan E. Eichler
1999–2001

National Science Foundation (DEB 9806913)

Title: Molecular Evolution of Pericentromeric Duplications among Higher Primates

Goal: Investigate phylogenetic history of pericentromeric DNA by comparative analysis.

PI: Evan E. Eichler
1998–1999 (converted in second year to NIH grant)

National Institutes of Health (R01 HG01847)

Title: Human Genomic Sequence Variation: X Chromosome
Goal: Examine the nature and frequency of sequence variation of the X chromosome in a population of humans and primates.
PI: Aravinda Chakravarti (Johns Hopkins)
Co-PI: Evan E. Eichler
1998–2001

National Institutes of Health (R01 HG01955)

Title: Human Genomic Polymorphisms.
Goal: SNP discovery and genotype frequency within 4 Mb of genomic DNA.
PI: Aravinda Chakravarti (Johns Hopkins)
Co-PI: Evan E. Eichler
1998–2001

Charles B. Wang Foundation

Title: Center for Computational Genomics
Goal: Develop computational infrastructure for high-throughput genomic analysis at CWRU.
PIs: Joseph Nadeau and Yoh-Han Pao
Co-PIs: Evan E. Eichler, John Witte, Cenk Sahinalp, Sunil Rao
2001–2004

Oklahoma Foundation

Title: The Evolution of New Genes and Gene Families within the Human Genome.
Goal: Develop a phylogenomic approach to recover rapidly evolving gene families in a panel of primate species.
PI: Evan E. Eichler
2003–2004

Ohio Board of Regents (PRI, CWRUID)

Title: Computational Tools
Goal: Develop computational algorithms for large-scale multiple sequence alignment.
PI: Cenk Sahinalp (Electrical Engineering and Computer Science)
Co-PI: Evan E. Eichler
2001–2003

PUBLICATIONS

(364 peer-reviewed publications: 172 first or corresponding senior-author publications – designated with *)

a) Research Articles

Verkerk AJ, de Graaff E, De Boulle K, **Eichler EE**, Konecki DS, Reyniers E, Manca A, Poustka A, Willems PJ, Nelson DL, Oostra BA. (1993). Alternative splicing in the fragile X gene FMR1. *Hum Mol Genet* Apr;2(4):399–404.

Ashley CT, Sutcliffe JS, Kunst CB, Leiner HA, **Eichler EE**, Nelson DL, Warren ST. (1993). Human and murine FMR-1: Alternative splicing and translational initiation downstream of the CGG-repeat. *Nat Genet* Jul;4(3):244–251.

***Eichler EE**, Richards S, Gibbs RA, Nelson DL. (1993). Fine structure of the human FMR1 gene. *Hum Mol Genet* Aug;2(8):1147–1153.

Chong SS, **Eichler EE**, Nelson DL, Hughes MR. (1994). Robust amplification and ethidium-visible detection of the fragile X syndrome CGG repeat using Pfu polymerase. *Am J Med Genet* Jul 15;51(4):522–526.

***Eichler EE**, Holden JJA, Popovich BW, Reiss AL, Snow K, Thibodeau SN, Richards CS, Ward PA, Nelson DL. (1994). Length of uninterrupted CGG repeats determines instability in the FMR1 gene. *Nat Genet* Sep;8(1):88–94.

***Eichler EE**, Kunst CB, Lugenbeel KA, Ryder OA, Davison D, Warren ST, Nelson DL. (1995). Evolution of the cryptic FMR1 CGG repeat. *Nat Genet* Nov;11(3):301–308.

***Eichler EE**, Hammond HA, Macpherson JN, Ward PA, Nelson DL. (1995). Population survey of the human FMR1 CGG repeat substructure suggests biased polarity for the loss of AGG interruptions. *Hum Mol Genet* Dec;4(12):2199–2208.

- Chastain PD, **Eichler EE**, Kang S, Nelson DL, Levene SD, Sinden RR. (1995). Anomalous rapid electrophoretic mobility of DNA containing triplet repeats associated with human disease genes. *Biochem Dec* 12;34(49):16125–16131.
- Kunst CB, Zerylnick C, Karickhoff L, **Eichler EE**, Bullard J, Chalifoux M, Holden JJ, Nelson DL, Warren ST. (1996). FMR1 in global populations. *Am J Hum Genet Mar*;58(3):513–522.
- ***Eichler EE**, Macpherson JN, Murray A, Jacobs PA, Chakravarti A, Nelson DL. (1996). Haplotype and interspersed analysis of the FMR1 CGG repeat identifies two different mutational pathways for the origin of the fragile X syndrome. *Hum Mol Genet Mar*;5(3):319–330.
- ***Eichler EE**, Lu F, Shen Y, Antonacci R, Jurecic V, Doggett NA, Moyzis RK, Baldini A, Gibbs RA, Nelson DL. (1996). Duplication of a gene-rich cluster between 16p11.1 and Xq28: a novel pericentromeric-directed mechanism for paralogous genome evolution. *Hum Mol Genet Jul*;5(7):899–912.
- ***Eichler EE**, Nelson DL. (1996). Genetic variation and evolutionary stability of the FMR1 CGG repeat in six closed human populations. *Am J Med Genet Jul* 12;64(1):220–225.
- *Falik-Zaccai TC, Shachak E, Yalon M, Lis Z, Borochowitz Z, Macpherson JN, Nelson DL, **Eichler EE**. (1997). Predisposition to the fragile X syndrome in Jews of Tunisian descent is due to the absence of AGG interruptions on a rare Mediterranean haplotype. *Am J Hum Genet Jan*;60(1):103–112.
- ***Eichler EE**, Budarf ML, Rocchi M, Deaven LL, Doggett NA, Baldini A, Nelson DL, Mohrenweiser HW. (1997). Interchromosomal duplications of the adrenoleukodystrophy locus: A phenomenon of pericentromeric plasticity. *Hum Mol Genet Jul*;6(7):991–1002.
- Wagtmann N, Rojo S, **Eichler EE**, Mohrenweiser H, Long EO. (1997). A new human gene complex the killer cell inhibitory receptors and related monocyte/macrophage receptors. *Curr Biol Aug*;7(8):615–618.
- Fan W, Christensen M, **Eichler EE**, Zhang X, Lennon G. (1997). Cloning, sequencing, gene organization, and localization of the human ribosomal protein RPL23A. *Genomics Dec*;46(2):234–239.
- Pearson CE, **Eichler EE**, Lorenzetti D, Kramer SF, Zoghbi HY, Nelson DL, Sinden RR. (1998). Interruptions in the triplet repeats of SCA1 and FRAXA reduce the propensity and complexity of slipped strand DNA (S-DNA) formation. *Biochem Feb*;37(8):2701–2708.
- ***Eichler EE**, Hoffman SM, Adamson AA, Gordon LA, McCreedy P, Lamerdin JE, Mohrenweiser HW. (1998). Complex beta-satellite repeat structures and the expansion of the zinc-finger gene cluster in 19p12. *Genome Res Aug*;8(8):791–808.
- Her C, Wood TC, **Eichler EE**, Mohrenweiser HW, Ramagli LS, Siciliano MJ, Weinshilboum RM. (1998). Human hydroxysteroid sulfotransferase SULT2B1: Two enzymes encoded by a single chromosome 19 gene. *Genomics Nov*;53(3):284–295.
- Trask BJ, Massa H, Brand-Arpon V, Chan K, Friedman C, Nguyen OT, **Eichler EE**, van den Engh G, Rouquier S, Shizuya H, Giorgi D. (1998). Large multi-chromosomal duplications encompass many members of the olfactory receptor gene family in the human genome. *Hum Mol Genet Dec*;7(13):2007–2020.
- Loftus BJ, Kim UJ, Sneddon VP, Kalush F, Brandon R, Fuhrmann J, Mason T, Crosby ML, Barnstead M, Cronin L, Deslattes Mays A, Cao Y, Xu RX, Kang HL, Mitchell S, **Eichler EE**, Harris PC, Venter JC, Adams MD. (1999). Genome duplications and other features in 12 Mbp of DNA sequence from human chromosome 16p and 16q. *Genomics Sep*;60(3):295–308.
- ***Eichler EE**, Archidiacono N, Rocchi M. (1999). CAGGG repeats and the pericentromeric duplication of the hominoid genome. *Genome Res Nov*;9(11):1048–1058.
- *Horvath JE, Viggiano L, Loftus BJ, Adams MD, Archidiacono N, Rocchi M, **Eichler EE**. (2000). Molecular structure and evolution of an alpha satellite/non-alpha satellite junction at 16p11. *Hum Mol Genet Jan*;9(1):113–123.
- *Horvath JE, Schwartz S, **Eichler EE**. (2000). The mosaic structure of human pericentromeric DNA: A strategy for characterizing complex regions of the human genome. *Genome Res Jun*;10(6):839–852.
- *Bailey JA, Carrel L, Chakravarti A, **Eichler EE**. (2000). Molecular evidence for a relationship between LINE-1 elements and X chromosome inactivation: The Lyon repeat hypothesis. *Proc Natl Acad Sci U S A Jun*;97(12):6634–6639.
- Cheung VG, Nowak N, Jang W, Kirsch IR, Zhao S, Chen XN, Furey TS, Kim UJ, Kuo WL, Olivier M, Conroy J, Kasprzyk A, Massa H, Yonescu R, Sait S, Thoreen C, Snijders A, Lemyre E, Bailey JA, Bruzel A, Burrill WD, Clegg SM, Collins S, Dhami P, Friedman C, Han CS, Herrick S, Lee J, Ligon AH, Lowry S, Morley M, Narasimhan S, Osoegawa K, Peng Z, Plajzer-Frick I, Quade BJ, Scott D, Sirotkin K, Thorpe AA, Gray JW, Hudson J, Pinkel D, Ried T, Rowen L, Shen-Ong GL, Strausberg RL, Birney E, Callen DF, Cheng JF, Cox DR, Doggett NA, Carter NP, **Eichler EE**, Haussler D, Korenberg JR, Morton CC, Albertson D, Schuler G, de Jong PJ, Trask BJ. (2001). Integration of cytogenetic landmarks in the draft sequence of the human genome. *Nature Feb* 15;409(6822):953–958.

- *Bailey JA, Yavor AM, Massa HF, Trask BJ, **Eichler EE**. (2001). Segmental duplications: Organization and impact within the current human genome project assembly. *Genome Res* Jun;11(6):1005–1017.
- Mathews DJ, Kashuk C, Brightwell G, **Eichler EE**, Chakravarti A. (2001). Sequence variation within the fragile X locus. *Genome Res* Aug;11(8):1382–1391.
- *Johnson ME, Viggiano L, Bailey JA, Abdul-Rauf M, Goodwin G, Rocchi M, **Eichler EE**. (2001). Positive selection of a gene family during the emergence of humans and African apes. *Nature* Oct;413(6855):514–519.
- Cutler DJ, Zwick ME, Carrasquillo MM, Yohn CT, Tobin KP, Kashuk C, Mathews DJ, Shah NA, **Eichler EE**, Warrington JA, Chakravarti A. (2001). High-throughput variation detection and genotyping using microarrays. *Genome Res* Nov;11(11):1913–1925.
- *Bailey JA, Yavor AM, Viggiano L, Misceo D, Horvath JE, Archidiacono N, Schwartz S, Rocchi M, **Eichler EE**. (2002). Human-specific duplication and mosaic transcripts: The recent paralogous structure of chromosome 22. *Am J Hum Genet* Jan;70(1):83–100.
- van Geel M, **Eichler EE**, Beck AF, Shan Z, Haaf T, van der Maarell SM, Frants RR, de Jong PJ. (2002). A cascade of complex subtelomeric duplications during the evolution of the hominoid and Old World monkey genomes. *Am J Hum Genet* Jan;70(1):269–278.
- Kashuk C, SenGupta S, **Eichler EE**, Chakravarti A. (2002). ViewGene: A graphical tool for polymorphism visualization and characterization. *Genome Res* Feb;12(2):333–338.
- *Bailey JA, Gu Z, Clark RA, Reinert K, Samonte RV, Schwartz S, Adams MD, Myers EW, Li PW, **Eichler EE**. (2002). Recent segmental duplications in the human genome. *Science* Aug;297(5583):1003–1007.
- ***Eichler EE**, Johnson ME, Alkan C, Tüzün E, Sahinalp C, Misceo D, Archidiacono N, Rocchi M. (2002). Divergent origins and concerted expansion of two segmental duplications on chromosome 16. *J Hered* Nov–Dec;92(6):468–472.
- Alkan C, Bailey JA, **Eichler EE**, Sahinalp CS, Tüzün E. (2002). An algorithmic analysis of the role of unequal crossover in alpha-satellite DNA evolution. *Genome Inform* 13:93–102.
- Guy J, Hearn T, Crosier M, Mudge J, Viggiano L, Koczan D, Thiesen HJ, Bailey JA, Horvath JE, **Eichler EE**, Earthrowl ME, Deloukas P, French L, Rogers J, Bentley D, Jackson MS. (2003). Genomic sequence and transcriptional profile of the boundary between pericentromeric satellites and genes on human chromosome arm 10p. *Genome Res* Feb;13(2):159–172.
- *Liu G, NISC Comparative Sequencing Program, Zhao S, Bailey JA, Sahinalp SC, Alkan C, Tüzün E, Green ED, **Eichler EE**. (2003). Analysis of primate genomic variation reveals a repeat-driven expansion of the human genome. *Genome Res* Mar;13(3):358–368.
- *Locke DP, Segraves R, Carbone L, Archidiacono N, Albertson DG, Pinkel D, **Eichler EE**. (2003). Large-scale variation among human and great ape genomes determined by array comparative genomic hybridization. *Genome Res* Mar;13(3):347–357.
- *Locke DP, Archidiacono N, Misceo D, Cardone MF, Deschamps S, Roe B, Rocchi M, **Eichler EE**. (2003). Refinement of a chimpanzee pericentric inversion breakpoint to a segmental duplication cluster. *Genome Biol* Jul;4(8):R50.
- *Horvath JE, Gulden CL, Bailey JA, Yohn C, McPherson JD, Prescott A, Roe BA, De Jong PJ, Ventura M, Misceo D, Archidiacono N, Zhao S, Schwartz S, Rocchi M, **Eichler EE**. (2003). Using a pericentromeric interspersed repeat to recapitulate the phylogeny and expansion of human centromeric segmental duplications. *Mol Biol Evol* Sep;20(9):1463–1479.
- Chai JH, Locke DP, Grealley JM, Knoll JH, Ohta T, Dunai J, Yavor A, **Eichler EE**, Nicholls RD. (2003). Identification of four highly conserved genes between breakpoint hotspots BP1 and BP2 of the Prader-Willi/Angelman syndromes deletion region that have undergone evolutionary transposition mediated by flanking duplicons (2003). *Am J Hum Genet* Oct;73(4):898–925.
- *Bailey JA, Liu G, **Eichler EE**. (2003). An Alu transposition model for the origin and expansion of human segmental duplications. *Am J Hum Genet* Oct;73(4):823–834.
- *Locke DP, Segraves R, Nicholls RD, Schwartz S, Pinkel D, Albertson DG, **Eichler EE**. (2004). BAC microarray analysis of 15q11-q13 rearrangements and the impact of segmental duplications. *J Med Genet* Mar;41(3):175–182.
- *Bailey JA, Baertsch R, Kent WJ, Haussler D, **Eichler EE**. (2004). Hotspots of mammalian chromosomal evolution. *Genome Biol* Mar;5(4):R23.
- Astbury C, Christ LA, Aughton DJ, Cassidy SB, Kumar A, **Eichler EE**, Schwartz S. (2004). Detection of deletions in de novo "balanced" chromosome rearrangements: Further evidence for their role in phenotypic abnormalities. *Genet Med* Mar–Apr;6(2):81–89.
- *Tüzün E, Bailey JA, **Eichler EE**. (2004). Recent segmental duplications in the working draft assembly of the Brown Norway Rat. *Genome Res* Apr;14(4):493–506.

- *Bailey JA, Church DM, Ventura M, Rocchi M, **Eichler EE**. (2004). Analysis of segmental duplications and genome assembly in the mouse. *Genome Res* May;14(5):789–801.
- Chen DC, Saarela J, Clark RA, Miettinen T, Chi A, **Eichler EE**, Peltonen L, Palotie A. (2004). Segmental duplications flank the multiple sclerosis locus on chromosome 17q. *Genome Res* Aug;14(8):1483–1492.
- Fredman D, White SJ, Potter S, **Eichler EE**, Den Dunnen JT, Brookes AJ. (2004). Complex SNP-related sequence variation in segmental genome duplications. *Nat Genet* Aug;36(8):861–866.
- Khaitovich P, Muetzel B, She X, Lachmann M, Hellmann I, Dietzsch J, Steigele S, Do HH, Weiss G, Enard W, Heissig F, Arendt T, Nieselt-Struwe K, **Eichler EE**, Pääbo S. (2004). Regional patterns of gene expression in human and chimpanzee brains. *Genome Res* Aug;14(8):1462–1473.
- *She X, Horvath JE, Jiang Z, Liu G, Furey TS, Christ L, Clark R, Graves T, Gulden CL, Alkan C, Bailey JA, Sahinalp C, Rocchi M, Haussler D, Wilson RK, Miller W, Schwartz S, **Eichler EE**. (2004). The structure and evolution of centromeric transition regions within the human genome. *Nature* Aug;430(7002):857–864.
- Ventura M, Weigl S, Carbone L, Cardone MF, Misceo D, Teti M, D’adabbo P, Wandall A, Bjoerck E, de Jong P, She X, **Eichler EE**, Archidiacono N, Rocchi M. (2004). Recurrent sites for new centromere seeding. *Genome Res* Sep;14(9):1696–1703.
- *She X, Jiang Z, Clark RA, Liu G, Cheng Z, Tüzün E, Church DM, Sutton G, Halpern AL, **Eichler EE**. (2004). Shotgun sequence assembly and recent segmental duplications within the human genome. *Nature* Oct;431(7011):927–930.
- Sahinalp SC, **Eichler E**, Goldberg P, Berenbrink P, Friedetzky T, Ergun F. (2004). Identifying uniformly mutated segments within repeats. *J Bioinform Comput Biol* Dec;2(4):657–668.
- Alkan C, **Eichler EE**, Bailey JA, Sahinalp SC, Tüzün E. (2004). The role of unequal crossover in alpha-satellite DNA evolution: A computational analysis. *J Comput Biol* 11(5):933–944.
- Kirsch S, Weiss B, Miner TL, Waterston RH, Clark RA, **Eichler EE**, Münch C, Schempp W, Rappold G. (2005). Interchromosomal segmental duplications of the pericentromeric region on the human Y chromosome. *Genome Res* Feb;15(2):195–204.
- *Yohn CT, Jiang Z, McGrath SD, Hayden KE, Khaitovich P, Johnson ME, Eichler MY, McPherson JD, Zhao S, Pääbo S, **Eichler EE**. (2005). Lineage-specific expansions of retroviral insertions within the genomes of African great apes but not humans and orangutans. *PLOS Biol* Apr;3(4):e110.
- *Tüzün E, Sharp AJ, Bailey JA, Kaul R, Morrison VA, Pertz LM, Haugen E, Hayden H, Albertson D, Pinkel D, Olson MV, **Eichler EE**. (2005). Fine-scale structural variation of the human genome. *Nat Genet* Jul;37(7):727–732.
- Alkan C, Tüzün E, Buard J, Lethiec F, **Eichler EE**, Bailey JA, Sahinalp SC. (2005). Manipulating multiple sequence alignments via MaM and WebMaM. *Nucleic Acids Res* Jul 1;33(Web Server issue):W295–298.
- *Horvath JE, Gulden CL, Samonte RU, Eichler MY, Ventura M, McPherson JD, Graves TA, Wilson RK, Schwartz S, Rocchi M, **Eichler EE**. (2005). Punctuated duplication seeding events during the evolution of human chromosome 2p11. *Genome Res* Jul;15(7):914–927.
- *Sharp AJ, Locke DP, McGrath SD, Cheng Z, Bailey JA, Samonte RU, Pertz LM, Clark R, Schwartz S, Seagraves R, Oseroff VV, Albertson DG, Pinkel D, **Eichler EE**. (2005). Segmental duplications and copy-number variation in the human genome. *Am J Hum Genet* Jul;77(1):78–88.
- *Cheng Z, Ventura M, She X, Khaitovich P, Graves T, Osoegawa K, Church D, DeJong P, Wilson RK, Pääbo S, Rocchi M, **Eichler EE**. (2005). A genome-wide comparison of recent chimpanzee and human segmental duplications. *Nature* Sep;437(7055):88–93.
- *Newman TL, Tüzün E, Morrison VA, Hayden KE, Ventura M, McGrath SD, Rocchi M, **Eichler EE**. (2005). A genome-wide survey of structural variation between human and chimpanzee. *Genome Res* Oct;15(10):1344–1356.
- *Locke DP, Jiang Z, Pertz LM, Misceo D, Archidiacono N, **Eichler EE**. (2005). Molecular evolution of the human chromosome 15 pericentromeric region. *Cytogenet Genome Res* 108(1–3):73–82.
- *Newman TL, Rieder MJ, Morrison VA, Sharp AJ, Smith JD, Sprague LJ, Kaul R, Carlson CS, Olson MV, Nickerson DA, **Eichler EE**. (2006). High-throughput genotyping of intermediate-size structural variation. *Hum Mol Genet* Apr;15(7):1159–1167.
- Perry GH, Tchinda J, McGrath SD, Zhang J, Picker SR, Cáceres AM, Iafrate AJ, Tyler-Smith C, Scherer SW, **Eichler EE**, Stone AC, Lee C. (2006). Hotspots for copy number variation in chimpanzees and humans. *Proc Natl Acad Sci U S A* May;103(21):8006–8011.

- *She X, Liu G, Ventura M, Zhao S, Misceo D, Roberto R, Cardone MF, Rocchi M, NISC Comparative Sequencing Program, Green ED, Archidiacono N, **Eichler EE**. (2006). A preliminary comparative analysis of primate segmental duplications shows elevated substitution rates and a great-ape expansion of intrachromosomal duplications. *Genome Res* May;16(5):576–583.
- *Locke DP, Sharp AJ, McCarroll SA, McGrath SD, Newman TL, Cheng Z, Schwartz S, Albertson DG, Pinkel D, Altschuler DM, **Eichler EE**. (2006). Linkage disequilibrium and heritability of copy-number polymorphisms within duplicated regions of the human genome. *Am J Hum Genet* Aug;79(2):275–290.
- *Sharp AJ, Hansen S, Selzer RR, Cheng Z, Regan R, Hurst JA, Stewart H, Price SM, Blair E, Hennekam RC, Fitzpatrick CA, Segraves R, Richmond TA, Guiver C, Albertson DG, Pinkel D, Eis PS, Schwartz S, Knight SJ, **Eichler EE**. (2006). Discovery of previously unidentified genomic disorders from the duplication architecture of the human genome. *Nat Genet* Sep;38(9):1038–1042.
- *Johnson ME, NISC Comparative Sequencing Program, Cheng Z, Morrison AV, Scherer S, Ventura M, Gibbs RA, Green ED, **Eichler EE**. (2006). Recurrent duplication-driven transposition of DNA during hominoid evolution. *Proc Natl Acad Sci U S A* Nov;103(47):17626–17631.
- Cardone MF, Alonso A, Paziienza M, Ventura M, Montemurro G, Carbone L, de Jong PJ, Stanyon R, D'Addabbo P, Archidiacono N, She X, **Eichler EE**, Warburton PE, Rocchi M. (2006). Independent centromere formation in a capricious, gene-free domain of chromosome 13q21 in Old World monkeys and pigs. *Genome Biol* 7(10):R91.
- Wong KK, deLeeuw RJ, Dosanjh NS, Kimm LR, Cheng Z, Horsman DE, MacAulay C, Ng RT, Brown CJ, **Eichler EE**, Lam WL. (2007). A comprehensive analysis of common copy-number variations in the human genome. *Am J Hum Genet* Jan;80(1):91–104 (5 Dec 2006).
- *Roberto R, Capozzi O, Wilson RK, Mardis ER, Lomiento M, Tüzün E, Cheng Z, Mootnick AR, Archidiacono N, Rocchi M, **Eichler EE**. (2007). Molecular refinement of gibbon genome rearrangement. *Genome Res* Feb;17(2):249–257 (21 Dec 2006).
- *Sharp AJ, Selzer RR, Veltman JA, Gimelli S, Gimelli G, Striano P, Coppola A, Regan R, Price SM, Knoers NV, Eis PS, Brunner HG, Hennekam RC, Knight SJ, de Vries BB, Zuffardi O, **Eichler EE**. (2007). Characterization of a recurrent 15q24 microdeletion syndrome. *Hum Mol Genet* Mar;16(5):567–572.
- Ventura M, Antonacci F, Cardone MF, Stanyon R, D'Addabbo P, Cellamare A, Sprague LJ, **Eichler EE**, Archidiacono N, Rocchi M. (2007). Evolutionary formation of new centromeres in macaque. *Science* Apr;316(5822):243–246.
- *Kidd JM, Newman TL, Tüzün E, Kaul R, **Eichler EE**. (2007). Population stratification of a common APOBEC gene deletion polymorphism. *PLOS Genet* Apr 20;3(4):e63.
- *Alkan C, Ventura M, Archidiacono N, Rocchi M, Sahinalp CS, **Eichler EE**. (2007). Organization and evolution of primate centromeric DNA from whole-genome shotgun sequence data. *PLOS Comput Biol* Sep;3(9):e181 (28 Sept 2007).
- Lyle R, Prandini P, Osoegawa K, ten Hallers B, Humphray S, Zhu B, Eyraas E, Castelo R, Bird CP, Gagos S, Scott C, Cox A, Deutsch S, Ucla C, Cruts M, Dahoun S, She X, Bena F, Wang SY, Van Broeckhoven C, **Eichler EE**, Guigo R, Rogers J, de Jong PJ, Reymond A, Antonarakis SE. (2007). Islands of euchromatin-like sequence and expressed polymorphic sequences within the short arm of human chromosome 21. *Genome Res* Nov;17(11):1690–1696.
- *Sharp AJ, Itsara A, Cheng Z, Alkan C, Schwartz S, **Eichler EE**. (2007). Optimal design of oligonucleotide microarrays for measurement of DNA copy-number. *Hum Mol Genet* Nov;16(22):2770–2779 (28 Aug 2007).
- *Mefford HC, Clauin S, Sharp AJ, Moller RS, Ullmann R, Kapur R, Pinkel D, Cooper GM, Ventura M, Ropers HH, Tommerup N, **Eichler EE**, Bellanne-Chantelot C. (2007). Recurrent reciprocal genomic rearrangements of 17q12 are associated with renal disease, diabetes and epilepsy. *Am J Hum Genet* Nov;81(5):1057–1069 (26 Sept 2007).
- *Jiang Z, Tang H, Ventura M, Cardone MF, Marques-Bonet T, She X, Pevzner P, **Eichler EE**. (2007). Ancestral reconstruction of segmental duplications reveals punctuated cores of human genome evolution. *Nat Genet* Nov;39(11):1361–1368 (7 Oct 2007).
- Bovee D, Zhou Y, Haugen E, Wu Z, Hayden HS, Gillett W, Tüzün E, Cooper GM, Sampas N, Phelps K, Levy R, Morrison VA, Sprague J, Jewett D, Buckley D, Subramaniam S, Chang J, Smith DR, Olson MV, **Eichler EE**, Kaul R. (2008). Closing gaps in the human genome with fosmid resources generated from multiple individuals. *Nat Genet* Jan;40(1):96–101. PMID: 18157130. PMID: N/A.
- *Bailey JA, Kidd JM, **Eichler EE**. (2008). Human copy number polymorphic genes. *Cytogenet Genome Res* 123(1–4):234–243. PMID: PMC2920189.
- Cardone MF, Jiang Z, D'Addabbo P, Archidiacono N, Rocchi M, **Eichler EE**, Ventura M. (2008). Hominoid chromosomal rearrangements on 17q map to complex regions of segmental duplication. *Genome Biol* Feb 7;9(2):R28. PMID: PMC2374708.
- *Sharp AJ, Mefford HC, Li K, Baker C, Skinner C, Stevenson RE, Schroer RJ, Novara F, De Gregori M, Ciccone R, Broomer A, Casuga I, Wang Y, Xiao C, Barbacioru C, Gimelli G, Bernardina BD, Torniero C, Giorda R, Regan R, Murday V, Mansour S, Fichera M, Castiglia L, Failla P, Ventura M, Jiang Z, Cooper GM, Knight SJ, Romano C, Zuffardi O, Chen C, Schwartz CE,

- Eichler EE.** (2008). A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. *Nat Genet* Mar;40(3):322–328. PMID: PMC2365467.
- Walsh T, McClellan JM, McCarthy SE, Addington AM, Pierce SB, Cooper GM, Nord AS, Kusenda M, Malhotra D, Bhandari A, Stray SM, Rippey CF, Roccanova P, Makarov V, Lakshmi B, Findling RL, Sikich L, Stromberg T, Merriman B, Gogtay N, Butler P, Eckstrand K, Noory L, Gochman P, Long R, Chen Z, Davis S, Baker C, **Eichler EE**, Meltzer PS, Nelson SF, Singleton AB, Lee MK, Rapoport JL, King MC, Sebat J. (2008). Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia. *Science* Apr 25;320(5875):539–543. PMID: 18369103. PMID: N/A.
- Kirsch S, Munch C, Jiang Z, Cheng Z, Chen L, Batz C, **Eichler EE**, Schempp W. (2008). Evolutionary dynamics of segmental duplications from human Y-chromosomal euchromatin/heterochromatin transition regions. *Genome Res* June;18(6):1030–1042. PMID: PMC2493392.
- *Kidd JM, Cooper GM, Donahue WF, Hayden HS, Sampas N, Graves T, Hansen N, ... (31 authors) ..., Nickerson DA, Mullikin JC, Wilson RK, Bruhn L, Olson MV, Kaul R, Smith DR, **Eichler EE.** (2008). Mapping and sequencing of structural variation from eight human genomes. *Nature* May 1;453(7191):56–64. PMID: PMC2424287.
- *She X, Cheng Z, Zöllner S, Church DM, **Eichler EE.** (2008). Mouse segmental duplication and copy number variation. *Nat Genet* Jul;40(7):909–14. PMID: PMC2574762.
- *Jiang Z, Hubley R, Smit A, **Eichler EE.** (2008). DupMasker: A tool for annotating primate segmental duplications. *Genome Res* Aug;18(8):1362–1368. PMID: PMC2493431.
- Martin J, Knight SJ, Sharp AJ, **Eichler EE**, Hurst J, Kini U. (2008). Potocki-Lupski syndrome mimicking a connective tissue disorder. *Clin Dysmorphol* Jul;17(3):211–213. PMID: 18541972. PMID: N/A.
- Koolen DA, Sharp AJ, Hurst JA, Firth HV, Knight SJ, Goldenberg A, Saugier-Veber P, Pfundt R, Vissers LE, Destree A, Grisart B, Rooms L, Van der Aa N, Field M, Hackett A, Bell K, Nowaczyk MJ, Mancini GM, Poddighe PJ, Schwartz CE, Rossi E, De Gregori M, Antonacci-Fulton LL, McLellan MD 2nd, Garrett JM, Wiechert MA, Miner TL, Crosby S, Ciccone R, Willatt L, Rauch A, Zenker M, Aradhya S, Manning MA, Strom TM, Wagenstaller J, Krepischi-Santos AC, Vianna-Morgante AM, Rosenberg C, Price SM, Stewart H, Shaw-Smith C, Brunner HG, Wilkie AO, Veltman JA, Zuffardi O, **Eichler EE**, de Vries BB. (2008). Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. *J Med Genet* Nov;45(11):710–720. PMID: PMC3071570.
- *Zody MC, Jiang Z, Fung HC, Antonacci F, Hillier LW, Cardone MF, Graves TA, Kidd JM, Cheng Z, Abouelleil A, Chen L, Wallis J, Glasscock J, Wilson RK, Reily AD, Duckworth J, Ventura M, Hardy J, Warren WC, **Eichler EE.** (2008). Evolutionary toggling of the MAPT 17q21.31 inversion region. *Nat Genet* Sep;40(9): 1076–1083. PMID: PMC2684794.
- Marques-Bonet T, Cheng Z, She X, **Eichler EE**, Navarro A. (2008). The genomic distribution of intraspecific and interspecific sequence divergence of human segmental duplications relative to human/chimpanzee chromosomal rearrangements. *BMC Genomics* Aug 12;9(1):384. PMID: PMC2542386.
- Perry GH, Yang F, Marques-Bonet T, Murphy C, Fitzgerald T, Lee AS, Hyland C, Stone AC, Hurles ME, Tyler-Smith C, **Eichler EE**, Carter NP, Lee C, Redon R. (2008). Copy number variation and evolution in humans and chimpanzees. *Genome Res* Nov;18(11):1698–1710. PMID: PMC2577862.
- Cooper GM, Zerr T, Kidd JM, **Eichler EE**, Nickerson DA. (2008). Systematic assessment of copy number variant detection via genome-wide SNP genotyping. *Nat Genet* Oct;40(10):1199–1203. PMID: PMC2759751.
- *Mefford HC, Sharp AJ, Baker C, Itsara A, Jiang Z, Buysse K, ... (71 authors) ..., Veltman JA, de Vries BB, Vermeesch JR, Barber JC, Willatt L, Tassabehji M, **Eichler EE.** (2008). Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *N Engl J Med* Oct 16;359(16):1685–1699. PMID: PMC2703742.
- *Kidd JM, Cheng Z, Graves T, Wilson R, **Eichler EE.** (2008). Haplotype sorting using human fosmid clone end-sequence pairs. *Genome Res* Dec;18(12):2016–2023. PMID: PMC2593576.
- Lomiento M, Jiang Z, D'Addabbo P, **Eichler EE**, Rocchi M. (2008). Evolutionary-new centromeres preferentially emerge within gene deserts. *Genome Biol* Dec 16;9(12):R173. PMID: PMC2646277.
- *Girirajan S, Chen L, Graves T, Marques T, Ventura M, Fronick C, Fulton L, Rocchi M, Fulton RS, Wilson RK, Mardis ER, **Eichler EE.** (2009). Sequencing human-gibbon breakpoints of synteny reveals mosaic new insertions at rearrangement sites. *Genome Res* Feb;19(2):178–190. Epub 2008 Nov 24. PMID: PMC2652201.
- Helbig I, Mefford HC, Sharp AJ, Guipponi M, Fichera M, Franke A, Muhle H, de Kovel C, Baker C, von Spiczak S, Kron KL, Steinich I, Kleefuss-Lie AA, Leu C, Gaus V, Schmitz B, Klein KM, Reif PS, Rosenow F, Weber Y, Lerche H, Zimprich F, Urak L, Fuchs K, Feucht M, Genton P, Thomas P, Visscher F, de Haan GJ, Møller RS, Hjalgrim H, Luciano D, Wittig M, Nothnagel M, Elger CE, Nürnberg P, Romano C, Malafosse A, Koeleman BP, Lindhout D, Stephani U, Schreiber S, **Eichler EE**, Sander T. (2009). 15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. *Nat Genet.* Feb;41(2):160–162. PMID: PMC3026630.

- *Nicholas TJ, Cheng Z, Ventura M, Mealey K, **Eichler EE**, Akey JM. (2009). The genomic architecture of segmental duplications and associated copy number variants in dogs. *Genome Res.* Mar;19(3):491–499. PMID: PMC2661811.
- Landsverk ML, Ruzzo EK, Mefford HC, Buysse K, Buchan JG, **Eichler EE**, Petty EM, Peterson EA, Knutzen DM, Barnett K, Farlow MR, Caress J, Parry GJ, Quan D, Gardner KL, Hong M, Simmons Z, Bird TD, Chance PF, Hannibal MC. (2009). Duplication within the SEPT9 gene associated with a founder effect in North American families with Hereditary Neuralgic Amyotrophy. *Hum Mol Genet.* Apr 1;18(7):1200–1208. PMID: PMC2722193.
- Hannes FD, Sharp AJ, Mefford HC, de Ravel T, Ruivenkamp CA, Breuning MH, Fryns JP, Devriendt K, Van Buggenhout G, Vogels A, Stewart HH, Hennekam RC, Cooper GM, Regan R, Knight SJ, **Eichler EE**, Vermeesch JR. (2009). Recurrent reciprocal deletions and duplications of 16p13.11: The deletion is a risk factor for MR/MCA while the duplication may be a rare benign variant. *J Med Genet* Apr;46(4):223–32. Epub 2008 Jun 11. PMID: PMC2658752.
- Degenhardt JD, de Candia P, Chabot A, Schwartz S, Henderson L, Ling B, Hunter M, Jiang Z, Palermo RE, Katze M, **Eichler EE**, Ventura M, Rogers J, Marx P, Gilad Y, Bustamante CD. (2009). Copy number variation of CCL3-like genes affects rate of progression to simian-AIDS in Rhesus Macaques (*Macaca mulatta*). *PLOS Genet.* Jan;5(1):e1000346. PMID: PMC2621346.
- *Itsara A, Cooper GM, Baker C, Girirajan S, Li J, Absher D, Krauss RM, Myers RM, Ridker PM, Chasman DI, Mefford H, Ying P, Nickerson DA, **Eichler EE**. (2009). Population analysis of large copy number variants and hotspots of human genetic disease. *Am J Hum Genet.* Feb;84(2):148–161. PMID: PMC2668011.
- de Cid R, Riveira-Munoz E, Zeeuwen PL, Robarge J, Liao W, Dannhauser EN, Giardina E, Stuart PE, Nair R, Helms C, Escaramís G, Ballana E, Martín-Ezquerria G, den Heijer M, Kamsteeg M, Joosten I, **Eichler EE**, Lázaro C, Pujol RM, Armengol L, Abecasis G, Elder JT, Novelli G, Armour JA, Kwok PY, Bowcock A, Schalkwijk J, Estivill X. (2009). Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. *Nat Genet.* Feb;41(2):211–215. PMID: PMC3128734.
- *Marques-Bonet T, Kidd JM, Ventura M, Graves TA, Cheng Z, Hillier LW, Jiang Z, Baker C, Malfavon-Borja R, Fulton LA, Alkan C, Aksay G, Girirajan S, Siswara P, Chen L, Cardone MF, Navarro A, Mardis ER, Wilson RK, **Eichler EE**. (2009). A burst of segmental duplications in the genome of the African great ape ancestor. *Nature* Feb 12;457(7231):877–881. PMID: PMC2751663.
- *Bekpen C, Marques-Bonet T, Alkan C, Antonacci F, Leogrande MB, Ventura M, Kidd JM, Siswara P, Howard JC, **Eichler EE**. (2009). Death and resurrection of the human IRGM gene. *PLOS Genet* Mar;5(3):e1000403. PMID: PMC2644816.
- van Bon BW, Mefford HC, Menten B, Koolen DA, Sharp AJ, Nillesen WM, Innis JW, ... (44 authors) ..., Brunner HG, **Eichler EE**, Kleefstra T, de Vries BB. (2009). Further delineation of the 15q13 microdeletion and duplication syndromes: A clinical spectrum varying from non-pathogenic to a severe outcome. *J Med Genet* Aug;46(8):511–523. PMID: PMC3395372.
- *Antonacci F, Kidd JM, Marques-Bonet T, Ventura M, Siswara P, Jiang Z, **Eichler EE**. (2009). Characterization of six human disease-associated inversion polymorphisms. *Hum Mol Genet* Jul 15;18(14):2555–2566. PMID: PMC2701327.
- *Liu GE, Alkan C, Jiang L, Zhao S, **Eichler EE**. (2009). Comparative analysis of Alu repeats in primate genomes. *Genome Res* May;19(5):876–885. PMID: PMC2675976.
- Zhao Y, Marotta M, **Eichler EE**, Eng C, Tanaka H. (2009). Linkage disequilibrium between two high-frequency deletion polymorphisms: implications for association studies involving the glutathione-S transferase (GST) genes. *PLOS Genet* May;5(5):e1000472. PMID: PMC2672168.
- Cellamare A, Catacchio CR, Alkan C, Giannuzzi G, Antonacci F, Cardone MF, Della Valle G, Malig M, Rocchi M, **Eichler EE**, Ventura M. (2009). New insights into centromere organization and evolution from the white-cheeked gibbon and marmoset. *Mol Biol Evol* Aug;26(8):1889–1900. PMID: PMC2734153.
- Hormozdiari F, Alkan C, **Eichler EE**, Sahinalp SC. (2009). Combinatorial algorithms for structural variation detection in high-throughput sequenced genomes. *Genome Res* Jul;19(7):1270–1278. PMID: PMC2704429.
- De Bustos C, Ramos E, Young JM, Tran RK, Menzel U, Langford CF, **Eichler EE**, Hsu L, Henikoff S, Dumanski JP, Trask BJ. (2009). Tissue-specific variation in DNA methylation levels along human chromosome 1. *Epigenetics Chromatin* Jun 8;2(1):7. PMID: PMC2706828.
- *Mefford HC, Cooper GM, Zerr T, Smith JD, Baker C, Shafer N, Thorland EC, Skinner C, Schwartz CE, Nickerson DA, **Eichler EE**. (2009). A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. *Genome Res* Sep;19(9):1579–1585. PMID: PMC2752120.
- Smith JJ, Antonacci F, **Eichler EE**, Amemiya CT. (2009) Programmed loss of millions of base pairs from a vertebrate genome. *Proc Natl Acad Sci U S A* Jul 7;106(27):11212–11217. PMID: PMC2708698.
- Dibbens LM, Mullen S, Helbig I, Mefford HC, Bayly MA, Bellows S, Leu C, Trucks H, Obermeier T, Wittig M, Franke A, Caglayan H, Yapici Z, EPICURE Consortium, Sander T, **Eichler EE**, Scheffer IE, Mulley JC, Berkovic SF. (2009) Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: Precedent for disorders with complex inheritance. *Hum Mol Gene* Oct 1;18(19):3626–3631. PMID: PMC3465696.

- 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. (2009). Targeted capture and massively parallel sequencing of 12 human exomes. *Nature* Sep 10;461(7261):272–276. PMID: PMC2844771.
- *Alkan C, Kidd JM, Marques-Bonet T, Aksay G, Antonacci F, Hormozdiari F, Kitzman JO, Baker C, Malig M, Mutlu O, Sahinalp SC, Gibbs RA, **Eichler EE**. (2009). Personalized copy number and segmental duplication maps using next-generation sequencing. *Nat Genet* Oct;41(10):1061–1067. PMID: PMC2875196.
- *Liu GE, Ventura M, Cellamare A, Chen L, Cheng Z, Zhu B, Li C, Song J, **Eichler EE**. (2009). Analysis of recent segmental duplications in the bovine genome. *BMC Genomics* Dec 1;10(1):571. PMID: PMC2796684.
- de Kovel CG, Trucks H, Helbig I, Mefford HC, Baker C, Leu C, Kluck C, Muhle H, von Spiczak S, Ostertag P, Obermeier T, KleefuB-Lie AA, Hallmann K, Steffens M, Gaus V, Klein KM, Hamer HM, Rosenow F, Brilstra EH, Kasteleijn-Nolst Trenite D, Swinkels ME, Weber YG, Unterberger I, Zimprich F, Urak L, Feucht M, Fuchs K, Moller RS, Hjalgrim H, De Jonghe P, Suls A, Ruckert IM, Wichmann HE, Franke A, Schreiber S, Nurnberg P, Elger CE, Lerche H, Stephani U, Koeleman BP, Lindhout D, **Eichler EE**, Sander T. (2010). Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. *Brain* Jan;133(Pt 1):23–32. Epub 2009 Oct 20. PMID: PMC2801323.
- Zerr T, Cooper GM, **Eichler EE**, Nickerson DA. (2010). Targeted interrogation of copy number variation using SCIMMkit. *Bioinformatics* Jan 1;26(1):120–122. PMID: PMC2796813.
- Hansen S, **Eichler EE**, Fullerton SM, Carrell D. (2010). SPANX Gene Variation in Fertile and Infertile Males. *Syst Biol Reprod Med* Feb;55:18–26. PMID: 20073942. PMID: N/A.
- Silengo M, Belligni E, Molinatto C, Baldassare G, Biamino E, Chiesa N, Zuffardi O, Girirajan S, **Eichler EE**, Ferrero GB. (2010). Eyebrow anomalies as a diagnostic sign of genomic disorders. *Clin Genet* Jan;77(1):28–31. PMID: 20092588. PMID: N/A.
- *Girirajan S, Rosenfeld JA, Cooper GM, Antonacci F, Siswara P, Itsara A, Vives L, Walsh T, McCarthy SE, Baker C, Mefford HC, Kidd JM, Browning SR, Browning BL, Dickel DE, Levy DL, Ballif BC, Platky K, Farber DM, Gowans GC, Wetherbee JJ, Asamoah A, Weaver DD, Mark PR, Dickerson J, Garg BP, Ellingwood SA, Smith R, Banks VC, Smith W, McDonald MT, Hoo JJ, French BN, Hudson C, Johnson JP, Ozmore JR, Moeschler JB, Surti U, Escobar LF, El-Khechen D, Gorski JL, Kussmann J, Salbert B, Lacassie Y, Biser A, McDonald-McGinn DM, Zackai EH, Deardorff MA, Shaikh TH, Haan E, Friend KL, Fichera M, Romano C, Gez J, Delisi LE, Sebat J, King MC, Shaffer LG, **Eichler EE**. (2010). A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. *Nat Genet* Mar;42(3):203–209. PMID: PMC2847896.
- Rosenfeld JA, Coppinger J, Bejjani BA, Girirajan S, **Eichler EE**, Shaffer LG, Ballif BC. (2010). Speech delays and behavioral problems are the predominant features in individuals with developmental delays and 16p11.2 microdeletions and microduplications. *Journal of Neurodevelopmental Disorders* Mar;2(1):26–38. PMID: PMC3125720.
- Liu GE, Hou Y, Zhu B, Cardone MF, Jiang L, Cellamare A, Mitra A, Alexander LJ, Coutinho LL, Dell'aquila ME, Gasbarre LC, Lacalandra G, Li RW, Matukumalli LK, Nonneman D, Regitano LC, Smith TP, Song J, Sonstegard TS, Van Tassell CP, Ventura M, **Eichler EE**, McDanel TG, Keele JW. (2010). Analysis of copy number variations among diverse cattle breeds. *Genome Res* May;20(5):693–703. PMID: PMC2860171.
- Green RE, Krause J, Briggs AW, Maricic T, Stenzel U, Kircher M, Patterson N, Li H, Zhai W, Fritz MH, Hansen NF, Durand EY, Malaspinas AS, Jensen JD, Marques-Bonet T, Alkan C, Prüfer K, Meyer M, Burbano HA, Good JM, Schultz R, Aximu-Petri A, Butthof A, Höber B, Höffner B, Siegemund M, Weihmann A, Nusbaum C, Lander ES, Russ C, Novod N, Affourtit J, Egholm M, Verna C, Rudan P, Brajkovic D, Kucan Z, Gusic I, Doronichev VB, Golovanova LV, Lalueza-Fox C, de la Rasilla M, Fordea J, Rosas A, Schmitz RW, Johnson PL, **Eichler EE**, Falush D, Birney E, Mullikin JC, Slatkin M, Nielsen R, Kelso J, Lachmann M, Reich D, Pääbo S. (2010). A draft sequence of the Neandertal genome. *Science* May 7;328(5979):710–722. PMID: 20448178. PMID: N/A.
- Hajirasouliha I, Hormozdiari F, Alkan C, Kidd JM, Birol I, **Eichler EE**, Sahinalp SC. (2010). Detection and characterization of novel sequence insertions using paired-end next-generation sequencing. *Bioinformatics* May 15;26(10):1277–1283. PMID: PMC2865866.
- *Kidd JM, Sampas N, Antonacci F, Graves T, Fulton R, Hayden HS, Alkan C, Malig M, Ventura M, Giannuzzi G, Kallicki J, Anderson P, Tsalenko A, Yamada NA, Tsang P, Kaul R, Wilson RK, Bruhn L, **Eichler EE**. (2010). Characterization of missing human genome sequences and copy-number polymorphic insertions. *Nat Methods* May;7(5):365–371. PMID: PMC2875995.
- *Mefford HC, Muhle H, Ostertag P, von Spiczak S, Buysse K, Baker C, Franke A, Malafosse A, Genton P, Thomas P, Gurnett CA, Schreiber S, Bassuk AG, Guipponi M, Stephani U, Helbig I, **Eichler EE**. (2010). Genome-wide copy number variation in epilepsy: novel susceptibility loci in idiopathic generalized and focal epilepsies. *PLOS Genet* May 20;6(5):e1000962. PMID: PMC2873910.
- Hormozdiari F, Hajirasouliha I, Dao P, Hach F, Yorukoglu D, Alkan C, **Eichler EE**, Sahinalp SC. (2010). Next-generation VariationHunter: Combinatorial algorithms for transposon insertion discovery. *Bioinformatics* Jun 15;26(12):i350–357. PMID: PMC2881400.

- Teague B, Waterman MS, Goldstein S, Potamouisis K, Zhou S, Reslewic S, Sarkar D, Valouev A, Churas C, Kidd JM, Kohn S, Runnheim R, Lamers C, Forrest D, Newton MA, **Eichler EE**, Kent-First M, Surti U, Livny M, Schwartz DC. (2010). High-resolution human genome structure by single-molecule analysis. *Proc Natl Acad Sci U S A* Jun 15;107(24):10848–10853. PMID: PMC2890719.
- Beck CR, Collier P, Macfarlane C, Malig M, Kidd JM, **Eichler EE**, Badge RM, Moran JV. (2010). LINE-1 retrotransposition activity in human genomes. *Cell* Jun 25;141(7):1159–1170. PMID: PMC3013285.
- Hach F, Hormozdiari F, Alkan C, Hormozdiari F, Birol I, **Eichler EE**, Sahinalp SC. (2010). mrsFAST: a cache-oblivious algorithm for short-read mapping. *Nat Methods* Aug;7(8):576–577. PMID: PMC3115707.
- Collie AM, Landsverk ML, Ruzzo E, Mefford HC, Buysse K, Adkins JR, Knutzen DM, Barnett K, Brown RH Jr, Parry GJ, Yum SW, Simpson DA, Olney RK, Chinnery PF, **Eichler EE**, Chance PF, Hannibal MC. (2010). Non-recurrent SEPT9 duplications cause hereditary neuralgic amyotrophy. *J Med Genet* Sep;47(9):601–607. Epub 2009 Nov 25. PMID: 19939853. PMID: N/A.
- Mefford HC, Shafer N, Antonacci F, Tsai JM, Park SS, Hing AV, Rieder MJ, Smyth MD, Speltz ML, **Eichler EE**, Cunningham ML. (2010). Copy number variation analysis in single-suture craniosynostosis: Multiple rare variants including RUNX2 duplication in two cousins with metopic craniosynostosis. *Am J Med Genet A* Sep;152A(9):2203–2210. PMID: PMC3104131.
- *Antonacci F, Kidd JM, Marques-Bonet T, Teague B, Ventura M, Girirajan S, Alkan C, Campbell CD, Vives L, Malig M, Rosenfeld JA, Ballif BC, Shaffer LG, Graves TA, Wilson RK, Schwartz DC, **Eichler EE**. (2010). A large and complex structural polymorphism at 16p12.1 underlies microdeletion disease risk. *Nat Genet* Sep;42(9):745–750. PMID: PMC2930074.
- Bachmann-Gagescu R, Mefford HC, Cowan C, Glew GM, Hing AV, Wallace S, Bader PI, Hamati A, Reitnauer PJ, Smith R, Stockton DW, Muhle H, Helbig I, **Eichler EE**, Ballif BC, Rosenfeld J, Tsuchiya KD. (2010). Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity. *Genet Med* Oct;12(10):641–647. PMID: 20808231. PMID: N/A.
- *Itsara A, Wu H, Smith JD, Nickerson DA, Romieu I, London SJ, **Eichler EE**. (2010). De novo rates and selection of large copy number variation. *Genome Res* Nov;20(11):1469–1481. PMID: PMC2963811.
- *Sudmant PH, Kitzman JO, Antonacci F, Alkan C, Malig M, Tsalenko A, Sampas N, Bruhn L, Shendure J, 1000 Genomes Project, **Eichler EE**. (2010). Diversity of human copy number variation and multicopy genes. *Science* October 29;330(6004):641–646. PMID: PMC3020103.
- *Kidd JM, Graves T, Newman TL, Fulton R, Hayden HS, Malig M, Kallicki J, Kaul R, Wilson RK, **Eichler EE**. (2010). A human genome structural variation sequencing resource reveals insights into mutational mechanisms. *Cell* Nov 24;143(5):837–847. PMID: PMC3026629.
- Reich D, Green RE, Kircher M, Krause J, Patterson N, Durand EY, Viola B, Briggs AW, Stenzel U, Johnson PL, Maricic T, Good JM, Marques-Bonet T, Alkan C, Fu Q, Mallick S, Li H, Meyer M, **Eichler EE**, Stoneking M, Richards M, Talamo S, Shunkov MV, Derevianko AP, Hublin JJ, Kelso J, Slatkin M, Pääbo S. (2010). Genetic history of an archaic hominin group from Denisova Cave in Siberia. *Nature* Dec 23;468(7327):1053–1060. PMID: PMC4306417.
- Alkan C, Cardone MF, Catacchio CR, Antonacci F, O'Brien SJ, Ryder OA, Purgato S, Zoli M, Della Valle G, **Eichler EE**, Ventura M. (2011). Genome-wide characterization of centromeric satellites from multiple mammalian genomes. *Genome Res* Jan;21(1):137–145. Epub 2010 Nov 16. PMID: PMC3012921.
- *Alkan C, Sajjadian S, **Eichler EE**. (2011). Limitations of next-generation genome sequence assembly. *Nat Methods* Jan;8(1):61–65. Epub 2010 Nov 21. PMID: PMC3115693.
- Hurle B, Marques-Bonet T, Antonacci F, Hughes I, Ryan JF, Comparative Sequencing Program N, **Eichler EE**, Ornitz DM, Green ED. (2011). Lineage-specific evolution of the vertebrate Otopetrin gene family revealed by comparative genomic analyses. *BMC Evol Biol* Jan 24;11(1):23. PMID: PMC3038909.
- Locke DP, Hillier LW, Warren WC, Worley KC, Nazareth LV, Muzny DM, Yang SP, Wang Z, Chinwalla AT, Minx P, Mitreva M, Cook L, Delehaunty KD, Fronick C, Schmidt H, Fulton LA, Fulton RS, Nelson JO, Magrini V, Pohl C, Graves TA, Markovic C, Cree A, Dinh HH, Hume J, Kovar CL, Fowler GR, Lunter G, Meader S, Heger A, Ponting CP, Marques-Bonet T, Alkan C, Chen L, Cheng Z, Kidd JM, **Eichler EE**, White S, Searle S, Vilella AJ, Chen Y, Flicek P, Ma J, Raney B, Suh B, Burhans R, Herrero J, Haussler D, Faria R, Fernando O, Darre F, Farre D, Gazave E, Oliva M, Navarro A, Roberto R, Capozzi O, Archidiacono N, Valle GD, Purgato S, Rocchi M, Konkel MK, Walker JA, Ullmer B, Batzer MA, Smit AF, Hubley R, Casola C, Schrider DR, Hahn MW, Quesada V, Puente XS, Ordonez GR, Lopez-Otin C, Vinar T, Brejova B, Ratan A, Harris RS, Miller W, Kosiol C, Lawson HA, Taliwal V, Martins AL, Siepel A, Roychoudhury A, Ma X, Degenhardt J, Bustamante CD, Gutenkunst RN, Mailund T, Dutheil JY, Hobolth A, Schierup MH, Ryder OA, Yoshinaga Y, de Jong PJ, Weinstock GM, Rogers J, Mardis ER, Gibbs RA, Wilson RK. (2011). Comparative and demographic analysis of orang-utan genomes. *Nature* Jan 27;469(7331):529–533. PMID: PMC3060778.
- Kitzman JO, Mackenzie AP, Adey A, Hiatt JB, Patwardhan RP, Sudmant PH, Ng SB, Alkan C, Qiu R, **Eichler EE**, Shendure J. (2011). Haplotype-resolved genome sequencing of a Gujarati Indian individual. *Nat Biotechnol* Jan;29(1):59–63. Epub 2010 Dec 19. PMID: PMC3116788.

- *Campbell CD, Sampas N, Tsalenko A, Sudmant PH, Kidd JM, Malig M, Vu TH, Vives L, Tsang P, Bruhn L, **Eichler EE**. (2011). Population-genetic properties of differentiated human copy-number polymorphisms. *Am J Hum Genet* Mar 11;88(3):317–332. PMID: PMC3059424.
- *Hormozdiari F, Alkan C, Ventura M, Hajirasouliha I, Malig M, Hach F, Yorukoglu D, Dao P, Bakhshi M, Sahinalp SC, **Eichler EE**. (2011). Alu repeat discovery and characterization within human genomes. *Genome Res* Jun;21(6):840–849. Epub 2010 Dec 3. PMID: PMC3106317.
- *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**. (2011). Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. *Nat Genet* Jun;43(6):585–589. PMID: PMC3115696.
- Hormozdiari F, Hach F, Sahinalp SC, **Eichler EE**, Alkan C. (2011). Sensitive and fast mapping of di-base encoded reads. *Bioinformatics* Jul 15;27(14):1915–1921. PMID: PMC3129524.
- Church DM, Schneider VA, Graves T, Auger K, Cunningham F, Bouk N, Chen HC, Agarwala R, McLaren WM, Ritchie GR, Albracht D, Kremitzki M, Rock S, Kotkiewicz H, Kremitzki C, Wollam A, Trani L, Fulton L, Fulton R, Matthews L, Whitehead S, Chow W, Torrance J, Dunn M, Harden G, Threadgold G, Wood J, Collins J, Heath P, Griffiths G, Pelan S, Grafham D, **Eichler EE**, Weinstock G, Mardis ER, Wilson RK, Howe K, Flicek P, Hubbard T. (2011). Modernizing reference genome assemblies. *PLOS Biol* Jul;9(7):e1001091. PMID: PMC3130012.
- *Cooper GM, Coe BP, Girirajan S, Rosenfeld JA, Vu TH, Baker C, Williams C, Stalker H, Hamid R, Hannig V, Abdel-Hamid H, Bader P, McCracken E, Niyazov D, Leppig K, Thiess H, Hummel M, Alexander N, Gorski J, Kussmann J, Shashi V, Johnson K, Rehder C, Ballif BC, Shaffer LG, **Eichler EE**. (2011). A copy number variation morbidity map of developmental delay. *Nat Genet* Aug 14;43(9):838–846. PMID: PMC3171215.
- Nicholas TJ, Baker C, **Eichler EE**, Akey JM. (2011). A high-resolution integrated map of copy number polymorphisms within and between breeds of the modern domesticated dog. *BMC Genomics* Aug 16;12(1):414. PMID: PMC3166287.
- *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**. (2011). Gorilla genome structural variation reveals evolutionary parallelisms with chimpanzee. *Genome Res* Oct;21(10):1640–1649. PMID: PMC3202281.
- Gazave E, Darre F, Morcillo-Suarez C, Petit-Marty N, Carreno A, Marigorta UM, Ryder OA, Blancher A, Rocchi M, Bosch E, Baker C, Marques-Bonet T, **Eichler EE**, Navarro A. (2011). Copy number variation analysis in the great apes reveals species-specific patterns of structural variation. *Genome Res* Oct;21(10):1626–39. PMID: PMC3202280.
- Renton AE, Majounie E, Waite A, Simon-Sahez J, Rollinson S, Gibbs JR, Schymick JC, Laaksovirta H, van Swieten JC, Myllykangas L, Kalimo H, Paetau A, Abramzon Y, Remes AM, Kaganovich A, Scholz SW, Duckworth J, Ding J, Harmer DW, Hernandez DG, Johnson JO, Mok K, Ryten M, Trabzuni D, Guerreiro RJ, Orrell RW, Neal J, Murray A, Pearson J, Jansen IE, Sondervan D, Seelaar H, Blake D, Young K, Halliwell N, Callister JB, Toulson G, Richardson A, Gerhard A, Snowden J, Mann D, Neary D, Nalls MA, Peuralinna T, Jansson L, Isoviita VM, Kaivorinne AL, Holtta-Vuori M, Ikonen E, Sulkava R, Benatar M, Wu J, Chio A, Restagno G, Borghero G, Sabatelli M; ITALSGEN Consortium, Heckerman D, Rogaeva E, Zinman L, Rothstein JD, Sendtner M, Drepper C, **Eichler EE**, Alkan C, Abdullaev Z, Pack SD, Dutra A, Pak E, Hardy J, Singleton A, Williams NM, Heutink P, Pickering-Brown S, Morris HR, Tienari PJ, Traynor BJ. (2011). A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9p21-linked ALS-FTD. *Neuron* Oct 20;72(2):257–268. PMID: PMC3200438.
- Sivakumaran TA, Igo RP Jr, Kidd JM, Itsara A, Kopplin LJ, Chen W, Hagstrom SA, Peachey NS, Francis PJ, Klein ML, Chew EY, Ramprasad VL, Tay WT, Mitchell P, Seielstad M, Stambolian DE, Edwards AO, Lee KE, Leontiev DV, Jun G, Wang Y, Tian L, Qiu F, Henning AK, LaFramboise T, Sen P, Aarthi M, George R, Raman R, Das MK, Vijaya L, Kumaramanickavel G, Wong TY, Swaroop A, Abecasis GR, Klein R, Klein BE, Nickerson DA, **Eichler EE**, Iyengar SK. (2011). A 32 kb critical region excluding Y402H in CFH mediates risk for age-related macular degeneration. *PLOS One* 6(10):e25598. PMID: PMC3192039.
- *Girirajan S, Brkanac Z, Coe BP, Baker C, Vives L, Vu TH, Shafer N, Bernier R, Ferrero GB, Silengo M, Warren ST, Moreno CS, Fichera M, Romano C, Raskind WH, **Eichler EE**. (2011). Relative burden of large CNVs on a range of neurodevelopmental phenotypes. *PLOS Genet* Nov;7(11):e1002334. PMID: PMC3213131.
- Vu TH, Coccaro EF, **Eichler EE**, Girirajan S. (2011). Genomic architecture of aggression: Rare copy number variants in intermittent explosive disorder. *Am J Med Genet B Neuropsychiatr Genet* Dec;156B(7):808–816. PMID: PMC3168586.
- Hormozdiari F, Hajirasouliha I, McPherson A, **Eichler EE**, Sahinalp SC. (2011). Simultaneous structural variation discovery in multiple paired-end sequenced genomes. *Genome Res* Dec;21(12):2203–2212. PMID: PMC3227108.
- Muhle H, Mefford HC, Obermeier T, von Spiczak S, **Eichler EE**, Stephani U, Sander T, Helbig I. (2011). Absence seizures with intellectual disability as a phenotype of the 15q13.3 microdeletion syndrome. *Epilepsia* Dec;52(12):e194–198. PMID: PMC3270691.

- *Karakoc E, Alkan C, O'Roak BJ, Dennis MY, Vives L, Mark K, Rieder MJ, Nickerson DA, **Eichler EE**. (2011). Detection of structural variants and indels within exome data. *Nat Methods* Dec 18;9(2):176–178. PMID: PMC3269549.
- Mefford HC, Yendle SC, Hsu C, Cook J, Geraghty E, McMahon JM, Eeg-Olofsson O, Sadleir LG, Gill D, Ben-Zeev B, Lerman-Sagie T, Mackay M, Freeman JL, Andermann E, Pelakanos JT, Andrews I, Wallace G, **Eichler EE**, Berkovic SF, Scheffer IE. (2011). Rare copy number variants are an important cause of epileptic encephalopathies. *Ann Neurol* Dec;70(6):974–985. PMID: PMC3245646.
- Chen YZ, Matsushita M, Girirajan S, Lisowski M, Sun E, Sul Y, Bernier R, Estes A, Dawson G, Minshew N, Shellenberg GD, **Eichler EE**, Rieder MJ, Nickerson DA, Tsuang DW, Tsuang MT, Wijsman EM, Raskind WH, Brkanac Z. (2012). Evidence for involvement of GNB1L in autism. *Am J Med Genet B Neuropsychiatr Genet* Jan;159B(1):61–71. Epub 2011 Nov 16. PMID: PMC3270696.
- *Mefford HC, Rosenfeld JA, Shur N, Slavotinek AM, Cox VA, Hennekam RC, Firth HV, Willatt L, Wheeler P, Morrow EM, Cook J, Sullivan R, Oh A, McDonald MT, Zonana J, Keller K, Hannibal MC, Ball S, Kussmann J, Gorski J, Zelewski S, Banks V, Smith W, Smith R, Paull L, Rosenbaum KN, Amor DJ, Silva J, Lamb A, **Eichler EE**. (2012). Further clinical and molecular delineation of the 15q24 microdeletion syndrome. *J Med Genet* Feb;49(2):110–118. Epub 2011 Dec 17. PMID: PMC3261729.
- Lamb AN, Rosenfeld JA, Neill NJ, Talkowski ME, Blumenthal I, Girirajan S, Keelean-Fuller D, Fan Z, Pouncey J, Stevens C, Mackay-Loder L, Terespolsky D, Bader P, Rosenbaum K, Vallee S, Moeschler JB, Ladda R, Sell S, Martin J, Ryan S, Jones MC, Moran R, Shealy A, Madan-Khetarpal S, McConnell J, Surti U, Delahaye A, Heron-Longe B, Pipiras E, Benzacken B, Passemard S, Verloes A, Isidor B, Caignec CL, Glew GM, Opheim KE, **Eichler EE**, Morton CC, Gusella JF, Schultz RA, Ballif BC, Shaffer LG. (2012). Haploinsufficiency of SOX5 at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. *Hum Mutat* Apr;33(4):728–740. PMID: PMC3618980.
- Bickhart DM, Hou Y, Schroeder SG, Alkan C, Cardone MF, Matukumalli LK, Song J, Schnabel RD, Ventura M, Taylor JF, Garcia JF, Van Tassell CP, Sonstegard TS, **Eichler EE**, Liu GE. (2012). Copy number variation of individual cattle genomes using next-generation sequencing. *Genome Res* Apr;22(4):778–790. PMID: PMC3317159.
- Veeramah KR, O'Brien JE, Meisler MH, Cheng X, Dib-Hajj SD, Waxman SG, Talwar D, Girirajan S, **Eichler EE**, Restifo LL, Erickson RP, Hammer MF. (2012). De novo pathogenic SCN8A mutation identified by whole-genome sequencing of a family quartet affected by infantile epileptic encephalopathy and SUDEP. *Am J Hum Genet* Mar 9;90(3):502–510. PMID: PMC3309181.
- *Bekpen C, Tastekin I, Siswara P, Akdis CA, **Eichler EE**. (2012). Primate segmental duplication creates novel promoters for the LRRC37 gene family within the 17q21.31 inversion polymorphism region. *Genome Res* Jun;22(6):1050–1058. PMID: PMC3371713.
- *Ventura M, Catacchio C, Sajjadian S, Vives L, Sudmant P, Marques-Bonet T, Graves TA, Wilson RK, **Eichler EE**. (2012). The evolution of African great ape subtelomeric heterochromatin and the fusion of human chromosome 2. *Genome Res* Jun;22(6):1036–1049. PMID: PMC3371704.
- *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**. (2012). Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. *Nature* Apr 4;485(7397):246–250. PMID: PMC3350576.
- *Itsara A, Vissers LELM, Steinberg KM, Meyer KJ, Zody MC, Koolen DA, de Ligt J, Cuppen E, Baker C, Lee C, Graves TA, Wilson RK, Jenkins RB, Veltman JA, **Eichler EE**. (2012). Resolving the breakpoints of the 17q21.31 microdeletion syndrome with next-generation sequencing. *Am J Hum Genet* Apr 6;90(4):599–613. PMID: PMC3322237.
- Priest JR, Girirajan S, Vu TH, Olson A, **Eichler EE**, Portman MA. (2012). Rare copy number variants in isolated sporadic and syndromic atrioventricular septal defects. *Am J Med Genet A* Jun;158A(6):1279–1784. PMID: PMC3564951.
- *Dennis MY, Nuttle X, Sudmant PH, Antonacci F, Graves TA, Nefedov M, Rosenfeld JA, Sajjadian S, Malig M, Kotkiewicz H, Curry CJ, Shafer S, Shaffer LF, de Jong PJ, Wilson RK, **Eichler EE**. (2012). Evolution of human-specific neural SRGAP2 genes by incomplete segmental duplication. *Cell* 03 May 11;149(4):912–922. PMID: PMC3365555.
- *Krumm N, Sudmant PH, Ko A, O'Roak BJ, Malig M, Coe BP, NHLBI Exome Sequencing Project N, Quinlan AR, Nickerson DA, **Eichler EE**. (2012). Copy number variation detection and genotyping from exome sequence data. *Genome Res* Aug;22(8):1525–1532. PMID: PMC3409265.
- 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. (2012). Noninvasive whole-genome sequencing of a human fetus. *Sci Transl Med* Jun 6;4(137):137ra76. PMID: PMC3379884.
- *Steinberg KM, Antonacci F, Sudmant PH, Kidd JM, Campbell CD, Vives L, Malig M, Scheinfeldt L, Beggs W, Ibrahim M, Lema G, Nyambo TB, Omar SA, Bodo JM, Froment A, Donnelly MP, Kidd KK, Tishkoff SA, **Eichler EE**. (2012). Structural diversity and African origin of the 17q21.31 inversion polymorphism. *Nat Genet* Jul 1;44(8):872–880. PMID: PMC3408829.

- Chen YZ, Matsushita MM, Robertson P, Rieder M, Girirajan S, Antonacci F, Lipe H, **Eichler EE**, Nickerson DA, Bird TD, Raskind WH. (2012). Autosomal dominant familial dyskinesia and facial myokymia: single exome sequencing identifies a mutation in adenylyl cyclase 5. *Arch Neurol* May 1;69(5):630–635. PMID: PMC3508680.
- Smith JJ, Baker C, **Eichler EE**, Amemiya CT. (2012). Genetic Consequences of Programmed Genome Rearrangement. *Curr Biol* Aug 21;22(16):1524–1529. PMID: PMC3427415.
- 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. (2012). Non-invasive fetal genome sequencing: Opportunities and challenges. *Am J Med Genet A* Oct;158A(10):2382–2384. PMID: PMC3448836.
- 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. (2012). A high-coverage genome sequence from an archaic Denisovan individual. *Science* Oct 12;338(6104):222–226. PMID: PMC3617501.
- *Girirajan S, Rosenfeld JA, Coe BP, Parikh S, Friedman N, Goldstein A, Filipink RA, McConnell JS, Angle B, Meschino WS, Nezarati MM, Asamoah A, Jackson KE, Gowans GC, Martin JA, Carmany EP, Stockton DW, Schnur RE, Penney LS, Martin DM, Raskin S, Leppig K, Thiese H, Smith R, Aberg E, Niyazov DM, Escobar LF, El-Khechen D, Johnson KD, Lebel RR, Siefkas K, Ball S, Shur N, McGuire M, Brasington CK, Spence JE, Martin LS, Clericuzio C, Ballif BC, Shaffer LG, **Eichler EE**. (2012). Phenotypic heterogeneity of genomic disorders and rare copy-number variants. *N Engl J Med* Oct 4;367(14):1321–1331. PMID: PMC3494411.
- *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**. (2012). Estimating the human mutation rate using autozygosity in a founder population. *Nat Genet* Nov;44(11):1277–1281. PMID: PMC3483378.
- *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. (2012). Multiplex targeted sequencing identifies recurrently mutated genes in autism spectrum disorders. *Science* Dec 21;338(6114):1619–1622. PMID: PMC3528801.
- *Giannuzzi G, Siswara P, Malig M, Marques-Bonet T, NISC Comparative Sequencing Program, Mullikin JC, Ventura M, **Eichler EE**. (2013). Evolutionary dynamism of the primate LRRC37 gene family. *Genome Res* Jan;23(1):46–59. doi: 10.1101/gr.138842.112. Epub 2012 Oct 11. PMID: PMC3530683.
- Mueller M, Barros P, Witherden AS, Roberts AL, Zhang Z, Schaschl H, Yu CY, Hurler ME, Schaffner C, Floto RA, Game L, Steinberg KM, Wilson RK, Graves TA, **Eichler EE**, Cook HT, Vyse TJ, Aitman TJ. (2013). Genomic pathology of SLE-associated copy-number variation at the FCGR2C/FCGR3B/FCGR2B locus. *Am J Hum Genet* Jan 10;92(1):28–40. Epub 2012 Dec 20. PMID: PMC3542466.
- *Lorente-Galdos B, Bleyhl J, Santpere G, Vives L, Ramirez O, Hernandez J, Anglada R, Cooper GM, Navarro A, **Eichler EE**, Marques-Bonet T. (2013). Accelerated exon evolution within primate segmental duplications. *Genome Biol* Jan 29;14(1):R9. PMID: PMC3906575.
- Tucci A, Kara E, Schossig A, Wolf NI, Plagnol V, Fawcett K, Paisan-Ruiz C, Moore M, Hernandez D, Musumeci S, Tennison M, Hennekam R, Palmeri S, Malandrini A, Raskin S, Donnai D, Hennig C, Tzschach A, Hordijk R, Bast T, Wimmer K, Lo CN, Shorvon S, Mefford H, **Eichler EE**, Hall R, Hayes I, Hardy J, Singleton A, Zschocke J, Houlden H. (2013). Kohlschütter-Tönz syndrome: Mutations in ROGDI and evidence of genetic heterogeneity. *Hum Mutat* Feb;34(2):296–300. Epub 2012 Nov 27. PMID: PMC3902979.
- Beunders G, Voorhoeve E, Golzio C, Pardo LM, Rosenfeld JA, Talkowski ME, Simonic I, Lionel AC, Vergult S, Pyatt RE, van de Kamp J, Nieuwint A, Weiss MM, Rizzu P, Verwer LE, van Spaendonk RM, Shen Y, Wu BL, Yu T, Yu Y, Chiang C, Gusella JF, Lindgren AM, Morton CC, van Binsbergen E, Bulk S, van Rossem E, Vanakker O, Armstrong R, Park SM, Greenhalgh L, Maye U, Neill NJ, Abbott KM, Sell S, Ladda R, Farber DM, Bader PI, Cushing T, Drautz JM, Konczal L, Nash P, Reyes ED, Carter MT, Hopkins E, Marshall CR, Osborne LR, Gripp KW, Thrush DL, Hashimoto S, Gastier-Foster JM, Astbury C, Ylstra B, Meijers-Heijboer H, Posthuma D, Menten B, Mortier G, Scherer SW, **Eichler EE**, Girirajan S, Katsanis N, Groffen AJ, Sistermans EA. (2013). Exonic deletions in AUTS2 cause a syndromic form of intellectual disability and suggest a critical role for the C terminus. *Am J Hum Genet* Feb 7;92(2):210–220. PMID: PMC3567268.
- *Girirajan S, Dennis MY, Baker C, Malig M, Coe BP, Campbell CD, Mark K, Vu TH, Alkan C, Cheng Z, Biesecker LG, Bernier R, **Eichler EE**. (2013). Refinement and discovery of new hotspots of copy-number variation associated with autism spectrum disorder. *Am J Hum Genet* Feb 7;92(2):221–237. PMID: PMC3567267.
- Watson CT, Steinberg KM, Huddleston J, Warren RL, Malig M, Schein J, Willsey AJ, Joy JB, Scott JK, Graves TA, Wilson RK, Holt RA, **Eichler EE**, Bredon F. (2013). Complete haplotype sequence of the human immunoglobulin heavy-chain variable, diversity, and joining genes and characterization of allelic and copy-number variation. *Am J Hum Genet* Apr 4;92(4):530–546. PMID: PMC3617388.

- Korvatska O, Strand NS, Berndt JD, Strovast T, Chen DH, Leverenz JB, Kiianitsa K, Mata IF, Karakoc E, Greenup JL, Bonkowski E, Chuang J, Moon RT, **Eichler EE**, Nickerson DA, Zabetian CP, Kraemer BC, Bird TD, Raskind WH. (2013). Altered splicing of ATP6AP2 causes X-linked parkinsonism with spasticity (XPDS). *Hum Mol Genet* Aug 15;22(16):3259–3268. PMID: PMC3723311.
- Chin CS, Alexander DH, Marks P, Klammer AA, Drake J, Heiner C, Clum A, Copeland A, Huddleston J, **Eichler EE**, Turner SW, Korlach J. (2013). Nonhybrid, finished microbial genome assemblies from long-read SMRT sequencing data. *Nat Methods* Jun;10(6):563–569. PMID: 23644548. PMID: N/A.
- Timms AE, Dorschner MO, Wechsler J, Choi KY, Kirkwood R, Girirajan S, Baker C, **Eichler EE**, Korvatska O, Roche KW, Horwitz MS, Tsuang DW. (2013). Support for the N-Methyl-D-Aspartate receptor hypofunction hypothesis of schizophrenia from exome sequencing in multiplex families. *JAMA Psychiatry* Jun 1;70(6):582–590. PMID: 23553203. PMID: N/A.
- Rosenfeld JA, Coe BP, **Eichler EE**, Cuckle H, Shaffer LG. (2013). Estimates of penetrance for recurrent pathogenic copy-number variations. *Genet Med* Jun;15(6):478–481. Epub 2012 Dec 20. PMID: PMC3664238.
- Girirajan S, Johnson RL, Tassone F, Balciuniene J, Katiyar N, Fox K, Baker C, Srikanth A, Yeoh KH, Khoo SJ, Nauth TB, Hansen R, Ritchie M, Hertz-Picciotto I, **Eichler EE**, Pessah IN, Selleck SB. (2013). Global increases in both common and rare copy number load associated with autism. *Hum Mol Genet* Jul 15;22(14):2870–2880. PMID: PMC3690969.
- McMichael G, Girirajan S, Moreno-De-Luca A, Gez J, Shard C, Nguyen LS, Nicholl J, Gibson C, Haan E, **Eichler E**, Martin CL, MacLennan A. (2014). Rare copy number variation in cerebral palsy. *Eur J Hum Genet* Jan;22(1):40–45. PMID: PMC3865415.
- Prado-Martinez J, Hernando-Herraez I, Lorente-Galdos B, Dabad M, Ramirez O, Baeza-Delgado C, Morcillo-Suarez C, Alkan C, Hormozdiari F, Rainieri E, Estelle J, Fernandez-Callejo M, Valles M, Ritscher L, Schoneberg T, de la Calle-Mustienes E, Casillas S, Rubio-Acero R, Mele M, Engelken J, Caceres M, Gomez-Skarmeta JL, Gut M, Bertranpetit J, Gut IG, Abello T, **Eichler EE**, Mingarro I, Lalueza-Fox C, Navarro A, Marques-Bonet T. (2013). The genome sequencing of an albino Western lowland gorilla reveals inbreeding in the wild. *BMC Genomics* May 31;14(1):363. PMID: PMC3673836.
- *Prado-Martinez J, Sudmant PH, Kidd JM, Li H, Kelley JL, Lorente-Galdos B, Veeramah KR, Woerner AE, O'Connor TD, Santpere G, Cagan A, Theunert C, Casals F, Laayouni H, Munch K, Hobolth A, Halager AE, Malig M, Hernandez-Rodriguez J, Hernando-Herraez I, Prüfer K, Pybus M, Johnstone L, Lachmann M, Alkan C, Twigg D, Petit N, Baker C, Hormozdiari F, Fernandez-Callejo M, Dabad M, Wilson ML, Stevison L, Camprubí C, Carvalho T, Ruiz-Herrera A, Vives L, Mele M, Abello T, Kondova I, Bontrop RE, Pusey A, Lankester F, Kiyang JA, Bergl RA, Lonsdorf E, Myers S, Ventura M, Gagneux P, Comas D, Siegmund H, Blanc J, Agueda-Calpena L, Gut M, Fulton L, Tishkoff SA, Mullikin JC, Wilson RK, Gut IG, Gonder MK, Ryder OA, Hahn BH, Navarro A, Akey JM, Bertranpetit J, Reich D, Mailund T, Schierup MH, Hvilsom C, Andrés AM, Wall JD, Bustamante CD, Hammer MF, **Eichler EE**, Marques-Bonet T. (2013). Great ape genetic diversity and population history. *Nature* Jul 25;499(7459):471–475. PMID: PMC3822165.
- *Sudmant PH, Huddleston J, Catacchio CR, Malig M, Hillier LW, Baker C, Mohajeri K, Kondova I, Bontrop RE, Persengiev S, Antonacci F, Ventura M, Prado Martinez J, Marques-Bonet T, **Eichler EE**. (2013). Evolution and diversity of copy number variation in the great ape lineage. *Genome Res* Sep;23(9):1373–1382. PMID: PMC3759715.
- *Hormozdiari F, Konkel MK, Prado-Martinez J, Chiatante G, Herraez IH, Walker JA, Nelson B, Alkan C, Sudmant PH, Huddleston J, Catacchio CR, Ko A, Malig M, Baker C; Great Ape Genome Project, Marques-Bonet T, Ventura M, Batzer MA, **Eichler EE**. (2013). Rates and patterns of great ape retrotransposition. *Proc Natl Acad Sci U S A* Aug 13;110(33):13457–13462. PMID: PMC3746892.
- *Nuttall X, Huddleston J, O'Roak BJ, Antonacci F, Fichera M, Romano C, Shendure J, **Eichler EE**. (2013). Rapid and accurate large-scale genotyping of duplicated genes and discovery of interlocus gene conversions. *Nat Methods* Aug;10(9):903–909. PMID: PMC3985568.
- Epi4K Consortium, Allen AS, Berkovic SF, Cossette P, Delanty N, Dlugos D, **Eichler EE**, Epstein MP, Glauser T, Goldstein DB, Han Y, Heinzen EL, Hitomi Y, Howell KB, Johnson MR, Kuzniecky R, Lowenstein DH, Lu YF, Madou MR, Marson AG, Mefford HC, Esmaeeli Nieh S, O'Brien TJ, Ottman R, Petrovski S, Poduri A, Ruzzo EK, Scheffer IE, Sherr EH, Yuskaitis CJ; Epilepsy Phenome/Genome Project, Abou-Khalil B, Aldredge BK, Bautista JF, Berkovic SF, Boro A, Cascino GD, Consalvo D, Crumrine P, Devinsky O, Dlugos D, Epstein MP, Fiol M, Fountain NB, French J, Friedman D, Geller EB, Glauser T, Glynn S, Haut SR, Hayward J, Helmers SL, Joshi S, Kanner A, Kirsch HE, Knowlton RC, Kossoff EH, Kuperman R, Kuzniecky R, Lowenstein DH, McGuire SM, Motika PV, Novotny EJ, Ottman R, Paolicchi JM, Parent JM, Park K, Poduri A, Scheffer IE, Shellhaas RA, Sherr EH, Shih JJ, Singh R, Sirven J, Smith MC, Sullivan J, Lin Thio L, Venkat A, Vining EP, Von Allmen GK, Weisenberg JL, Widdess-Walsh P, Winawer MR. (2013). De novo mutations in epileptic encephalopathies. *Nature* Sep 12;501(7466):217–221. PMID: PMC3773011.
- *Krumm N, O'Roak BJ, Karakoc E, Mohajeri K, Nelson B, Vives L, Jacquemont S, Munson J, Bernier R, **Eichler EE**. (2013). Transmission disequilibrium of small CNVs in simplex autism. *Am J Hum Genet* Oct 3;93(4):595–606. PMID: PMC3791263.
- Giannuzzi G, Paziienza M, Huddleston J, Antonacci F, Malig M, Vives L, **Eichler EE**, Ventura M. (2013). Hominoid fission of chromosome 14/15 and the role of segmental duplications. *Genome Res* Nov;23(11):1763–1773. PMID: PMC3814877.

*Dumont BL, **Eichler EE**. (2013). Signals of historical interlocus gene conversion in human segmental duplications. *PLOS One* Oct 4;8(10):e75949. PMID: PMC3790853.

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. (2014). Rare-variant extensions of the transmission disequilibrium test: Application to autism exome sequence data. *Am J Hum Genet* Jan 2;94(1):33–46. Epub 2013 Dec 19. PMID: PMC3882934.

Dao P, Numanagic I, Lin YY, Hach F, Karakoc E, Donmez N, Collins C, **Eichler EE**, Sahinalp SC. (2014). ORMAN: optimal resolution of ambiguous RNA-Seq multimappings in the presence of novel isoforms. *Bioinformatics* Mar 1;30(5):644–651. Epub 2013 Oct 15. PMID: 24130305. PMID: N/A.

*Jacquemont S, Coe BP, Hersch M, Duyzend MH, Krumm N, Bergmann S, Beckmann JS, Rosenfeld JA, **Eichler EE**. (2014). A higher mutational burden in females supports a "Female Protective Model" in neurodevelopmental disorders. *Am J Hum Genet* Mar 6;94(3):415–425. PMID: PMC3951938.

*Huddleston J, Ranade S, Malig M, Antonacci F, Chaisson M, Hon L, Sudmant PH, Graves TA, Alkan C, Dennis MY, Wilson RK, Turner SW, Korlach J, **Eichler EE**. (2014). Reconstructing complex regions of genomes using long-read sequencing technology. *Genome Res* Apr;24(4):688–696. PMID: PMC3975067.

Morris DW, Pearson RD, Cormican P, Kenny EM, O'Dushlaine CT, Lemieux Perreault LP, Giannoulatou E, Tropea D, Maher BS, Wormley B, Kelleher E, Fahey C, Molinos I, Bellini S, Pirinen M, Strange A, Freeman C, Thiselton DL, Elves RL, Regan R, Ennis S, Dinan TG, McDonald C, Murphy KC, O'Callaghan E, Waddington JL, Walsh D, O'Donovan M, Grozeva D, Craddock N, Stone J, Scolnick E, Purcell S, Sklar P, Coe B, **Eichler EE**, Ophoff R, Buizer J, Szatkiewicz J, Hultman C, Sullivan P, Gurling H, McQuillin A, St Clair D, Rees E, Kirov G, Walters J, Blackwood D, Johnstone M, Donohoe G; International Schizophrenia Consortium; SGENE+ Consortium, O'Neill FA; Wellcome Trust Case Control Consortium 2, Kendler KS, Gill M, Riley BP, Spencer CC, Corvin A. (2014). An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. *Hum Mol Genet* Jun 15;23(12):3316–3326. PMID: PMC4030770.

Helsmoortel C, Vulto-van Silfhout AT, Coe BP, Vandeweyer G, Rooms L, van den Ende J, Schuurs-Hoeijmakers JH, Marcelis CL, Willemsen MH, Vissers LE, Yntema HG, Bakshi M, Wilson M, Witherspoon KT, Malmgren H, Nordgren A, Annerén G, Fichera M, Bosco P, Romano C, de Vries BB, Kleefstra T, Kooy RF, **Eichler EE**, Van der Aa N. (2014). A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. *Nat Genet* Apr;46(4):380–384. PMID: PMC3990853.

Stong N, Deng Z, Gupta R, Hu S, Paul S, Weiner AK, **Eichler EE**, Graves T, Fronick CC, Courtney L, Wilson RK, Lieberman P, Davuluri RV, Riethman H. (2014). Subtelomeric CTCF and cohesin binding site organization using improved subtelomere assemblies and a novel annotation pipeline. *Genome Res* Jun;24(6):1039–1050. PMID: PMC4032850.

Falchi M, El-Sayed Moustafa JS, Takousis P, Pesce F, Bonnefond A, Andersson-Assarsson JC, Sudmant PH, Dorajoo R, Al-Shafai MN, Bottolo L, Ozdemir E, So HC, Davies RW, Patrice A, Dent R, Mangino M, Hysi PG, Dechaume A, Huyvaert M, Skinner J, Pigeyre M, Caiazzo R, Raverdy V, Vaillant E, Field S, Balkau B, Marre M, Visvikis-Siest S, Weill J, Poulain-Godefroy O, Jacobson P, Sjöström L, Hammond CJ, Deloukas P, Sham PC, McPherson R, Lee J, Tai ES, Sladek R, Carlsson LM, Walley A, **Eichler EE**, Pattou F, Spector TD, Froguel P. (2014). Low copy number of the salivary amylase gene predisposes to obesity. *Nat Genet* May;46(5):492–497. PMID: 24686848. PMID: N/A.

Vulto-van Silfhout AT, Rajamanickam S, Jensik PJ, Vergult S, de Roker N, Newhall KJ, Raghavan R, Reardon SN, Jarrett K, McIntyre T, Bulinski J, Ownby SL, Huggenvik JI, McKnight GS, Rose GM, Cai X, Willaert A, Zweier C, Ende S, de Ligt J, van Bon BW, Lugtenberg D, de Vries PF, Veltman JA, van Bokhoven H, Brunner HG, Rauch A, de Brouwer AP, Carvill GL, Hoischen A, Mefford HC, **Eichler EE**, Vissers LE, Menten B, Collard MW, de Vries BB. (2014). Mutations affecting the SAND domain of DEAF1 cause intellectual Disability with severe speech impairment and behavioral problems. *Am J Hum Genet* May 1;94(5):649–661. PMID: PMC4067565.

Hach F, Sarrafi I, Hormozdiari F, Alkan C, **Eichler EE**, Sahinalp SC. (2014). mrsFAST-Ultra: A compact, SNP-aware mapper for high performance sequencing applications. *Nucleic Acids Res* Jul;42(W1):W494–W500. PMID: PMC4086126.

*Bernier R, Golzio C, Xiong B, Stessman HA, Coe BP, Penn O, Witherspoon K, Gerds 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, Francescato 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**. (2014). Disruptive CHD8 mutations define a subtype of autism early in development. *Cell* Jul 17;158(2):263–276. PMID: PMC4136921.

*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**. (2014). Whole-genome sequencing of individuals from a founder population identifies candidate genes for asthma. *PLOS One* Aug 12;9(8):e104396. PMID: PMC4130548.

Vandeweyer G, Helsmoortel C, Van Dijck A, Vulto-van Silfhout AT, Coe BP, Bernier R, Gerds J, Rooms L, van den Ende J, Bakshi M, Wilson M, Nordgren A, Hendon LG, Abdulrahman OA, Romano C, de Vries BB, Kleefstra T, **Eichler EE**, Van der Aa

- N, Kooy RF. (2014). The transcriptional regulator ADNP links the BAF (SWI/SNF) complexes with autism. *Am J Med Genet C Semin Med Genet* Sep;166(3):315–326. PMID: PMC4195434.
- Lozano R, Hagerman RJ, Duyzend M, Budimirovic DB, **Eichler EE**, Tassone F. (2014). Genomic studies in fragile X premutation carriers. *J Neurodev Disord* 2014;6(1):27. PMID: PMC4147387.
- *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, Geetz J, de Vries BB, Romano C, **Eichler EE**. (2014). Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nat Genet* Oct;46(10):1063–1071. PMID: PMC4177294.
- Lazaridis I, Patterson N, Mittnik A, Renaud G, Mallick S, Kirsanow K, Sudmant PH, Schraiber JG, Castellano S, Lipson M, Berger B, Economou C, Bollongino R, Fu Q, Bos KI, Nordenfelt S, Li H, de Filippo C, Prufer K, Sawyer S, Posth C, Haak W, Hallgren F, Fornander E, Rohland N, Delsate D, Francken M, Guinet JM, Wahl J, Ayodo G, Babiker HA, Bailliet G, Balanovska E, Balanovsky O, Barrantes R, Bedoya G, Ben-Ami H, Bene J, Berrada F, Bravi CM, Brisighelli F, Busby GB, Cali F, Churnosov M, Cole DE, Corach D, Damba L, van Driem G, Dryomov S, Dugoujon JM, Fedorova SA, Gallego Romero I, Gubina M, Hammer M, Henn BM, Hervig T, Hodoglugil U, Jha AR, Karachanak-Yankova S, Khusainova R, Khusnutdinova E, Kittles R, Kivisild T, Klitz W, Kuinskis V, Kushniarevich A, Laredj L, Litvinov S, Loukidis T, Mahley RW, Melegh B, Metspalu E, Molina J, Mountain J, Nakkalajarvi K, Nesheva D, Nyambo T, Osipova L, Parik J, Platonov F, Posukh O, Romano V, Rothhammer F, Rudan I, Ruizbakiev R, Sahakyan H, Sajantila A, Salas A, Starikovskaya EB, Tarekegn A, Toncheva D, Turdikulova S, Uktveryte I, Utevska O, Vasquez R, Villena M, Voevoda M, Winkler CA, Yepiskoposyan L, Zalloua P, Zemanek T, Cooper A, Capelli C, Thomas MG, Ruiz-Linares A, Tishkoff SA, Singh L, Thangaraj K, Vilems R, Comas D, Sukernik R, Metspalu M, Meyer M, **Eichler EE**, Burger J, Slatkin M, Paabo S, Kelso J, Reich D, Krause J. (2014). Ancient human genomes suggest three ancestral populations for present-day Europeans. *Nature* Sep 18;513(7518):409–413. PMID: PMC4170574.
- Deriziotis P, O'Roak BJ, Graham SA, Estruch SB, Dimitropoulou D, Bernier RA, Gerds J, Shendure J, **Eichler EE**, Fisher SE. (2014). De novo TBR1 mutations in sporadic autism disrupt protein functions. *Nat Commun* Sep 18;5:4954. PMID: PMC4212638.
- *Antonacci F, Dennis MY, Huddleston J, Sudmant PH, Steinberg KM, Rosenfeld JA, Miroballo M, Graves TA, Vives L, Malig M, Denman L, Raja A, Stuart A, Tang J, Munson B, Shaffer LG, Amemiya CT, Wilson RK, **Eichler EE**. (2014). Palindromic GOLGA8 core duplicons promote chromosome 15q13.3 microdeletion and evolutionary instability. *Nat Genet* Dec;46(12):1293–1302. PMID: PMC4244265.
- *Iossifov I, O'Roak BJ, Sanders SJ, Ronemus M, Krumm N, Levy D, Stessman HA, Witherspoon KT, Vives L, Patterson KE, Smith JD, Paepfer 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. (2014). The contribution of de novo coding mutations to autism spectrum disorder. *Nature* Nov 13;515(7526):216–221. PMID: PMC4313871.
- *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**. (2014). Recurrent de novo mutations implicate novel genes underlying simplex autism risk. *Nat Commun* Nov 24;5:5595. PMID: PMC4249945.
- Steinberg KM, Schneider VA, Graves-Lindsay TA, Fulton RS, Agarwala R, Huddleston J, Shiryev SA, Morgulis A, Surti U, Warren WC, Church DM, **Eichler EE**, Wilson RK. (2014). Single haplotype assembly of the human genome from a hydatidiform mole. *Genome Res* Dec;24(12):2066–2076. PMID: PMC4248323.
- Watson CT, Steinberg KM, Graves TA, Warren RL, Malig M, Schein J, Wilson RK, Holt RA, **Eichler EE**, Breden F. (2015). Sequencing of the human IG light chain loci from a hydatidiform mole BAC library reveals locus-specific signatures of genetic diversity. *Genes Immun* Jan-Feb;16(1):24–34. Epub 2014 Oct 23. PMID: PMC4304971.
- *Chaisson MJ, Huddleston J, Dennis MY, Sudmant PH, Malig M, Hormozdiari F, Antonacci F, Surti U, Sandstrom R, Boitano M, Landolin JM, Stamatoyannopoulos JA, Hunkapiller MW, Korlach J, **Eichler EE**. (2015). Resolving the complexity of the human genome using single-molecule sequencing. *Nature* Jan 29;517(7536):608–611. Epub 2014 Nov 10. PMID: PMC4317254.
- *Hormozdiari F, Penn O, Borenstein E, **Eichler EE**. (2015). The discovery of integrated gene networks for autism and related disorders. *Genome Res* Jan;25(1):142–154. Epub 2014 Nov 5. PMID: PMC4317170.
- De Roker N, Vergult S, Koolen D, Jacobs E, Hoischen A, Zeesman S, Bang B, Bena F, Bockaert N, Bongers EM, de Ravel T, Devriendt K, Giglio S, Faivre L, Joss S, Maas S, Marle N, Novara F, Nowaczyk MJ, Peeters H, Polstra A, Roelens F, Rosenberg C, Thevenon J, Tumer Z, Vanhauwaert S, Varvagiannis K, Willaert A, Willemsen M, Willems M, Zuffardi O, Coucke P, Speleman F, **Eichler EE**, Kleefstra T, Menten B. (2015). Refinement of the critical 2p25.3 deletion region: The role of MYT1L in intellectual disability and obesity. *Genet Med* Jun;17(6):460–466. Epub 2014 Sep 18. PMID: 25232846. PMID: N/A.
- Pino-Yanes M, Gignoux CR, Galanter JM, Levin AM, Campbell CD, Eng C, Huntsman S, Nishimura KK, Gourraud PA, Mohajeri K, O'Roak BJ, Hu D, Mathias RA, Nguyen EA, Roth LA, Padhukasahasram B, Moreno-Estrada A, Sandoval K, Winkler CA,

Lurmann F, Davis A, Farber HJ, Meade K, Avila PC, Serebrisky D, Chapela R, Ford JG, Lenoir MA, Thyne SM, Brigino-Buenaventura E, Borrell LN, Rodriguez-Cintron W, Sen S, Kumar R, Rodriguez-Santana JR, Bustamante CD, Martinez FD, Raby BA, Weiss ST, Nicolae DL, Ober C, Meyers DA, Bleecker ER, Mack SJ, Hernandez RD, **Eichler EE**, Barnes KC, Williams LK, Torgerson DG, Burchard EG. (2015). Genome-wide association study and admixture mapping reveal new loci associated with total IgE levels in Latinos. *J Allergy Clin Immunol* Jun;135(6):1502–1510. Epub 2014 Dec 6. PMID: PMC4458233.

Mazina V, Gerdt J, Trinh S, Ankenman K, Ward T, Dennis MY, Girirajan S, **Eichler EE**, Bernier R. (2015). Epigenetics of autism-related impairment: Copy number variation and maternal infection. *J Dev Behav Pediatr* Feb-Mar;36(2):61–67. PMID: PMC4318761.

Carlson KD, Sudmant PH, Press MO, **Eichler EE**, Shendure J, Queitsch C. (2015). MIPSTR: a method for multiplex genotyping of germline and somatic STR variation across many individuals. *Genome Res* May;25(5):750–761. PMID: PMC4417122.

Snyder MW, Simmons LE, Kitzman JO, Coe BP, Henson JM, Daza RM, **Eichler EE**, Shendure J, Gammill HS. (2015). Copy-number variation and false positive prenatal aneuploidy screening results. *N Engl J Med* Apr 23;372(17):1639–1645. PMID: PMC4411081.

Xue Y, Prado-Martinez J, Sudmant PH, Narasimhan V, Ayub Q, Szpak M, Frandsen P, Chen Y, Yngvadottir B, Cooper DN, de Manuel M, Hernandez-Rodriguez J, Lobon I, Siegmund HR, Pagani L, Quail MA, Hvilson C, Mudakikwa A, **Eichler EE**, Cranfield MR, Marques-Bonet T, Tyler-Smith C, Scally A. (2015). Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding. *Science* Apr 10;348(6231):242–245. PMID: PMC4668944.

Kloosterman WP, Francioli LC, Hormozdiari F, Marschall T, Hehir-Kwa JY, Abdellaoui A, Lameijer EW, Moed MH, Koval V, Renkens I, van Roosmalen MJ, Arp P, Karssen LC, Coe BP, Handsaker RE, Suchiman ED, Cuppen E, Thung DT, McVey M, Wendl MC, Uitterlinden A, van Duijn CM, Swertz M, Wijmenga C, van Ommen G, Slagboom PE, Boomsma DI, Schonhuth A, **Eichler EE**, de Bakker PI, Ye K, Guryev V. (2015). Characteristics of de novo structural changes in the human genome. *Genome Res* Jun;25(6):792–801. PMID: PMC4448676.

*Krumm N, Turner TN, Baker C, Vives L, Mohajeri K, Witherspoon K, Raja A, Coe BP, Stessman HA, He ZX, Leal SM, Bernier R, **Eichler EE**. (2015). Excess of rare, inherited truncating mutations in autism. *Nat Genet* Jun;47(6):582–588. PMID: PMC4449286.

Houge G, Haesen D, Vissers LE, Mehta S, Parker MJ, Wright M, Vogt J, McKee S, Tolmie JL, Cordeiro N, Kleefstra T, Willemsen MH, Reijnders MR, Berland S, Hayman E, Lahat E, Brilstra EH, van Gassen KL, Zonneveld-Huijssoon E, de Bie CI, Hoischen A, **Eichler EE**, Holdhus R, Steen VM, Doskeland SO, Hurler ME, FitzPatrick DR, Janssens V. (2015). B56-related protein phosphatase 2A dysfunction identified in patients with intellectual disability. *J Clin Invest* Aug 3;125(8):3051–3062. PMID: PMC4623570.

Snijders Blok L, Madsen E, Juusola J, Gilissen C, Baralle D, Reijnders MR, Venselaar H, Helsmoortel C, Cho MT, Hoischen A, Vissers LE, Koemans TS, Wissink-Lindhout W, **Eichler EE**, Romano C, Van Esch H, Stumpel C, Vreeburg M, Smeets E, Oberdorff K, van Bon BW, Shaw M, Geck J, Haan E, Bienek M, Jensen C, Loeys BL, Van Dijk A, Innes AM, Racher H, Vermeer S, Di Donato N, Rump A, Tatton-Brown K, Parker MJ, Henderson A, Lynch SA, Fryer A, Ross A, Vasudevan P, Kini U, Newbury-Ecob R, Chandler K, Male A; DDD Study, Dijkstra S, Schieving J, Giltay J, van Gassen KL, Schuurs-Hoeijmakers J, Tan PL, Padiaditakis I, Haas SA, Retterer K, Reed P, Monaghan KG, Haverfield E, Natowicz M, Myers A, Kruer MC, Stein Q, Strauss KA, Brigatti KW, Keating K, Burton BK, Kim KH, Charrow J, Norman J, Foster-Barber A, Kline AD, Kimball A, Zackai E, Harr M, Fox J, McLaughlin J, Lindstrom K, Haude KM, van Roozendaal K, Brunner H, Chung WK, Kooy RF, Pfundt R, Kalscheuer V, Mehta SG, Katsanis N, Kleefstra T. (2015). Mutations in DDX3X are a common cause of unexplained intellectual disability with gender-specific effects on Wnt signaling. *Am J Hum Genet* Aug 6;97(2):343–352. PMID: PMC4573244.

*Sudmant PH, Mallick S, Nelson BJ, Hormozdiari F, Krumm N, Huddleston J, Coe BP, Baker C, Nordenfelt S, Bamshad M, Jorde LB, Posukh OL, Sahakyan H, Watkins WS, Yepiskoposyan L, Abdullah MS, Bravi CM, Capelli C, Hervig T, Wee JT, Tyler-Smith C, van Driem G, Romero IG, Jha AR, Karachanak-Yankova S, Toncheva D, Comas D, Henn B, Kivisild T, Ruiz-Linares A, Sajantila A, Metspalu E, Parik J, Vilems R, Starikovskaya EB, Ayodo G, Beall CM, Di Rienzo A, Hammer M, Khusainova R, Khusnutdinova E, Klitz W, Winkler C, Labuda D, Metspalu M, Tishkoff SA, Dryomov S, Sukernik R, Patterson N, Reich D, **Eichler EE**. (2015). Global diversity, population stratification, and selection of human copy number variation. *Science* Sep 11;349(6253):aab3761. PMID: PMC4568308.

Mitchell E, Douglas A, Kjaegaard S, Callewaert B, Vanlander A, Janssens S, Lawson Yuen A, Skinner C, Failla P, Alberti A, Avola E, Fichera M, Kibaek M, Digilio MC, Hannibal MC, den Hollander NS, Bizzarri V, Renieri A, Mencarelli MA, Fitzgerald T, Piazzolla S, van Oudenhove E, Romano C, Schwartz C, **Eichler EE**, Slavotinek A, Escobar L, Rajan D, Crolla J, Carter N, Hodge JC, Mefford HC. (2015). Recurrent duplications of 17q12 associated with variable phenotypes. *Am J Med Genet A* Dec;167(12):3038–3045. PMID: 26420380. PMID: N/A.

*Sudmant PH, Rausch T, Gardner EJ, Handsaker RE, Abyzov A, Huddleston J, Zhang Y, Ye K, Jun G, Hsi-Yang Fritz M, Konkel MK, Malhotra A, Stutz AM, Shi X, Paolo Casale F, Chen J, Hormozdiari F, Dayama G, Chen K, Malig M, Chaisson MJ, Walter K, Meiers S, Kashin S, Garrison E, Auton A, Lam HY, Jasmine Mu X, Alkan C, Antaki D, Bae T, Cerveira E, Chines P, Chong Z, Clarke L, Dal E, Ding L, Emery S, Fan X, Gujral M, Kahveci F, Kidd JM, Kong Y, Lameijer EW, McCarthy S, Flicek P, Gibbs RA, Marth G, Mason CE, Menelaou A, Muzny DM, Nelson BJ, Noor A, Parrish NF, Pendleton M, Quitadamo A, Raeder B, Schadt EE, Romanovitch M, Schlattl A, Sebra R, Shabalin AA, Untergasser A, Walker JA, Wang M, Yu F, Zhang C, Zhang J, Zheng-Bradley

- X, Zhou W, Zichner T, Sebat J, Batzer MA, McCarroll SA; The 1000 Genomes Project Consortium, Mills RE, Gerstein MB, Bashir A, Stegle O, Devine SE, Lee C, **Eichler EE**, Korbel JO. (2015). An integrated map of structural variation in 2,504 human genomes. *Nature* Oct 1;526(7571):75–81. PMID: PMC4617611.
- Chen DH, Meneret A, Friedman JR, Korvatska O, Gad A, Bonkowski ES, Stessman HA, Doummar D, Mignot C, Anheim M, Bernes S, Davis MY, Damon-Perriere N, Degos B, Grabli D, Gras D, Hisama FM, Mackenzie KM, Swanson PD, Tranchant C, Vidailhet M, Winesett S, Trouillard O, Amendola LM, Dorschner MO, Weiss M, **Eichler EE**, Torkamani A, Roze E, Bird TD, Raskind WH. (2015). ADCY5-related dyskinesia: Broader spectrum and genotype-phenotype correlations. *Neurology* Dec 8;85(23):2026–2035. PMID: PMC4676753.
- Chen J, Huddleston J, Buckley RM, Malig M, Lawhon SD, Skow LC, Lee MO, **Eichler EE**, Andersson L, Womack JE. (2015). Bovine NK-lysin: Copy number variation and functional diversification. *Proc Natl Acad Sci U S A* Dec 29;112(52):E7223–E7229. PMID: PMC4702975.
- *van Bon BW, Coe BP, Bernier R, Green C, Gerds J, Witherspoon K, Kleefstra T, Willemsen MH, Kumar R, Bosco P, Fichera M, Li D, Amaral D, Cristofoli F, Peeters H, Haan E, Romano C, Mefford HC, Scheffer I, Gecz J, de Vries BB, **Eichler EE**. (2016). Disruptive de novo mutations of DYRK1A lead to a syndromic form of autism and ID. *Mol Psychiatry* Jan;21(1):126–132. doi: 10.1038/mp.2015.5. Epub 2015 Feb 24. PMID: PMC4547916.
- *Duyzend MH, Nuttle X, Coe BP, Baker C, Nickerson DA, Bernier R, **Eichler EE**. (2016). Maternal modifiers and parent-of-origin bias of the autism-associated 16p11.2 CNV. *Am J Hum Genet* Jan 7;98(1):45–57. Epub 2015 Dec 31. PMID: PMC4716684.
- *Turner TN, Hormozdiari F, Duyzend MH, McClymont SA, Hook PW, Iossifov I, Raja A, Baker C, Hoekzema K, Stessman HA, Zody MC, Nelson BJ, Huddleston J, Sandstrom R, Smith JD, Hanna D, Swanson JM, Faustman EM, Bamshad MJ, Stamatoyannopoulos J, Nickerson DA, McCallion AS, Darnell R, **Eichler EE**. (2016). Genome sequencing of autism-affected families reveals disruption of putative noncoding regulatory DNA. *Am J Hum Genet* Jan 7;98(1):58–74. Epub 2015 Dec 31. PMID: PMC4716689.
- Lugtenberg D, Reijnders MR, Fenckova M, Bijlsma EK, Bernier R, van Bon BW, Smeets E, Vulto-van Silfhout AT, Bosch D, **Eichler EE**, Mefford HC, Carvill GL, Bongers EM, Schuurs-Hoeijmakers JH, Ruivenkamp CA, Santen GW, van den Maagdenberg AM, Peeters-Scholte CM, Kuenen S, Verstreken P, Pfundt R, Yntema HG, de Vries PF, Veltman JA, Hoischen A, Gilissen C, de Vries BB, Schenck A, Kleefstra T, Vissers LE. (2016). De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. *Eur J Hum Genet* Aug;24(8):1145–1153. PMID: PMC4970694.
- *Stessman HA, Turner TN, **Eichler EE**. (2016). Molecular subtyping and improved treatment of neurodevelopmental disease. *Genome Med* Feb 25;8(1):22. PMID: PMC4766622.
- Ba W, Yan Y, Reijnders MR, Schuurs-Hoeijmakers JH, Feenstra I, Bongers EM, Bosch DG, de Leeuw N, Pfundt R, Gilissen C, de Vries PF, Veltman JA, Hoischen A, Mefford HC, **Eichler EE**, Eipper BA, Mains RE, Vissers LE, Nadif Kasri N, de Vries BB. (2016). TRIO loss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. *Hum Mol Genet* Mar 1;25(5):892–902. Epub 2015 Dec 31. PMID: PMC4754042.
- *Stessman HA, Willemsen MH, Fenckova M, Penn O, Hoischen A, Xiong B, Wang T, Hoekzema K, Vives L, Vogel I, Brunner HG, van der Burgt I, Ockeloen CW, Schuurs-Hoeijmakers JH, Klein Wassink-Ruiter JS, Stumpel C, Stevens SJ, Vles HS, Marcellis CM, van Bokhoven H, Cantagrel V, Colleaux L, Nicouveau M, Lyonnet S, Bernier RA, Gerds J, Coe BP, Romano C, Alberti A, Grillo L, Scuderi C, Nordenskjöld M, Kvarnang M, Guo H, Xia K, Piton A, Gerard B, Genevieve D, Delobel B, Lehalle D, Perrin L, Prieur F, Thevenon J, Gecz J, Shaw M, Pfundt R, Keren B, Jacqueline A, Schenck A, **Eichler EE**, Kleefstra T. (2016). Disruption of POGZ is associated with intellectual disability and autism spectrum disorders. *Am J Hum Genet* Mar 3;98(3):541–552. PMID: PMC4890241.
- *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**. (2016). Long-read sequence assembly of the gorilla genome. *Science* Apr 1;352(6281):aae0344. PMID: PMC4920363.
- Priest JR, Osoegawa K, Mohammed N, Nanda V, Kundu R, Schultz K, Lammer EJ, Girirajan S, Scheetz T, Waggott D, Haddad F, Reddy S, Bernstein D, Burns T, Steimle JD, Yang XH, Moskowitz IP, Hurler M, Lifton RP, Nickerson D, Bamshad M, **Eichler EE**, Mital S, Sheffield V, Quertermous T, Gelb BD, Portman M, Ashley EA. (2016). De novo and rare variants at multiple loci support the oligogenic origins of atrioventricular septal heart defects. *PLoS Genet* Apr 8;12(4):e1005963. PMID: PMC4825975.
- Koolen DA, Pfundt R, Linda K, Beunders G, Veenstra-Knol HE, Conta JH, Fortuna AM, Gillessen-Kaesbach G, Dugan S, Halbach S, Abdul-Rahman OA, Winesett HM, Chung WK, Dalton M, Dimova PS, Mattina T, Prescott K, Zhang HZ, Saal HM, Hehir-Kwa JY, Willemsen MH, Ockeloen CW, Jongmans MC, Van der Aa N, Failla P, Barone C, Avola E, Brooks AS, Kant SG, Gerkes EH, Firth HV, Ounap K, Bird LM, Masser-Frye D, Friedman JR, Sokunbi MA, Dixit A, Splitt M; DDD Study, Kukulich MK, McGaughan J, Coe BP, Florez J, Nadif Kasri N, Brunner HG, Thompson EM, Gecz J, Romano C, **Eichler EE**, de Vries BB. (2016). The Koolen-de Vries syndrome: A phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. *Eur J Hum Genet* May;24(5):652–9. Epub 2015 Aug 26. PMID: PMC4930086.

Rafati N, Andersson LS, Mikko S, Feng C, Raudsepp T, Pettersson J, Janecka J, Wattle O, Ameer A, Thyreen G, Eberth J, Huddleston J, Malig M, Bailey E, **Eichler EE**, Dalin G, Chowdary B, Andersson L, Lindgren G, Rubin CJ. (2016). Large deletions at the SHOX locus in the pseudoautosomal region are associated with skeletal atavism in Shetland ponies. *G3 (Bethesda)* Jul 7;6(7):2213–2223. PMID: PMC4938674.

Mamiya PC, Richards TL, Coe BP, **Eichler EE**, Kuhl PK. (2016). Brain white matter structure and COMT gene are linked to second-language learning in adults. *Proc Natl Acad Sci U S A* Jun 28;113(26):7249–7254. PMID: PMC4932981.

Shi L, Guo Y, Dong C, Huddleston J, Yang H, Han X, Fu A, Li Q, Li N, Gong S, Lintner KE, Ding Q, Wang Z, Hu J, Wang D, Wang F, Wang L, Lyon GJ, Guan Y, Shen Y, Evgrafov OV, Knowles JA, Thibaud-Nissen F, Schneider V, Yu CY, Zhou L, **Eichler EE**, So KF, Wang K. (2016). Long-read sequencing and de novo assembly of a Chinese genome. *Nat Commun* Jun 30;7:12065. PMID: PMC4931320.

*Nuttall X, Giannuzzi G, Duyzend M, Schraiber JG, Narvaiza I, Sudmant PH, Penn O, Chiatante G, Malig M, Huddleston J, Benner C, Camponeschi F, Ciofi-Baffoni S, Stessman HA, Marchetto MC, Denman L, Harshman L, Baker C, Raja A, Penewit K, Janke N, Tang WJ, Ventura M, Banci L, Antonacci F, Akey JM, Amemiya CT, Gage FH, Reymond A, **Eichler EE**. (2016) Emergence of a Homo sapiens-specific gene family and chromosome 16p11.2 CNV susceptibility. *Nature* Aug 11;536(7615):205–209. PMID: PMC4988886.

Fox K, Johnsen JM, Coe BP, Frazar CD, Reiner AP; NHLBI Exome Sequencing Project, Minority Health-GRID Network, **Eichler EE**, Nickerson DA. (2016). Analysis of exome sequencing data sets reveals structural variation in the coding region of ABO in individuals of African ancestry. *Transfusion* 56:2744–2749. PMC Journal – In Process.

Hehir-Kwa JY, Marschall T, Kloosterman WP, Francioli LC, Baaijens JA, Dijkstra LJ, Abdellaoui A, Koval V, Thung DT, Wardenaar R, Renkens I, Coe BP, Deelen P, de Ligt J, Lameijer EW, van Dijk F, Hormozdiari F; Genome of the Netherlands Consortium., Uitterlinden AG, van Duijn CM, **Eichler EE**, de Bakker PI, Swertz MA, Wijmenga C, van Ommen GB, Slagboom PE, Boomsma DI, Schönhuth A, Ye K, Guryev V. (2016). A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. *Nat Commun* Oct 6;7:12989. PMID: PMC5059695.

*Turner TN, Yi Q, Krumm N, Huddleston J, Hoekzema K, Stessman HAF, Doebley A, Bernier RA, Nickerson DA, **Eichler EE**. (2016). denovo-db: a compendium of human de novo variants. *Nucl Acids Res* Oct 5; doi: 10.1093/nar/gkw865. PMID: PMC5210614.

*Mohajeri K, Cantsilieris S, Huddleston J, Nelson BJ, Coe BP, Campbell CD, Baker C, Harshman L, Munson KM, Kronenberg ZN, Kremitzki M, Raja A, Catacchio CR, Graves TA, Wilson RK, Ventura M, **Eichler EE**. (2016). Interchromosomal core duplicons drive both evolutionary instability and disease susceptibility of the Chromosome 8p23.1 region. *Genome Res* Nov;26(11):1453–1467. PMID: PMC5088589.

*Wang T, Guo H, Xiong B, Stessman HA, Wu H, Coe BP, Turner TN, Liu Y, Zhao W, Hoekzema K, Vives L, Xia L, Tang M, Ou J, Chen B, Shen Y, Xun G, Long M, Lin J, Kronenberg ZN, Peng Y, Bai T, Li H, Ke X, Hu Z, Zhao J, Zou X, Xia K, **Eichler EE**. (2016). De novo genic mutations among a Chinese autism spectrum disorder cohort. *Nat Commun* Nov 8;7:13316. PMID: PMC5105161.

Bramswig NC, Lüdecke HJ, Pettersson M, Albrecht B, Bernier RA, Cremer K, **Eichler EE**, Falkenstein D, Gerds J, Jansen S, Kuechler A, Kvarnung M, Lindstrand A, Nilsson D, Nordgren A, Pfundt R, Spruijt L, Surowy HM, de Vries BB, Wieland T, Engels H, Strom TM, Kleefstra T, Wiczorek D. (2017). Identification of new TRIP12 variants and detailed clinical evaluation of individuals with non-syndromic intellectual disability with or without autism. *Hum Genet* Feb;136(2):179–192. Epub 2016 Nov 15. PMC Journal – In Process.

*Huddleston J, Chaisson MJ, Meltz Steinberg K, Warren W, Hoekzema K, Gordon DS, Graves-Lindsay TA, Munson KM, Kronenberg ZN, Vives L, Peluso P, Boitano M, Chin CS, Korlach J, Wilson RK, **Eichler EE**. (2017). Discovery and genotyping of structural variation from long-read haploid genome sequence data. *Genome Res* May;27(5):677–685. doi: 10.1101/gr.214007.116. Epub 2016 Nov 28. PMC Journal – In Process.

*Dennis MY, Harshman L, Nelson BJ, Penn O, Cantsilieris S, Huddleston J, Antonacci F, Penewit K, Denman L, Raja A, Baker C, Mark K, Malig M, Janke N, Espinoza C, Stessman HAF, Nuttall X, Hoekzema K, Lindsay-Graves TA, Wilson RK, **Eichler EE**. (2017). The evolution and population diversity of human-specific segmental duplications. *Nature Ecology and Evolution* Feb 17;1:69. doi:10.1038/s41559-016-0069. [Epub ahead of print] PMC Journal – In Process.

*Stessman HA, Xiong B, Coe BP, Wang T, Hoekzema K ... (42 authors) ... Xia K, Peeters H, Nordenskjöld M, Schenck A, Bernier RA, **Eichler EE**. (2017). Targeted sequencing identifies 90 neurodevelopmental disorder risk genes with autism and developmental disability biases. *Nat Genet* Apr;49(4):515–526. PMID: PMC5374041.

*Dougherty ML, Nuttall X, Penn O, Nelson BJ, Huddleston J, Baker C, Harshman L, Duyzend MH, Ventura M, Antonacci F, Sandstrom R, Dennis MY, **Eichler EE**. (2017). The birth of a human-specific neural gene by incomplete duplication and gene fusion. *Genome Biol* Mar 9;18(1):49. PMID: PMC5345166.

Kim DS, Burt AA, Ranchalis JE, Wilmot B, Smith JD, Patterson KE, Coe BP, Li YK, Bamshad MJ, Nikolas M, **Eichler EE**, Swanson JM, Nigg JT, Nickerson DA, Jarvik GP; University of Washington Center for Mendelian Genomics. (2017). Sequencing

of sporadic Attention-Deficit Hyperactivity Disorder (ADHD) identifies novel and potentially pathogenic de novo variants and excludes overlap with genes associated with autism spectrum disorder. *Am J Med Genet B Neuropsychiatr Genet* Mar 22. doi: 10.1002/ajmg.b.32527. [Epub ahead of print] PMC Journal – In Process.

Chiatante G, Giannuzzi G, Calabrese FM, **Eichler EE**, Ventura M. (2017). Centromere destiny in dicentric chromosomes: New insights from the evolution of human chromosome 2 ancestral centromeric region. *Mol Biol Evol* Mar 15. doi: 10.1093/molbev/msx108. [Epub ahead of print] PMC Journal – In Process.

Schneider VA, Graves-Lindsay T, Howe K, Bouk N, Chen HC, Kitts PA, Murphy TD, Pruitt KD, Thibaud-Nissen F, Albracht D, Fulton RS, Kremitzki M, Magrini V, Markovic C, McGrath S, Steinberg KM, Auger K, Chow W, Collins J, Harden G, Hubbard T, Pelan S, Simpson JT, Threadgold G, Torrance J, Wood JM, Clarke L, Koren S, Boitano M, Peluso P, Li H, Chin CS, Phillippy AM, Durbin R, Wilson RK, Flicek P, **Eichler EE**, Church DM. (2017). Evaluation of GRCh38 and de novo haploid genome assemblies demonstrates the enduring quality of the reference assembly. *Genome Res* May;27(5):849–864. PMC Journal – In Process.

Xia EH, Zhang HB, Sheng J, Li K, Zhang QJ, Kim C, Zhang Y, Liu Y, Zhu T, Li W, Huang H, Tong Y, Nan H, Shi C, Shi C, Jiang JJ, Mao SY, Jiao JY, Zhang D, Zhao Y, Zhao YJ, Zhang LP, Liu YL, Liu BY, Yu Y, Shao SF, Ni DJ, **Eichler EE**, Gao LZ. (2017). The tea tree genome provides insights into tea flavor and independent evolution of caffeine biosynthesis. *Mol Plant* Apr 29. pii: S1674-2052(17)30103-X. doi: 10.1016/j.molp.2017.04.002. [Epub ahead of print] PMC Journal – In Process.

b) Genome Sequencing Consortium Papers

International Sequencing Consortium. (2001). Initial sequencing and analysis of the human genome. *Nature* Feb;409(6822):860–921. §Contributors Bailey JA, **Eichler EE** to Segmental Duplication section of the manuscript, pages 889–892.

Hillier LW, Fulton RS, Fulton LA, Graves TA, Pepin KH, Wagner-McPherson C, ... (95 authors) ..., McPherson JD, Olson MV, **Eichler EE**, Green ED, Waterston RH, Wilson RK. (2003). The DNA sequence of human chromosome 7. *Nature* Jul;424(6945):157–164.

Istrail S, Sutton GG, Florea L, Halpern AL, Mobarry CM, Lippert R, ... (23 authors) ..., Clark AG, Waterman MS, **Eichler EE**, Adams MD, Hunkapiller MW, Myers EW, Venter JC. (2004). Whole-genome shotgun assembly and comparison of human genome assemblies. *Proc Natl Acad Sci U S A* Feb;101(7):1916–1921.

Grimwood J, Gordon LA, Olsen A, ... (87 authors) ..., **Eichler EE**, Pennacchio LA, Richardson P, Stubbs L, Rokhsar DS, Myers RM, Rubin EM, Lucas SM. (2004). The DNA sequence and biology of human chromosome 19. *Nature* Apr;428(6982):529–535.

Gibbs RA, Weinstock GM, Metzker ML, Muzny DM, Sodergren EJ, ... (94 authors), **Eichler EE**, ... (130 authors), Rat Genome Sequencing Project Consortium. (2004). Genome sequence of the Brown Norway rat yields insights into mammalian evolution. *Nature* Apr;428(6982):493–521

Schmutz J, Martin J, Terry A, Couronne O, Grimwood J, Lowry S, Gordon LA, ... (60 authors) ..., Cheng JF, **Eichler EE**, Olsen A, Pennacchio LA, Rokhsar DS, Richardson P, Lucas SM, Myers RM, Rubin EM. (2004). The DNA sequence and comparative analysis of human chromosome 5. *Nature* Sep;431(7006):268–274.

International Human Genome Sequencing Consortium. (2004). Finishing the euchromatic sequence of the human genome. *Nature* Oct;431(7011):931–45. §Contributors Tüzün E, **Eichler EE** to segmental duplication, heterochromatin and gap analyses sections of the manuscript, pages 938–941.

Martin J, Han C, Gordon LA, Terry A, Prabhakar S, She X, ... (109 authors) ..., Rokhsar DS, **Eichler EE**, Gilna P, Lucas SM, Myers RM, Rubin EM, Pennacchio LA. (2004). The sequence and analysis of duplication-rich human chromosome 16. *Nature* Dec;432(7020):988–994.

International Chicken Genome Sequencing Consortium. (2004). Sequence and comparative analysis of the chicken genome provide unique perspectives on vertebrate evolution. *Nature* Dec;432(7018):695–716. §Contributors Tüzün E, **Eichler EE** to segmental duplication section of the manuscript, pages 708–709.

Hillier LW, Graves TA, Fulton RS, Fulton LA, ... (110 authors) ..., Furey TS, Miller W, **Eichler EE**, Bork P, Suyama M, Torrents D, Waterston RH, Wilson RK. (2005). Generation and annotation of the DNA sequences of human chromosomes 2 and 4. *Nature* Apr 7;434(7034):724–731.

Chimpanzee Sequencing and Analysis Consortium. (2005). Initial sequencing of the chimpanzee genome and comparison with the human genome. *Nature* Sep;437(7055):69–87. §Contributors Tüzün E, Cheng Z, **Eichler EE** to segmental duplication and structural variation analyses of the manuscript, pages 73–75.

Zody MC, Garber M, Sharpe T, Young SK, Rowen L, O'Neill K, Whittaker CA, Kamal M, Chang JL, Cuomo CA, Dewar K, FitzGerald MG, Kodira CD, Madan A, Qin S, Yang X, Abbasi N, Abouelleil A, Arachchi HM, Baradarani L, Birditt B, Bloom S, Bloom T, Borowsky ML, Burke J, Butler J, Cook A, DeArellano K, DeCaprio D, Dorris L 3rd, Dors M, **Eichler EE**, Engels R, Fahey J, Fleetwood P, Friedman C, Gearin G, Hall JL, Hensley G, Johnson E, Jones C, Kamat A, Kaur A, Locke DP, Madan A, Munson G, Jaffe DB, Lui A, Macdonald P, Mauceli E, Naylor JW, Nesbitt R, Nicol R, O'Leary SB, Ratcliffe A, Rounsley S, She X, Sneddon KM, Stewart S, Sougnez C, Stone SM, Topham K, Vincent D, Wang S, Zimmer AR, Birren BW, Hood L, Lander ES,

- Nusbaum C. (2006). Analysis of the DNA sequence and duplication history of human chromosome 15. *Nature* Mar;440(7084):671–675.
- Taylor TD, Noguchi H, Totoki Y, Toyoda A, Kuroki Y, Dewar K, Lloyd C, Itoh T, Takeda T, Kim DW, She X, Barlow KF, Bloom T, Bruford E, Chang JL, Cuomo CA, **Eichler EE**, FitzGerald MG, Jaffe DB, LaButti K, Nicol R, Park HS, Seaman C, Sougnez C, Yang X, Zimmer AR, Zody MC, Birren BW, Nusbaum C, Fujiyama A, Hattori M, Rogers J, Lander ES, Sakaki Y. (2006). Human chromosome 11 DNA sequence and analysis including novel gene identification. *Nature* Mar;440(7083):497–500.
- Rhesus Macaque Genome Sequencing and Analysis Consortium, ... (12 authors), **Eichler EE**, ... (162 authors), Zwiq AS. (2007). Evolutionary and biomedical insights from the rhesus macaque genome. *Science* Apr;316(5822):222–234.
- Warren WC, Hillier LW, Marshall Graves JA, Birney E, Ponting CP, Grutzner F, Belov K, Miller W, Clarke L, Chinwalla AT, Yang SP, Heger A, Locke DP, Miethke P, Waters PD, Veyrunes F, Fulton L, Fulton B, Graves T, Wallis J, Puente XS, Lopez-Otin C, Ordóñez GR, **Eichler EE**, Chen L, Cheng Z, ... (74 authors) ..., Mardis ER, Wilson RK. (2008). Genome analysis of the platypus reveals unique signatures of evolution. *Nature* May 8;453(7192):175–83. Erratum in: Sep 11;455(7210):256. PMID: PMC2803040.
- Bovine Genome Sequencing and Analysis Consortium, Elsik CG, Tellam RL, Worley KC, Gibbs RA, Muzny DM, Weinstock GM, Adelson DL, **Eichler EE**, ... (298 authors), Zhao FQ. (2009). The genome sequence of taurine cattle: A window to ruminant biology and evolution. *Science* Apr 24;324(5926):522–528. PMID: PMC2943200.
- Church DM, Goodstadt L, Hillier LW, Zody MC, Goldstein S, She X, Bult CJ, Agarwala R, Cherry JL, DiCuccio M, Hlavina W, Kapustin Y, Meric P, Maglott D, Birtle Z, Marques AC, Graves T, Zhou S, Teague B, Potamousis K, Churas C, Place M, Herschleb J, Runnheim R, Forrest D, Amos-Landgraf J, Schwartz DC, Cheng Z, Lindblad-Toh K, **Eichler EE**, Ponting CP; Mouse Genome Sequencing Consortium. (2009). Lineage-specific biology revealed by a finished genome assembly of the mouse. *PLOS Biol* May 5;7(5):e1000112. PMID: PMC2680341.
- McKernan KJ, Peckham HE, Costa G, McLaughlin S, Tsung E, Fu Y, Clouser C, Duncan C, Ichikawa J, Lee C, Zhang Z, Sheridan A, Fu H, Ranade S, Dimilanta E, Sokolsky T, Zhang L, Hendrickson C, Li B, Kotler L, Stuart J, Malek J, Manning J, Antipova A, Perez D, Moore M, Hayashibara K, Lyons M, Beaudoin R, Coleman B, Laptewicz M, Sanicandro A, Rhodes M, De La Vega F, Gottimukkala RK, Hyland F, Reese M, Yang S, Bafna V, Bashir A, Macbride A, Aklan C, Kidd JM, **Eichler EE**, Blanchard AP. (2009) Sequence and structural variation in a human genome uncovered by short-read, massively parallel ligation sequencing using two base encoding. *Genome Res* Sep;19(9):1527–1541. PMID: PMC2752135.
- Schuster SC, Miller W, Ratan A, Tomsho LP, Giardine B, Kasson LR, Harris RS, Petersen DC, Zhao F, Qi J, Alkan C, Kidd JM, Sun Y, Drautz DI, Bouffard P, Muzny DM, Reid JG, Nazareth LV, Wang Q, Burhans R, Riemer C, Wittekindt NE, Moorjani P, Tindall EA, Danko CG, Teo WS, Buboltz AM, Zhang Z, Ma Q, Oosthuysen A, Steenkamp AW, Oostuisen H, Venter P, Gajewski J, Zhang Y, Pugh BF, Makova KD, Nekrutenko A, Mardis ER, Patterson N, Pringle TH, Chiaromonte F, Mullikin JC, **Eichler EE**, Hardison RC, Gibbs RA, Harkins TT, Hayes VM. (2010). Complete Khoisan and Bantu genomes from southern Africa. *Nature* Feb 18;463(7283):943–947. PMID: PMC3890430.
- Warren WC, Clayton DF, Ellegren H, Arnold AP, Hillier LW, Kunstner A, Searle S, White S, Vilella AJ, Fairley S, Heger A, Kong L, Ponting CP, Jarvis ED, Mello CV, Minx P, Lovell P, Velho TA, Ferris M, Balakrishnan CN, Sinha S, Blatti C, London SE, Li Y, Lin YC, George J, Sweedler J, Southey B, Gunaratne P, Watson M, Nam K, Backstrom N, Smeds L, Nabholz B, Itoh Y, Whitney O, Pfenning AR, Howard J, Volker M, Skinner BM, Griffin DK, Ye L, McLaren WM, Flicek P, Quesada V, Velasco G, Lopez-Otin C, Puente XS, Olender T, Lancet D, Smit AF, Hubley R, Konkel MK, Walker JA, Batzer MA, Gu W, Pollock DD, Chen L, Cheng Z, **Eichler EE**, Stapley J, Slate J, Ekblom R, Birkhead T, Burke T, Burt D, Scharff C, Adam I, Richard H, Sultan M, Soldatov A, Lehrach H, Edwards SV, Yang SP, Li X, Graves T, Fulton L, Nelson J, Chinwalla A, Hou S, Mardis ER, Wilson RK. (2010). The genome of a songbird. *Nature* Apr 1;464(7289):757–762. PMID: PMC3187626.
- The 1000 Genomes Project Consortium. (2010). A map of human genome variation from population-scale sequencing. *Nature* Oct 28;467(7319):1061–1073. [§]Contributors Aksay G, Alkan C, Hormozdiari F, Kidd JM, Sudmant PH, **Eichler EE** to structural variation analyses of the manuscript, pages 1063–1066. PMID: PMC3042601.
- Mills RE, Walter K, Stewart C, Handsaker RE, Chen K, Alkan C, Abyzov A, Yoon SC, Ye K, Cheetham RK, Chinwalla A, Conrad DF, Fu Y, Grubert F, Hajirasouliha I, Hormozdiari F, Iakoucheva LM, Iqbal Z, Kang S, Kidd JM, Konkel MK, Korn J, Khurana E, Kural D, Lam HY, Leng J, Li R, Li Y, Lin CY, Luo R, Mu XJ, Nemes J, Peckham HE, Rausch T, Scally A, Shi X, Stromberg MP, Stutz AM, Urban AE, Walker JA, Wu J, Zhang Y, Zhang ZD, Batzer MA, Ding L, Marth GT, McVean G, Sebat J, Snyder M, Wang J, Ye K, **Eichler EE**, Gerstein MB, Hurles ME, Lee C, McCarroll SA, Korbel JO; 1000 Genomes Project. (2011). Mapping copy number variation by population-scale genome sequencing. *Nature* Feb 3;470(7332):59–65. PMID: PMC3077050.
- Scally A, Duthel JY, Hillier LW, Jordan GE, Goodhead I, Herrero J, Hobolth A, Lappalainen T, Mailund T, Marques-Bonet T, McCarthy S, Montgomery SH, Schwalie PC, Tang YA, Ward MC, Xue Y, Yngvadottir B, Alkan C, Andersen LN, Ayub Q, Ball EV, Beal K, Bradley BJ, Chen Y, Clee CM, FitzGerald S, Graves TA, Gu Y, Heath P, Heger A, Karakoc E, Kolb-Kokocinski A, Laird GK, Lunter G, Meader S, Mort M, Mullikin JC, Munch K, O'Connor TD, Phillips AD, Prado-Martinez J, Rogers AS, Sajjadian S, Schmidt D, Shaw K, Simpson JT, Stenson PD, Turner DJ, Vigilant L, Vilella AJ, Whitener W, Zhu B, Cooper DN, de Jong P, Dermitzakis ET, **Eichler EE**, Flicek P, Goldman N, Mundy NI, Ning Z, Odom DT, Ponting CP, Quail MA, Ryder OA, Searle SM, Warren WC, Wilson RK, Schierup MH, Rogers J, Tyler-Smith C, Durbin R. (2012). Insights into hominid evolution from the gorilla genome sequence. *Nature* Mar 7;483(7388):169–75. PMID: PMC3303130.

Prüfer K, Munch K, Hellmann I, Akagi K, Miller JR, Walenz B, Koren S, Sutton G, Kodira C, Winer R, Knight JR, Mullikin JC, Meader SJ, Ponting CP, Lunter G, Higashino S, Hobolth A, Dutheil J, Karakoç E, Alkan C, Sajjadian S, Catacchio CR, Ventura M, Marques-Bonet T, **Eichler EE**, André C, Atencia R, Mugisha L, Junhold J, Patterson N, Siebauer M, Good JM, Fischer A, Ptak SE, Lachmann M, Symer DE, Mailund T, Schierup MH, Andrés AM, Kelso J, Pääbo S. (2012). The bonobo genome compared with the chimpanzee and human genomes. *Nature* Jun 28;486(7404):527–531. PMID: PMC3498939.

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, Kuhlwillm 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. (2014). The complete genome sequence of a Neanderthal from the Altai Mountains. *Nature* Jan 2;505(7481):43–49. Epub 2013 Dec 18. PMID: PMC4031459.

Marmoset Genome Sequencing and Analysis Consortium. (2014). The common marmoset genome provides insight into primate biology and evolution. *Nat Genet* Aug;46(8):850–857. PMID: PMC4138798.

Carbone L, Harris RA, Gnerre S, Veeramah KR, Lorente-Galdos B, Huddleston J, Meyer TJ, Herrero J, Roos C, Aken B, Anaclerio F, Archidiacono N, Baker C, Barrell D, Batzer MA, Beal K, Blancher A, Bohrson CL, Brameier M, Campbell MS, Capozzi O, Casola C, Chiatante G, Cree A, Damert A, de Jong PJ, Dumas L, Fernandez-Callejo M, Flicek P, Fuchs NV, Gut I, Gut M, Hahn MW, Hernandez-Rodriguez J, Hillier LW, Hubley R, Ianc B, Izsvak Z, Jablonski NG, Johnstone LM, Karimpour-Fard A, Konkel MK, Kostka D, Lazar NH, Lee SL, Lewis LR, Liu Y, Locke DP, Mallick S, Mendez FL, Muffato M, Nazareth LV, Nevenon KA, O'Blenski M, Ochis C, Odum DT, Pollard KS, Quilez J, Reich D, Rocchi M, Schumann GG, Searle S, Sikela JM, Skollar G, Smit A, Sonmez K, ten Hallers B, Terhune E, Thomas GW, Ullmer B, Ventura M, Walker JA, Wall JD, Walter L, Ward MC, Wheelan SJ, Whelan CW, White S, Wilhelm LJ, Woerner AE, Yandell M, Zhu B, Hammer MF, Marques-Bonet T, **Eichler EE**, Fulton L, Fronick C, Muzny DM, Warren WC, Worley KC, Rogers J, Wilson RK, Gibbs RA. (2014). Gibbon genome and the fast karyotype evolution of small apes. *Nature* Sep 11;513(7517):195–201. PMID: PMC4249732.

Yue F, Cheng Y, Breschi A, Vierstra J, Wu W, Ryba T, Sandstrom R, Ma Z, Davis C, Pope BD, Shen Y, Pervouchine DD, Djebali S, Thurman RE, Kaul R, Rynes E, Kirilusha A, Marinov GK, Williams BA, Trout D, Amrhein H, Fisher-Aylor K, Antoshechkin I, DeSalvo G, See LH, Fastuca M, Drenkow J, Zaleski C, Dobin A, Prieto P, Lagarde J, Bussotti G, Tanzer A, Denas O, Li K, Bender MA, Zhang M, Byron R, Groudine MT, McCleary D, Pham L, Ye Z, Kuan S, Edsall L, Wu YC, Rasmussen MD, Bansal MS, Kellis M, Keller CA, Morrissey CS, Mishra T, Jain D, Dogan N, Harris RS, Cayting P, Kawli T, Boyle AP, Euskirchen G, Kundaje A, Lin S, Lin Y, Jansen C, Malladi VS, Cline MS, Erickson DT, Kirkup VM, Learned K, Sloan CA, Rosenbloom KR, Lacerda de Sousa B, Beal K, Pignatelli M, Flicek P, Lian J, Kahveci T, Lee D, Kent WJ, Ramalho Santos M, Herrero J, Notredame C, Johnson A, Vong S, Lee K, Bates D, Neri F, Diegel M, Canfield T, Sabo PJ, Wilken MS, Reh TA, Giste E, Shafer A, Kutayavin T, Haugen E, Dunn D, Reynolds AP, Neph S, Humbert R, Hansen RS, De Bruijn M, Selli L, Rudensky A, Josefowicz S, Samstein R, **Eichler EE**, Orkin SH, Levasseur D, Papayannopoulou T, Chang KH, Skoultchi A, Gosh S, Distech C, Treuting P, Wang Y, Weiss MJ, Blobel GA, Cao X, Zhong S, Wang T, Good PJ, Lowdon RF, Adams LB, Zhou XQ, Pazin MJ, Feingold EA, Wold B, Taylor J, Mortazavi A, Weissman SM, Stamatoyannopoulos JA, Snyder MP, Guigo R, Gingeras TR, Gilbert DM, Hardison RC, Beer MA, Ren B; Mouse ENCODE Consortium. (2014). A comparative encyclopedia of DNA elements in the mouse genome. *Nature* Nov 20;515(7527):355–364. PMID: PMC4266106.

Vierstra J, Rynes E, Sandstrom R, Zhang M, Canfield T, Hansen RS, Stehling-Sun S, Sabo PJ, Byron R, Humbert R, Thurman RE, Johnson AK, Vong S, Lee K, Bates D, Neri F, Diegel M, Giste E, Haugen E, Dunn D, Wilken MS, Josefowicz S, Samstein R, Chang KH, **Eichler EE**, De Bruijn M, Reh TA, Skoultchi A, Rudensky A, Orkin SH, Papayannopoulou T, Treuting PM, Selli L, Kaul R, Groudine M, Bender MA, Stamatoyannopoulos JA. (2014). Mouse regulatory DNA landscapes reveal global principles of cis-regulatory evolution. *Science* Nov 21;346(6212):1007–1012. PMID: PMC4337786.

Epi4K Consortium. (2016). De novo mutations in SLC1A2 and CACNA1A are important causes of epileptic encephalopathies. *Am J Hum Genet* Aug 4;99(2):287–98. PMID: PMC4974067.

c) Reviews and Book Chapters

***Eichler EE**, Nelson DL. (1998). FRAXA and the fragile X syndrome. In: Rubinsztein DC, Hayden MR, editors. Trinucleotide repeat diseases. Oxford Press. p 11–42.

***Eichler EE**. (1998). Masquerading repeats: Paralogous pitfalls of the human genome. *Genome Res* Aug;8(8):758–762.

***Eichler EE**. (1999). Repetitive conundrums of centromere structure and function. *Hum Mol Genet* Feb;8(2):151–155.

Ji Y, **Eichler EE**, Schwartz S, Nicholls RD. (2000). Structure of chromosomal duplicons and their role in mediating human genomic disorders. *Genome Res* May;10(5):597–610.

*O'Keefe CO, **Eichler EE**. (2000). The pathological consequences and evolutionary implications of recent human genomic duplications. In: Sankoff D, Nadeau JH, editors. Comparative genomics: Empirical and analytical approaches to gene order dynamics, map alignment and the evolution of gene families. New York (NY): Springer. p 29–46.

***Eichler EE**. (2001). Segmental duplications: What's missing, misassigned, and misassembled—and should we care? *Genome Res* May;11(5):653–656.

- *Horvath JE, Bailey JA, Locke DL, **Eichler EE**. (2001). Lessons from the human genome: Transitions between euchromatin and heterochromatin. *Hum Mol Genet* Oct;10(20):2215–2223.
- ***Eichler EE**. (2001). Recent duplication, domain accretion and the dynamic mutation of the human genome. *Trends Genet* Nov;17(11):661–669.
- *Samonte RV, **Eichler EE**. (2002). Segmental duplications and the evolution of the primate genome. *Nat Rev Genet* Jan;3(1):65–72.
- ***Eichler EE**, DeJong PJ. (2002). Biomedical applications and studies of molecular evolution: A proposal for a primate genomic library resource. *Genome Res* May;12(5):673–678.
- Potier M-C, Golfier G, **Eichler EE**. (2002). Chromosome-specific repeats. In: Gardiner K, section editor. *Nature Encyclopaedia of the Human Genome*. London: Nature Publishing Group.
- *Locke DP, Horvath JE, **Eichler EE**. (2003). Mapping pericentromeric regions. In: Dunham I, editor. *Genome mapping and sequencing*. Wymondham (UK): Horizon Scientific Press. p 236–256.
- *Bailey JA, **Eichler EE**. (2003). Genome-wide detection and analysis of segmental duplications within mammalian organisms. *Cold Spring Harb Symp Quant Biol* 68:115–124.
- ***Eichler EE**, Sankoff D. (2003). Structural dynamics of eukaryotic chromosome evolution. *Science* Aug;301(5634):793–797.
- ***Eichler EE**, Patel NH. (2003). Genomes and evolution: From sequence to organism. *Curr Opin Genes Dev* Dec;13(6):559–561.
- ***Eichler EE**, Frazer, KA. (2004). The nature, pattern and function of human sequence variation. *Genome Biol* 5(4):318.
- ***Eichler EE**, Clark RA, She X. (2004). An assessment of the sequence gaps: Unfinished business in a finished human genome. *Nat Rev Genet* May;5(5):345–354.
- Coghlan A, **Eichler EE**, Oliver SG, Paterson AH, Stein L. (2005). Chromosome evolution in eukaryotes: A multi-kingdom perspective. *Trends Genet* Dec;21(12):673–682.
- Samonte RU, **Eichler EE**. (2005). Segmental duplications and the human genome. In: Jorde LB, editor. *Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics*. Chichester: John Wiley & Sons Ltd.
- ***Eichler EE**. (2006). Widening the spectrum of human genetic variation. *Nat Genet* Jan;38(1):9–11.
- *Sharp AJ, **Eichler EE**. (2006). Segmental duplications. In: Stankiewicz P, Lupski JR, editors. *Genomic disorders: The genomic basis of disease*. Totowa (NJ): Humana Press. p 73–88.
- *Sharp AJ, Cheng Z, **Eichler EE**. (2006). Structural variation of the human genome. *Annu Rev Genomics Hum Genet* 7:407–442.
- *Bailey JA, **Eichler EE**. (2006). Primate segmental duplications: Crucibles of evolution, diversity and disease. *Nat Rev Genet* Jul;7(7):552–564.
- Scherer SW, Lee C, Birney E, Altshuler DM, **Eichler EE**, Carter NP, Hurles ME, Feuk L. (2007). Challenges and standards in integrating surveys of structural variation. *Nat Genet* Jul;39(7 Suppl):S7–S15 (27 June 2007).
- *Cooper GM, Nickerson DA, **Eichler EE**. (2007). Mutational and selective effects on copy-number variants in the human genome. *Nat Genet* Jul;39(7 Suppl):S22–S29 (27 June 2007).
- ***Eichler EE**, Zimmerman AW. (2008). A hot spot of genetic instability in autism. *N Engl J Med* Feb 14;358(7):737–739 (Jan 9 2008).
- Varki A, Geschwind DH, **Eichler EE**. (2008). Explaining human uniqueness: Genome interactions with environment, behaviour and culture. *Nat Rev Genet* Oct;9(10):749–763. PMID: PMC2756412.
- *Mefford HC, **Eichler EE**. (2009). Duplication hotspots, rare genomic disorders, and common disease. *Curr Opin Genet Dev* Jun;19(3):196–204. PMID: PMC2746670.
- *Marques-Bonet T, Ryder OA, **Eichler EE**. (2009). Sequencing primate genomes: What have we learned? *Annu Rev Genomics Hum Genet* 10:355–386.
- *Marques-Bonet T, **Eichler EE**. (2009). The evolution of human segmental duplications and the core duplication hypothesis. *Cold Spring Harb Symp Quant Biol* 74:355–362. PMID: PMC4114149.
- *Marques-Bonet T, Girirajan S, **Eichler EE**. (2009). The origins and impact of primate segmental duplications. *Trends Genet* Oct;25(10):443–454. PMID: PMC2847396.

Manolio TA, Collins FS, Cox NJ, Goldstein DB, Hindorf LA, Hunter DJ, McCarthy MI, Ramos EM, Cardon LR, Chakravarti A, Cho JH, Guttmacher AE, Kong A, Kruglyak L, Mardis E, Rotimi CN, Slatkin M, Valle D, Whittemore AS, Boehnke M, Clark AG, **Eichler EE**, Gibson G, Haines JL, Mackay TF, McCarroll SA, Visscher PM. (2009). Finding the missing heritability of complex diseases. *Nature* Oct 8;461(7265):747–753. PMID: PMC2831613.

Miller DT, Adam MP, Aradhya S, Biesecker LG, Brothman AR, Carter NP, Church DM, Crolla JA, **Eichler EE**, Epstein CJ, Faucett WA, Feuk L, Friedman JM, Hamosh A, Jackson L, Kaminsky EB, Kok K, Krantz ID, Kuhn RM, Lee C, Ostell JM, Rosenberg C, Scherer SW, Spinner NB, Stavropoulos DJ, Tepperberg JH, Thorland EC, Vermeesch JR, Waggoner DJ, Watson MS, Martin CL, Ledbetter DH. (2010). Consensus statement: Chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. *Am J Hum Genet*. May 14;86(5):749–764. PMID: PMC2869000.

***Eichler EE**, Flint J, Gibson G, Kong A, Leal SM, Moore JH, Nadeau JH. (2010). Missing heritability and strategies for finding the underlying causes of complex disease. *Nat Rev Genet* Jun;11(6):446–450. PMID: PMC2942068.

*Girirajan S, **Eichler EE**. (2010). Phenotypic variability and genetic susceptibility to genomic disorders. *Hum Mol Genet* Oct 15;19(R2):R176–187. PMID: PMC2953748.

*Bekpen C, Xavier RJ, **Eichler EE**. (2010). Human IRGM gene "to be or not to be". *Semin Immunopathol* Dec;32(4):437–444.

*Alkan C, Coe BP, **Eichler EE**. (2011). Genome structural variation discovery and genotyping. *Nat Rev Genet* May;12(5):363–376. PMID: PMC4108431.

*Girirajan S, Campbell CD, **Eichler EE**. (2011). Human copy number variation and complex genetic disease. *Annu Rev Genet* 45:203–226.

*Girirajan S, **Eichler EE**. (2011). De novo CNVs in bipolar disorder: Recurrent themes or new directions? *Neuron* Dec 22;72(6):885–887.

*Coe BP, Girirajan S, **Eichler EE**. (2012). The genetic variability and commonality of neurodevelopmental disease. *Am J Med Genet C Semin Med Genet* May 15;160C(2):118–129. PMID: PMC4114147.

*Coe BP, Girirajan S, **Eichler EE**. (2012). A genetic model for neurodevelopmental disease. *Curr Opin Neurobiol* Oct;22(5):829–836. PMID: PMC3437230. PMID: PMC3437230.

*Campbell CD, **Eichler EE**. (2013). Properties and rates of germline mutations in humans. *Trends Genet* Oct;29(10):575–584. PMID: PMC3785239.

*Krumm N, O’Roak BJ, Shendure J, **Eichler EE**. (2014). A de novo convergence of autism genetics and molecular neuroscience. *Trends Neurosci* Feb;37(2):95–105. Epub 2013 Dec 30. PMID: PMC4077788.

*Stessman HA, Bernier R, **Eichler EE**. (2014). A genotype-first approach to defining the subtypes of a complex disease. *Cell* Feb 27;156(5):872–877. PMID: PMC4076166.

*Hoischen A, Krumm N, **Eichler EE**. (2014). Prioritization of neurodevelopmental disease genes by discovery of new mutations. *Nat Neurosci* Jun;17(6):764–772. PMID: PMC4077789.

*Nuttall X, Itsara A, Shendure J, **Eichler EE**. (2014). Resolving genomic disorder-associated breakpoints within segmental DNA duplications using massively parallel sequencing. *Nat Protoc* Jun;9(6):1496–1513. PMID: PMC4114152.

*Duyzend MH, **Eichler EE**. (2015). Genotype-first analysis of the 16p11.2 deletion defines a new type of "autism". *Biol Psychiatry* May 1;77(9):769–771. PMID: PMC4657856.

*Chaisson MJ, Wilson RK, **Eichler EE**. (2015). Genetic variation and the de novo assembly of human genomes. *Nat Rev Genet* Nov;16(11):627–640. PMID: PMC4745987.

*Huddleston J, **Eichler EE**. (2016). An incomplete understanding of human genetic variation. *Genetics* Apr;202(4):1251–1254. PMID: PMC4905539.

*Dennis MY, **Eichler EE**. (2016). Human adaptation and evolution by segmental duplication. *Curr Opin Genet Dev* Dec;41:44–52. PMID: PMC5161654.

*Cantsilieris S, Stessman HA, Shendure J, **Eichler EE**. (2017). Targeted capture and high-throughput sequencing using molecular inversion probes (MIPs). *Methods Mol Biol* 1492:95–106. PMC Journal – In Process.

d) Whitepapers

Eichler EE. (2001). Proposal for BAC library construction of Orangutan (*Pongo pygmaeus*).
<http://www.genome.gov/Pages/Research/Sequencing/BACLibrary/orangutanBornean.pdf>

Eichler EE. (2002). Proposal for construction of a primate BAC library resource.
<http://www.genome.gov/Pages/Research/Sequencing/BACLibrary/primateProposal.pdf>

Olson MV, **Eichler EE**, Varki A, Myers RM, Erwin JE, McConkey EH. (2004). A whitepaper advocating complete sequencing of the genome of the common chimpanzee, *Pan troglodytes*.
http://www.genome.gov/Pages/Research/Sequencing/SeqProposals/Chimp_Genome1_editted.pdf

Waterston RH, **Eichler EE**, Gibbs RA, Green ED, Haussler DH, Lander ES, McKnight S, O'Brien S, Olson MV, Rogers JA, Strausberg R. (2004). A modified version of the proposal from the working group on annotating the human genome.
<http://www.genome.gov/Pages/Research/Sequencing/NewGenSeqTargets/Summaries/AHGProposal.pdf>

Mansfield K, Tardiff S, **Eichler EE.** (2005). White paper for complete sequencing of the common marmoset (*Callithrix jacchus*) genome.
<http://www.genome.gov/Pages/Research/Sequencing/SeqProposals/MarmosetSeq.pdf>

Eichler EE, Altshuler, D, Nickerson, DA and members of the medical working sequencing group. (2006). Human Genome Structural Variation. <http://www.genome.gov/Pages/Research/Sequencing/SeqProposals/StructuralVariationproject.pdf>

Eichler EE, Nickerson DA, Altshuler D, Bowcock AM, Brooks LD, Carter NP, Church DM, Felsenfeld A, Guyer M, Lee C, Lupski JR, Mullikin JC, Pritchard JK, Sebat J, Sherry ST, Smith D, Valle D, Waterston RH. (2007). Completing the map of human genetic variation. *Nature* May;447(7141):161–165 (10 May 2007).