CURRICULUM VITAE

Evan Eugene Eichler
Professor
Howard Hughes Medical Institute
University of Washington
Genome Sciences, Box 355065
Seattle, WA 98195-5065
(206) 543-9526
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

2021–Present	Director (interim) of the Northwest Genomics Center (NWGC) University of Washington (UW) School of Medicine, Seattle, WA
2008–Present	Professor (with tenure) Department of Genome Sciences, University of Washington (UW), Seattle, WA
2005–Present	Howard Hughes Medical Institute Investigator (HHMI)
2020–Present	Member, The Brotman Baty Institute (BBI)
2015–Present	Associate Member, New York Genome Center (NYGC), New York City, NY
2004–2018	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

2022	Falling Walls Science Breakthroughs of the Year 2022
2022	TIME100: The Most Influential People of 2022
2018	National Academy of Medicine (NAM; Elected 2017)
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 (ESHG), 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

2020–Present	Advisory Board of *Cell*
2009–Present	Editorial Board of *Molecular Autism*
2004–2012	Academic Editor, *Public Library of Science (PLOS)*
2003	Section Editor, *Curr Opin Genet Dev*, Genomes and Evolution Special Issue
2002–Present	Editor of *Genome Research*
2002–2004	Editorial Board of *American Journal of Human Genetics*
2002–2007	Editorial Board of *DNA Sequence*
2002–2009	Editorial Board of *BMC Genomics*
1999–2002	Editorial Board of *Genome Research*

b) Scientific Advisory Boards (SABs)

2021–Present	International FOXP1 Foundation SAB
2021–Present	BioVariant, Inc. SAB
2013–2015	New York Genome Center (NYGC) SAB
2012–2020	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 (*ad hoc)*, Genome Center North Carolina, UNC Chapel Hill
2004	Member, SAB (*ad hoc*), Regulatory Genetics and GRAND Project, Genome Center, McGill University, Montreal (Director: Tom Hudson)
2003	Member, SAB (*ad hoc*), Department of Evolutionary Genetics, Max Planck Institute for Molecular Anthropology, Leipzig (Director: Svante Pääbo)

c) International

2021–Present	International SAB (Fachbeirat), Max Planck Institute for Psycholinguistics, Nijmegen, Netherlands
2021–Present	Member, Consortium for Long Read Sequencing (CoLoRS) database
2020–Present	Co-chair, Telomere-to-Telomere (T2T) Sequencing Consortium (primate) (with Adam Phillippy & Kateryna Makova)
2019–Present	Co-chair, Telomere-to-Telomere (T2T) Sequencing Consortium (human) (with Adam Philippy & Karen Miga)
2017–2018	Organizer, Keystone Symposium: "Mobile Genetic Elements and Genome Plasticity"
2015–2020	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
2014–Present	Co-Chair, Human Genome Structural Variation Consortium (HGSVC)
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
2008–2014	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

2021–Present Member Executive Committee, TOPMed
2021–Present Co-Chair, *All of Us* Working Group on Application of Long-read Sequencing Data
2021–Present Member, Steering Committee of the NIH GREGoR Consortium
2021–Present Member, Simons Foundation Autism Research Initiative (SFARI) Review Panel
2020–Present Member/PI, Steering Committee of the Human Pangenome Reference Consortium (HPRC)
2018–2021 Member, *All of Us* Working Group on Application of Long-read Sequencing Data
2018–2020 Member, Center for Mendelian Genomics (CMG UW)
2016–2020 Member, NIH/NHGRI CCDG Steering Committee
2015–2020 Member, Center for Common Disease Genomics (CCDG), Neuropsychiatric Working Group
2015–2023 Member, External Advisory Committee, MIND Institute IDDRC, UC Davis
2015 Chair, External Advisory Committee, Department of Human Genetics, University of Michigan
2014–2018 NIH/NHGRI Study Section, Genome Research Review Committee, GNOM-G
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

2021–Present Director, Northwest Genomics Center (NWGC)
2022–2023 Member, Director of NWGC Search Committee, Genome Sciences, UW
2020–2021 Chair, Genome Sciences 20-Year Anniversary Symposium

2018–Present	Member, Information Technology Advisory Committee, Genome Sciences, UW
2017–2022	PI, Interdisciplinary Training in Genome Sciences (Genome Training Grant: GTG; 2 T32 HG000035)
2017	Member, University of Washington Brain Health Solutions Initiative
2016–Present	Co-chair, Pediatric Mental Health Initiative, UW Medicine (with Emily Neuhaus)
2016–2017	Chair, Genome Sciences Faculty Search Committee (with Debbie Nickerson)
2016–2017	Member, Department of Genome Sciences Seminar Series Organizing Committee
2014–2017	Co-PI, Interdisciplinary Training in Genome Sciences (Genome Training Grant: GTG; 2 T32 HG000035)
2014–Present	UW Medical School Training Program (MSTP) Admissions Committee
2012–2013	Chair, Department of Genome Sciences Seminar Organizing Committee
2011–2021	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, Interdisciplinary Training in Genome Sciences 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
Science
Cell
New England Journal of Medicine
Nature Genetics
Nature Biotechnology
Nature Medicine
Genome Research
Nature Review Genetics
Genes and Development
Nature Methods
Neuron
Trends in Genetics
American Journal of Human Genetics
Human Molecular Genetics
Genetic Epidemiology
Chromosoma
Journal of Medical Genetics
PLOS Computational Biology
American Journal of Psychiatry
Molecular Endocrinology
Genes Chromosomes and Cancer
BMC Genomics
European Journal of Human Genetics
Genomics
Mammalian Genome
Neurogenetics
Human Genetics

EMBO Journal
Current Opinion Genetics and Development
PLOS Genetics
Proceedings of the National Academy of Sciences
Genome Biology
PLOS Biology
Science Translational Medicine
Nucleic Acids Research
Journal of Molecular Evolution
Gene
Molecular Phylogenetics and Evolution
Mutation Research
Molecular Autism
Cytogenetics and Cell Genetics
Journal of Molecular Genetics
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 (19 students—5 current and 14 graduated)

2022–Present Elizabeth (Lizzie) Plender, UW, predoctoral candidate

2021–Present Taylor Real, UW, predoctoral candidate

2020–Present Francis (Xavi) Guitart, UW, predoctoral candidate, advanced to candidacy August 2021.

2019–Present Michelle Noyes, UW, predoctoral candidate, advanced to candidacy September 2020.

2018–Present Philip Dishuck, UW, predoctoral candidate, advanced to candidacy September 2019.

2017–2021 Mitchell Vollger, UW, doctoral candidate, advanced to candidacy July 2018, graduated March 2021. Thesis: Assembly of segmental duplications and their variation in humans. Current: Postdoctoral Fellow, Stergachis Lab, UW, Seattle, WA

2014–2019 Madeleine Geisheker, MSTP, UW, doctoral candidate, advanced to candidacy September 2016, graduated May 2019. Thesis: De novo missense mutations in neurodevelopmental disorders. Current: Resident, Oregon Health & Science University, Portland, OR

2014–2018 Max Dougherty, MSTP, UW, doctoral candidate, advanced to candidacy December 2016, graduated May 2018. Thesis: Transcription of human-specific duplicate genes. Current: Resident, Internal Medicine Research Track, Icahn SOM at Mount Sinai, Manhattan, NY

2011–2016 Michael Duyzend, MSTP, UW, advanced to candidacy August 2013, graduated June 2016; M.D. 2017 UW. Thesis: Understanding the genetic basis of phenotype variability in individuals with neurocognitive disorders. Current: Resident Physician, Boston Children's Hospital, Boston, MA

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, MA

2010–2014 Niklas (Nik) Krumm, MSTP, UW, advanced to candidacy July 2012, graduated June 2014; M.D. 2017 UW. Thesis: Discovery and convergence of inherited mutations in autism spectrum disorder. Current: Assistant Professor, Laboratory Medicine and Pathology, UW, Seattle, WA

2009–2013 Peter Sudmant, UW, advanced to candidacy August 2010, graduated September 2013. Thesis: Evolution and diversity of hominid genomes. Current: Assistant Professor, Department of Integrative Biology, University of California, Berkeley, CA

2007–2011 Andrew (Andy) Itsara, MSTP, UW, advanced to candidacy May 2009, graduated May 2011, M.D. 2012 UW. Thesis: Detection and characterization of human copy-number variation. Past: Hospitalist, Seattle Cancer Care Alliance, Seattle, WA; Clinical Fellow, Hematology-Oncology. Current: Staff Clinician, Hematology Branch, NHLBI, NIH, Bethesda, MD

2006–2010 Jeffrey Kidd, UW, advanced to candidacy June 2007, graduated January 2010. Thesis: Mapping and sequencing human genomic structural variation. Current: Associate Professor (tenure-track), Department of Human Genetics & Department of Computational Medicine and Biology, University of Michigan, Ann Arbor, MI

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.; Associate Director, Bioinformatics, Gilead Sciences. Current: VP of Target Discovery, BioMap, San Francisco, CA

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: Technical Director, Center for Spatial and Functional Genomics, Children's Hospital of Philadelphia, PA

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.; SVP & General Manager, BioPharma at Seven Bridges Genomics, Cambridge. Current: Senior Director, Franchise Development at Foundation Medicine, Boston, MA

1999–2002 Jeffrey Bailey, advanced to candidacy December 1997 (Genetics), joined laboratory December 1999 from Chakravarti lab, graduated April 2002; M.D. 2005 CWRU. Thesis: Genome-wide analysis and detection of segmental duplications. Past: Assistant Professor of Medicine and Physician (Transfusion Medicine), University of Massachusetts Medical School, Worcester. Current: Mencoff Family Associate Professor of Translational Research, Associate Professor of Pathology and Laboratory Medicine, Brown University, Providence, RI

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, NC

b) Postdoctoral Fellows/Research Associates (50 postdocs: 4 current; 28 tenured/tenure-track faculty; 18 hold positions in industry; remainder clinicians, research associates, faculty instructors or scientific writers)

2022–Present Yang (Kate) Sui, Ph.D., postdoctoral research: Autism genetic variant discovery by long-read sequencing

2022–Present DongAhn Yoo, Ph.D., postdoctoral research: Telomere-to-telomere assembly of nonhuman primate genomes

2022–Present Francesco Kumara Mastrorosa, Ph.D., postdoctoral research: Long-read sequencing for the study of pathologic variation in Mendelian disorders

2022–2022 Hyeonsoo Jeong, Ph.D., postdoctoral research: Long-read functional characterization of duplicated genes. Current: Computational Scientist, Altos Labs, San Francisco, CA

2021–2022 Peiyao Zhao, Ph.D., postdoctoral research: Integrative analyses of genetic and epigenetic contributions to autism aetiology.

2019–2022 Danny Miller, M.D, Ph.D., postdoctoral research: Targeted long-read sequencing of clinical samples. Current: Assistant Professor, University of Washington, Seattle, WA

2019–2022 Yafei Mao, Ph.D., postdoctoral research: Primate structural variation evolution. Current: Associate Professor, Shanghai Jiao Tong University, Shanghai, China

2018–2021 David Porubsky, Ph.D., postdoctoral research: Great ape inversions and genetic diversity. Current: Acting Instructor, UW, Seattle, WA

2018–2021 Tzu-Hsueh (Stella) Huang, Ph.D., postdoctoral research: Recent human segmental duplication evolution and autism etiology through interlocus gene conversion discovery.

2018–Present Glennis Logsdon, Ph.D., postdoctoral research: Sequence, assembly, and variation of centromeric regions of the human genome.

2018–2021 Madelyn Gillentine, Ph.D., postdoctoral research: Modeling neurodevelopmental disorder candidate genes in human cells. Current: Lab Variant Scientist, Seattle Children's Hospital, Seattle, WA

2017–2022 Tianyun Wang, Ph.D., postdoctoral research: Targeted sequencing of autism risk candidate genes. Current: Assistant Professor (tenure-track), Department of Medical Genetics, Peking University, Beijing, China

2017–2020 Amy Wilfert, Ph.D., postdoctoral research: Identifying genetic drivers underlying the female protective effect and inherited autism. Current: Bioinformatics Scientist II, Guardant Health, Seattle, WA

2017–2020 Arvis Sulovari, Ph.D., postdoctoral research: Integrated discovery of dosage sensitivity genes in neurodevelopmental disorders. Current: Computational Genetics Lead (Senior Scientist), Cajal Neuroscience Inc., Seattle, WA

2017–2018 Hui Guo, Ph.D., postdoctoral research: Genetics of autism. Current: Associate Professor, Central South University, Changsha, China

2017–2017 Davide Risso, Ph.D., postdoctoral research: Characterization of the function of *Homo sapiens*-specific gene families. Current: Senior Research Scientist, Global Nutrition, Tate & Lyle, Torino, Piedmont, Italy

2016–2022 PingHsun Hsieh, Ph.D., postdoctoral research: Paralogous copy number variation and disease association. Current: Assistant Professor of Department of Genetics, Cell Biology, and Development at University of Minnesota Medical School, Minneapolis, MN

2015–2018 Jason Underwood, Ph.D., postdoctoral research: Long-read transcript sequencing. Current: Pacific Biosciences, Inc.

2015–2016 Chris Hill, Ph.D., postdoctoral research: Sequence and assembly of complex genomes using SMRT sequencing. Current: Staff Software Engineer, DataBricks, Seattle, WA

2015–2017 Zev Kronenberg, Ph.D., postdoctoral research: Disease association and positive selection of structural variation. Past: Senior Computational Biologist, Phase Genomics Inc., Seattle, WA. Current: Manager, Bioinformatics Engineering, Pacific Biosciences, Inc.

2014–2018 Stuart Cantsilieris, Ph.D., postdoctoral research: Structural diversity of duplicated immune response genes and disease association. Past: Research Scholar, Centre for Eye Research Australia, Royal Victorian Eye and Ear Hospital. Current: Senior Project Manager, Garvan Institute of Medical Research, Melbourne, Australia

2014–2019 Tychele Turner, Ph.D., postdoctoral research: Characterization of autism genetic risk factors. Current: Assistant Professor, Department of Genetics, Washington University School of Medicine, St. Louis, MO

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

2012–2017 Mark Chaisson, Ph.D., postdoctoral research: *De novo* assembly of next-generation sequencing data and structural variation detection. Current: Assistant Professor, University of Southern California, Los Angeles, CA

2012–2017 Osnat Penn, Ph.D., postdoctoral research: Gene expression analysis of recently duplicated genes. Past: Scientist II, Modeling, Analysis and Theory group, Allen Institute for Brain Science, Seattle, WA; Senior Bioinformatician Scientist, MyHeritage, Or Yehuda, Israel. Current: Head of Bioinformatics, RNA Therapeutics, Dexcel Pharma, Israel

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, Department of Biochemistry and Molecular Medicine; M.I.N.D. Institute, UC Davis Genome Center, CA

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, CA

2010–2018 Bradley Coe, Ph.D., Canadian Institutes of Health Research (CIHR) Fellow: Development of a morbidity map for copy number variation in neurocognitive disorders. Past: Acting Instructor, Department of Genome Sciences, University of Washington. Current: Clinical Assistant Professor, The University of British Columbia & Laboratory Scientist, Pathology Department Genome Diagnostics Lab, BC Children's and Women's Hospital and Health Centre, Vancouver, BC

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, ME

2009–2012 Karyn Meltz Steinberg, Ph.D., National Research Service Award (NRSA) Fellow: Exploring regions of extreme diversity in the human genome. Past: Staff Scientist, The Genome Institute at Washington University, St. Louis. Current: Assistant Technical Director of Clinical Development at GeneDx, St. Louis, MO

2009–2013 Emre Karakoc, Ph.D., postdoctoral research: Computational methods for characterization of genome and exome structural variation. Past: Assistant Professor, School of Engineering & Natural Sciences, Istanbul Medipol University, Turkey. Current: Principal Bioinformatician, Wellcome Sanger Institute, Hinxton, Cambridge, UK

2009–2013 Brian O'Roak, Ph.D., postdoctoral research: Next-generation sequencing approaches to gene discovery in autism spectrum disorders. Current: Associate Professor, Department of Molecular & Medical Genetics, Oregon Health & Sciences University, Portland, OR

2008–2013 Catarina (Katie) Campbell, Ph.D., National Research Service Award (NRSA) fellow: High-throughput genotyping of structural variants. Current: Director, Data Science, Novartis Institutes for BioMedical Research (NIBR), Boston, MA

2008–2012 Santhosh Girirajan, Ph.D., postdoctoral research: Mechanisms and implications of large-scale genome rearrangements. Current: Associate Professor (tenure-track), Department of Biochemistry and Molecular Biology & Department of Anthropology, Pennsylvania (Penn) State University, University Park, PA

2007–2012 Francesca Antonacci, Ph.D., postdoctoral research: Discovery and characterization of chromosomal inversions as common variants in the human genome. Current: Associate Professor, Department of Biology, University of Bari, Italy

2007–2011 Jeramiah J. Smith, Ph.D. (jointly supervised w/ Dr. Chris T. Amemiya), postdoctoral research: Developmentally programmed rearrangement of the lamprey genome. Current: Associate Professor, University of Kentucky, Lexington, KY

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, & Associate Professor Adjunct Faculty, Department of Genetics, University of Alabama at Birmingham, AL

2006–2009 Cemali Bekpen, Ph.D., HHMI Fellow: Functional characterization of Morpheus gene family. Past: Postdoc, Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Biology, Plön, Germany. Current: Assistant Professor, Department of Molecular Biology and Genetics, Bahçeşehir University, Istanbul, Turkey

2006–2008 Heather Mefford, M.D., Ph.D., Burroughs-Wellcome Scientist and Medical Genetics Fellow: Duplication-mediated rearrangement within fetal demise. Past: Associate Professor & Attending Physician, Department of Pediatrics, UW School of Medicine & Seattle Children's Hospital, Seattle, WA. Current: Faculty Member, St. Jude Children's Research Hospital, Memphis, TN

2005–2011 Can Alkan, Ph.D., HHMI Fellow: Development of mapping algorithms for next-generation sequence data. Current: Associate Professor (tenure-track), Department of Computer Engineering & PI, Lab for Bioinformatics & Computational Genomics, 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.; Business Owner/VP, Amplicon Consulting LLC & Director of R&D, Iverson Genetics; Senior Director, Clinical Operations & Development, Adaptive Biotechnologies Corp., Seattle, WA. Current: VP, Product Development, Oncology, Natera

2003–2007 Andrew Sharp, Ph.D., Rosetta Postdoctoral Fellow: Detection of segmental aneusomy in duplicated DNA. Current: Professor (w/ tenure), Genetics and Genomic Sciences, Mt. Sinai School of Medicine, New York City, NY

2002–2006 Xinwei She, Ph.D., Rosetta Postdoctoral Fellow: Computational analysis of segmental duplications. Past: Senior Computational Scientist, Merck; Principal Scientist/Bioinformatics Team Leader, Functional Genomics, Constellation Pharmaceuticals. Current: Director, Data Science at Parthenon Therapeutics, Boston, MA

2002–2004 Audrey Lynn, Ph.D. (jointly supervised w/ Dr. Terry Hassold), postdoctoral research: Genetic and physical correlation of recombination. Last known position: Project Coordinator, Department of Family Medicine, CWRU, Cleveland, OH

2001–2002 Vicky Choi, Ph.D., PMMB Fellow: Computational methods for sequence assembly of duplicated regions within the human genome. Last known position: Assistant Professor, Department of Computer Science, Virginia Tech, Blacksburg, VA

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; Last known position: Lab Director, PreventionGenetics, Marshfield, WI

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; Medical Writer, Cleveland HeartLab, Inc. Current: Industry Analyst at The Freedonia Group, Cleveland, OH

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: Assistant Professor, Center for Biomolecular Science and Engineering, University of California, Santa Cruz, CA

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, CA

d) Undergraduates

2022 James (Cy) Chittenden, B.S., Neuroscience with a Minor in Biology, The University of Chicago
2022 Andrew Bauer, B.S., Molecular, Cellular and Developmental Biology, UW
2019–2020 Di Lu, B.S., Molecular, Cellular and Developmental Biology, UW
2019 Caitlin Johnson, B.S., Biology, University of California, San Diego
2019 Yashi Singh, summer intern, Interlake High School, Bellevue, WA
2019 Nicholas (Nick) Rose, B.S., Molecular, Cellular, & Developmental Biology, UW
2018–2019 Ruiyang (Rick) Li, B.S., Biology, UW
2016, 2017 Idara Akpandak, B.S., Biology, University of Maryland
2016–2018 Naheed Arang, B.S., Microbiology & B.A., Integrated Science, UW
2015–2016 AnneMarie Welch, B.S., Microbiology, UW
2015–2017 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

2023 Joris Vermeesch, Laboratory of Cytogenetics and Genome Research, University of Leuven, Belgium
2018 A. Bernardo Carvalho, Universidade Federal do Rio de Janeiro, Brazil
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

2018 Davide Vecchio, Sapienza University of Rome, Italy
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 Giannuzzi, University of Bari, Italy
2008	Karen Buysse, Ghent University Hospital, Belgium

g) Thesis Committees (*Chair)

2020–Present	Robin Aguilar	Genome Sciences, UW	Advisor: Brian Beliveau
2019–2022	Michael Goldberg	Genome Sciences, UW	Advisor: Kelley Harris
2018–2019	John E. Lazar	Genome Sciences, UW	Advisor: John Stamatoyannopoulos
2016–2022	Alberto Rivera	Genome Sciences, UW	Advisor: Willie Swanson
2016–2019	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 Chakrravarti
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

2019	Esko A. Kautto	The Ohio State University	Advisor: Richard K. Wilson
2010	Andrés Ingason	University of Copenhagen	Advisor: Thomas Werge
2010	Anna Wetterbom	Uppsala University	Advisor: Ulf Gyllensten
2007	Tomas Marques-Bonet	Unversity of Pompua-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: 532 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, Gordon Research Conference: Molecular Cytogenetics, 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, SMBE, 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 (ESHG) 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 Autonoma of Barcelona, Barcelona, Spain, November

2007

- Invited Lecture, Pompua-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, Anna 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 Hereditable 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, 2012 SFARI Annual Meeting (Simons Foundation), Washington, DC, November
- 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 Speaker, 2014 SSC Whole Exome Sequencing Project Analysis Meeting (Simons Foundation), New York, NY, Jan.
- 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 (IDDRC) 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, Human, 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, AlCoB 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
- Invited Seminar Speaker, UCSF Biomedical Sciences (BMS) Seminar Series, San Francisco, CA, April
- Invited Speaker, Clinical Genomics and NGS Course, Bertinoro, Italy, May
- Invited Keynote, 9th International Conference on Canine and Feline Genetics and Genomics, St. Paul, MN, May
- Invited Speaker, MRC: Centre for Neurodevelopmental Disorders Inaugural Symposium: The Developing Brain in Health and Disease, London, UK, June
- Invited Closing Keynote, 11th European Cytogenetics Association (ECA) Conference, Florence, Italy, July
- Invited Speaker, University of Bari Department of Biology-Genetics, Bari, Italy, July
- Invited Speaker, Gordon Research Conference: Human Genetics & Genomics Conference, Stowe, VT, July
- Invited Speaker, Gordon Research Conference: Lung Development, Injury & Repair, New London, NH, August
- Invited Session Speaker, AGBT 2017 Precision Health Meeting, Scottsdale, AZ, September
- Invited Session Speaker, FENS: Federation of European Neuroscience Societies, The Brain Conference on Cortex Evolution and Development, Copenhagen, Denmark, September
- Invited Speaker, CARTA Symposium: Cellular and Molecular Explorations of Anthropogeny, La Jolla, CA, September
- Invited Speaker, Institute for Systems Genetics Inaugural Symposium at New York University (NYU) Langone Medical Center, New York, NY, October
- Invited Speaker, 18th International Fragile X & Related Neurodevelopmental Disorders Workshop, Quebec, Canada, October
- Invited Session Speaker, 67th ASHG Annual Meeting, Orlando, FL, October
- Invited Talk, Eli and Edythe Broad Center of Regeneration Medicine and Stem Cell Research, UCSF - University of California, San Francisco, CA, October
- Invited Speaker (two talks), The USERN Congress 2017, Kharkiv National Medical University, Kharkiv, Ukraine, November
- Invited Speaker, Carl Friedrich von Siemens Foundation, Munich, Germany, December

2018

- Invited Faculty, Workshop on Genomics Application, Český Krumlov, Czech Republic, January
- Invited Speaker, Genome 10K Workshop: Ordinal Level Vertebrate Genomes Project (VGP), San Diego, CA, January
- Invited Keynote, NYGC's 5th Human Genetics in NYC Conference, New York City, NY, February
- Speaker & Organizer, Keystone Symposium: Mobile Genetic Elements and Genome Plasticity, Sante Fe, NM, February
- Lecturer, University of Pittsburgh School of Medicine, Department of Pathology, Aron E. Szulman Memorial Lecture, Pittsburgh, PA, March
- Invited Seminar Speaker, University of Michigan, Department of Biostatistics, Ann Arbor, MI, April
- Invited Speaker, 2018 SFARI Science Meeting (Simons Foundation), New York, NY, April
- Invited Speaker, 13th International Meeting on Genetics of Neurodevelopmental Disorders, Troina, Italy, April
- Invited Speaker, University of Minnesota Seminar for Biochemistry, Molecular Biology and Biophysics (BMBB), Minneapolis, MN, April
- Invited Speaker, Clinical Genomics and NGS Course, Bertinoro, Italy, April
- Invited Speaker, Pasteur Institute, Paris, France, May
- Invited Speaker, 3rd GENMED Workshop on Medical Genomics, Paris, France, May
- Invited Speaker, Gordon Research Conference: Human Genetic Variation and Disease, Biddeford, ME, June
- Invited Speaker, Teratology Society's 58th Annual Meeting: Mechanisms, Models, Mothers and Babies: Bringing Birth Defects Research into Practice, Clearwater, FL, June
- Invited Speaker, 1st Alberta Neurodevelopmental Neuroscience Meeting, Edmonton Clinic Health Academy (ECHA), Edmonton, AB, Canada, July
- Invited Speaker, Allen Discovery Center at Boston Children's Hospital and Harvard Medical School, Boston, MA, September

- Invited Keynote, USERN 2018 Congress & ESGM Course on Primary Immunodeficiencies, Reggio Calabria, Italy, November
- Invited Keynote, Keystone Meeting: Leveraging Genomic Diversity to Promote Animal and Human Health, Uganda, Africa, November
- Invited Speaker, The 41st Annual Meeting of the Molecular Biology Society of Japan, Tokyo, Japan, November
- Invited Keynote, The 29th International Conference on Genome Informatics, Kunming, China, December
- Invited Speaker, Functional Genomics 2018: Big Data to Clinic, Doha, Qatar, December

2019

- Invited Faculty, McGill University's Bellairs Research Institute: Transposable Elements at the Crossroad of Health and Disease, Bellairs, Barbados, January
- Invited Speaker, CSHL: Double Helix Day: Insights into the Human Condition, Cold Spring Harbor, NY, February
- Invited Speaker, CSHL: Simons Center for Quantitative Biology Seminar, Cold Spring Harbor, NY, February
- Invited Speaker, CARTA Symposium 10th Anniversary: Revisiting the Agenda, La Jolla, CA, March
- Invited Session, Revolutionizing Next-Generation Sequencing (3rd Edition; RNGS3), Antwerp, Belgium, March
- Invited Plenary, ACMG Annual Clinical Genetics Meeting, Seattle, WA, April
- Invited Speaker, 16th Annual N. Ronald Morris Lecture, Rutgers University, Piscataway, NJ, April
- Invited Speaker, Clinical Genomics and NGS Course, Bertinoro, Italy, April
- Invited Speaker, Kjeldgaard Lecture, Aarhus University, Aarhus, Denmark, May
- Invited Keynote Speaker, SMRT Scientific Symposium, Leiden, The Netherlands, May
- Invited Seminar, Genetics, Bioinformatics & Systems Biology, UCSD, San Diego, CA, May
- Invited Speaker, New Insights Into Autism, NYGC, NYC, NY, October
- Invited Keynote Speaker, Australasian Genomic Technologies Association: AGTA19, Melbourne, Australia, October
- Invited Grand Rounds Speaker, UT Health San Antonio, 3rd Annual Patrick Holden Lectureship in Neurodevelopmental Disorders at Psychiatry Grand Rounds, San Antonio, TX, October
- Invited Keynote, 2019 Peking University Health Science Conferences on Autism Spectrum Disorders, Beijing, China, October
- Invited Speaker, Genome Informatics Meeting at CSHL, Cold Spring Harbor, NY, November
- Invited Speaker, 'Non-human primates - Novel insights into evolution and medicine' workshop, Center for Evolution & Medicine (CEM), Arizona State University, Phoenix, AZ, November
- Invited Speaker, Intellectual and Developmental Disabilities Research Center (IDDRC) Directors' Mtg, University of Washington, Seattle, WA November
- Invited Speaker, MASTERING GENOMES: It's a BLAST, A symposium on the occasion of Gene Myers' 65th birthday, Dresden, Germany, November

2020

- Invited Faculty, Workshop on Genomics Application, Český Krumlov, Czech Republic, January
- Participant, NHGRI Genomics2020 Strategic Planning Finale Meeting, Virtual, April
- Invited Speaker, Human and Mammalian Genetics and Genomics: The 61st McKusick Short Course, Virtual, July
- Invited Keynote, Cancer Genomics Consortium (CGC) 11th Annual Meeting, Virtual, August
- Speaker & Participant, Human Pangenome Reference Consortium (HPRC) & T2T Meetings, Virtual, September
- Invited Speaker, Simons Foundation Lecture, Virtual, October
- Invited Keynote, Pacific Biosciences of California, Inc., Virtual, November
- Invited Lecture, University of Tokyo, Virtual, November

2021

- Invited Speaker, NHGRI Bold Predictions Seminar, Virtual, February
- Invited Course Lecture, University of Southern California Introduction to Quantitative Biology Seminar, Virtual, February
- Invited Speaker, The Jackson Laboratory: Long-Read Sequencing Workshop, Virtual, April
- Invited Speaker, VII Brazilian Cytogenetics and Cytogenomics Meeting, Virtual, April
- Invited Speaker, 2021 Stanford Genetics Conference on Structural Variants and DNA Repeats, Virtual, April
- Invited Speaker, 15th Troina Meeting on Genetics of Neurodevelopmental Disorders (Italy), Virtual, April
- Invited Seminar, MIT Bioinformatics Seminar, Virtual, May
- Invited Speaker, Clinical Genomics & NGS - 33rd Course Jointly Organized by ESHG & CEUB, Bertinoro, Italy, Virtual, May
- Invited Keynote, 13th European Cytogenetics Conference (ECA), Virtual, July
- Invited Plenary Speaker, ESHG 2021 Virtual Conference, Virtual, August
- Invited Keynote, Precision Genomics Midwest 2021, Virtual, October

- Invited Seminar, Department of Microbiology, Biochemistry and Molecular Genetics and Public Health Research Institute Joint Seminar Series at Rutgers New Jersey Medical School, Virtual, November
- Invited Seminar, Department of Genetics, Genomics and Bioinformatics, University of Tennessee, Virtual, December
- Invited Plenary Speaker, 9th Pan Arab Human Genetics Conference (PAHGC), Virtual, December

2022

- Invited Speaker, 16th Troina Meeting on Genetics of Neurodevelopmental Disorders (Italy), Virtual, April
- Invited Seminar, Texas A&M Genetics and Genomics Seminar Series, College Station, TX, April
- Invited Speaker, Clinical Genomics & NGS - 34th Course Jointly Organized by ESHG & CEUB, Bertinoro, Italy, May
- Invited Speaker, The Jackson Laboratory: Long-Read Sequencing Workshop, Farmington, CT, May
- Invited Seminar, Stowers Institute, Kansas City, MO, May
- Invited Speaker, Genome Stability & Integrity Symposium at CSHL, Cold Spring Harbor, NY, June
- Invited Keynote, 4th International Conference of the Trisomy 21 (T21) Research Society, Long Beach, CA, June
- Invited Speaker, Telomere-to-Telomere Consortium, Santa Cruz, CA, August
- Invited Keynote, International Conference on Environmental Mutagens (ICEM), Ottawa, ON, Canada, August
- Invited Speaker, BMFZ Genomic Structural Variants Meeting, Düsseldorf, Germany, August
- Invited Speaker, Center for Autism and Neurodevelopment, Northwestern University Feinberg School of Medicine, Chicago, IL, November
- Invited Plenary Session, Association for Molecular Pathology's (AMP), Phoenix, AZ, November
- Invited Speaker, CARTA Fall 2022, Virtual, November
- Invited Speaker/Awardee, Falling Walls Symposium 2022, Berlin, Germany, November
- Invited Speaker, From Nucleotides to Neurons – A Symposium in Honor of the Centenary of Rosalind Franklin, New York, NY, December

2023

- Invited Speaker, MEDLAB Middle East 2023, Dubai, United Arab Emirates, February
- Invited Speaker, Pacific Northwest Research Institute (PNRI), Seattle, WA, February
- Invited Lecture, University of Utah, Salt Lake City, UT, March

RESEARCH GRANTS

a) Active

National Institutes of Health/ National Institutes of Mental Health (R01 MH101221-11)
Title: Rare Mutations and Autism Spectrum Disorders (previously: Sporadic Mutations and Autism Spectrum Disorders)
Goal: Use whole-genome sequence data to discover inherited and *de novo* mutations and genes responsible for autism and target 200 genes for deep resequencing in an additional set of 15,000 patients.
PI: Evan E. Eichler
2013–2025

National Institutes of Health (R01 HG002385-21)
Title: Sequence and Assembly of Segmental Duplications
Goal: Systematically sequence and assemble human and great ape segmental duplications, generate gene models from Iso-Seq data, and develop a genotyping platform for duplicated genes using molecular inversion probe assays.
PI: Evan E. Eichler
2007–2027

National Institutes of Health (R01 HG010169-05)
Title: Sequence-resolved structural variation of human genomes
Goal: Apply a multi-platform approach to sequence complex structural variation at the haplotype level and accurately genotype it from short-read sequence data generated from the 1000 Genomes Project.
PI: Evan E. Eichler
2018–2023

Simons Foundation (RFA 810018EE)
Title: Testing causal hypotheses for autism sex difference
Goal: Understand genetic basis for sex bias associated with autism.
Multi-PI: Aravinda Chakravarti; Evan E. Eichler
2021–2025

National Institutes of Health (1U01HG010971-04)
Title: Center for Human Reference Genome Diversity
Goal: Construction of a pangenome graph based on phased assemblies from diverse human genomes.
Multi-PI: David Haussler; Evan E. Eichler; Ira Hall; Erich Jarvis
2019–2024

National Institutes of Health (1U41HG007497-07)
Title: Identifying and Characterizing the Full Spectrum of Haplotype-resolved Structural Variation in Human Genomes
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; Evan E. Eichler; Jan Korbel
2013–2023

National Institutes of Health (3OT2OD002748-01S4)
Title: Northwest Genomics Center for *All of Us*
Goal: Establish a Genome Center for the *All of Us* Research Program. The NWGC for *All of Us* will provide whole-genome sequencing, genotyping and clinical validation of variants in the ACMG 59 genes.
Multi-PI: Gail Jarvik; Evan E. Eichler
2018–2023

National Institutes of Health (1U01HG011744-02)
Title: University of Washington Mendelian Genomics Research Center (UW-MGRC)
Goal: Establish the University of Washington Mendelian Genomics Research Center (UW-MGRC) with the overarching goal to maximize novel gene discovery for MCs, with an emphasis on canonical MCs that have gone unsolved using ES/WGS, and noncoding variants underlying MCs.
Multi-PI: Evan E. Eichler; Michael Bamshad
2021–2026

National Institutes of Health (1U01MH119705-04)
Title: Leveraging rare genetic etiologies to advance knowledge and treatment of neuropsychiatric disorders
Goal: Understand genetically defined subgroups with ASD through the support for a nationwide, clinical program collaborative as well as targeted comprehensive assessment of a large number of individuals with gene disrupting mutations.
Multi-PI: Christa Martin; David Ledbetter
2019–2024

National Institutes of Health (1U01HG10973-03)
Title: Representing structural haplotypes and complex genetic variation in pan-genome graphs
Goal: Evaluate and validate segmental duplication integration into the pan-genome repeat graphs and to benchmark genotyping methods of such regions.
PI: Mark Chaisson
2020–2024

National Heart, Lung, and Blood Institute (NHLBI) (HHSN268201600032I)
Title: UW Centralized Omics REsource (CORE) Task Order Nos. HHSN26800004, 75N92019F00074, and 75N92020F00001
Goal: Provide next-generation RNA sequencing (RNA-seq) and DNA methylation profiles for the Trans-Omics for Precision Medicine (TOPMed) Program.
PI: Evan E. Eichler
2016–2023

National Institutes of Health (NHLBI) (1R01NS122766-01A1)
Title: Novel approaches to identify tandem repeat expansions in neurodegenerative disease
Goal: Establish a novel paradigm to interrogate the mechanism of repeat expansion and reveal insight into novel genetic factors that cause or modulate risk for AD.
PI: Paul Valdmanis
2022–2027

b) Previous

Brotman Baty Institute (2021 Catalytic Collaborations)
Title: Targeted long-read sequencing to resolve complex structural variants and identify missing variants

Goal: Optimize methods for targeted long-read sequencing to identify disease-causing variation missed by existing sequencing and analysis methods.
PI: Evan E. Eichler
2021–2022

Sidra Medicine (NPRP10-1219-160035)
Title: Building haplotype-resolved de novo genome maps for Qatari family trios with severe developmental disorders as a premium resource to enhance diagnosis & uncover their etiology in the Qatari population
Goal: Generate high-resolution reference-unbiased genetic maps for families in Qatar with children suffering from severe or profound developmental disorders, together with underlying pathogenic variants (SNVs and SVs) and a comprehensive catalogue of Qatari-specific variation.
PI: Younes Mokrab
2018–2022

Simons Foundation (RFA 713892)
Title: Copy-number variation and pathogenic variant analyses of SPARK exomes
Goal: Discover pathogenic CNVs in SPARK family exomes.
PI: Evan E. Eichler
2020–2021

National Institutes of Health (5R01MH109912-04)
Title: 1/3 Building Integrative CNS Networks for Genomic Analysis of Autism
Goal: Understand the relative strengths of each network construction method for defining disease-related network relationships. *Eichler portion in Year 2.
PI: Dan Geschwind
2017–2021

Brotman Baty Institute (2019 Catalytic Collaborations)
Title: Long-read whole-genome sequencing of unsolved Mendelian cases of disease
Goal: Establish of a method for targeted long-read sequencing of native DNS from small regions of high clinical impact from the human genome.
PI: Evan E. Eichler
2020–2021

Simons Foundation (RFA 608045)
Title: Integrated CNV analysis of SPARK exomes
Goal: Discover pathogenic CNVs for ~15,000 exomes from 4,500 SPARK families.
PI: Evan E. Eichler
2018–2020

National Human Genome Research Institute (NHGRI) (5UM1HG008901-04)
Title: Robust STR calling from High-throughput Sequencing Technologies
Goal: Characterization of more complex structural variation not routinely detected by standard methods applied by the New York Genome Center (NYGC).
PI: Tom Maniatis
2016–2020

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 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: Ira Hall; Evan E. Eichler
2016–2019

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
2014–2018

National Institutes of Health (1U01NS077275)
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
2011–2018

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
2015–2018

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

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)
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 336475)
Title: Genome Pilot project of Simons Simplex Collection (SSC)
Goal: conduct whole-genome sequencing of 40 quads (parents + 1 affected proband + 1 unaffected sibling) from the SSC.
PI: Evan E. Eichler
2014–2015

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)
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 (5RO1NS069719)
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 (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)
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)
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)
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

(514 peer-reviewed publications: 223 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 interspersion 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, Neslon 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, McCready 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, Greally 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, Scwartz 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, Segraves 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, Archidiacano 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, Altshuler 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, Pazienza 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, Eyras 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. PMCID: N/A.

*Bailey JA, Kidd JM, **Eichler EE**. (2008). Human copy number polymorphic genes. *Cytogenet Genome Res* 123(1–4):234–243. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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-Ezquerra 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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, Gecz 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. PMCID: 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. PMCID: 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**, McDaneld TG, Keele JW. (2010). Analysis of copy number variations among diverse cattle breeds. *Genome Res* May;20(5):693–703. PMCID: 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, Fortea 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. PMCID: PMC5100745.

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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC2881400.

Teague B, Waterman MS, Goldstein S, Potamousis 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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, Thiese 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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, Wuu

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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC3508680.

Smith JJ, Baker C, **Eichler EE**, Amemiya CT. (2012). Genetic Consequences of Programmed Genome Rearrangement. *Curr Biol* Aug 21;22(16):1524–1529. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC3530683.

Mueller M, Barros P, Witherden AS, Roberts AL, Zhang Z, Schaschl H, Yu CY, Hurles 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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**, Breden 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. PMCID: PMC3617388.

Korvatska O, Strand NS, Berndt JD, Strovas 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC3690969.

McMichael G, Girirajan S, Moreno-De-Luca A, Gecz 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. PMCID: 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. PMCID: 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, Siegismund 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. PMCID: 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. PMCID: 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. PMCID: PMC3746892.

*Nuttle 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. PMCID: 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, Alldredge 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. PMCID: 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. PMCID: PMC3791263.

Giannuzzi G, Pazienza 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. PMCID: PMC3814877.

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

Vulto-van Silfhout AT, Rajamanickam S, Jensik PJ, Vergult S, de Rocker 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, Endele 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. PMCID: 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. PMCID: PMC4086126.

*Bernier R, Golzio C, Xiong B, Stessman HA, Coe BP, Penn O, Witherspoon K, Gerdts J, Baker C, Vulto-van Silfhout AT, Schuurs-Hoeijmakers JH, Fichera M, Bosco P, Buono S, Alberti A, Failla P, Peeters H, Steyaert J, Vissers LE, Francescatto L, Mefford HC, Rosenfeld JA, Bakken T, O'Roak BJ, Pawlus M, Moon R, Shendure J, Amaral DG, Lein E, Rankin J, Romano C, de Vries BB, Katsanis N, **Eichler EE**. (2014). Disruptive CHD8 mutations define a subtype of autism early in development. *Cell* Jul 17;158(2):263–276. PMCID: 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. PMCID: PMC4130548.

Vandeweyer G, Helsmoortel C, Van Dijck A, Vulto-van Silfhout AT, Coe BP, Bernier R, Gerdts 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. PMCID: 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. PMCID: 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, Gecz 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. PMCID: 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, Ku.inskas 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, Zemunik T, Cooper A, Capelli C, Thomas MG, Ruiz-Linares A, Tishkoff SA, Singh L, Thangaraj K, Villems 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. PMCID: PMC4170574.

Deriziotis P, O'Roak BJ, Graham SA, Estruch SB, Dimitropoulou D, Bernier RA, Gerdts J, Shendure J, **Eichler EE**, Fisher SE. (2014). De novo TBR1 mutations in sporadic autism disrupt protein functions. *Nat Commun* Sep 18;5:4954. PMCID: 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. PMCID: PMC4244265.

*Iossifov I, O'Roak BJ, Sanders SJ, Ronemus M, Krumm N, Levy D, Stessman HA, Witherspoon KT, Vives L, Pattterson KE, Smith JD, Paeper B, Nickerson DA, Dea J, Dong S, Gonzalez LE, Mandell JD, Mane SM, Murtha MT, Sullivan CA, Walker MF, Waqar Z, Wei L, Willsey AJ, Yamrom B, Lee YH, Grabowska E, Dalkic E, Wang Z, Marks S, Andrews P, Leotta A, Kendall J, Hakker I, Rosenbaum J, Ma B, Rodgers L, Troge J, Narzisi G, Yoon S, Schatz MC, Ye K, McCombie WR, Shendure J, **Eichler EE**,

State MW, Wigler M. (2014). The contribution of de novo coding mutations to autism spectrum disorder. *Nature* Nov 13;515(7526):216–221. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC4317170.

De Rocker 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. PMCID: 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. PMCID: PMC4458233.

Mazina V, Gerdts 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. PMCID: 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. PMCID: 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. PMCID: 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, Siegismund HR, Pagani L, Quail MA, Hvilsom 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. PMCID: 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. PMCID: 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. PMCID: 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, Hurles 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. PMCID: 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, Oberndorff K, van Bon BW, Shaw M, Gecz J, Haan E, Bienek M, Jensen C, Loeys BL, Van Dijck 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, Pediaditakis 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. PMCID: 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, Villems 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC4702975.

*van Bon BW, Coe BP, Bernier R, Green C, Gerdts 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC4970694.

*Stessman HA, Turner TN, **Eichler EE**. (2016). Molecular subtyping and improved treatment of neurodevelopmental disease. *Genome Med* Feb 25;8(1):22. PMCID: 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. PMCID: 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, Marcelis CM, van Bokhoven H, Cantagrel V, Colleaux L, Nicouleau M, Lyonnet S, Bernier RA, Gerdts J, Coe BP, Romano C, Alberti A, Grillo L, Scuderi C, Nordenskjöld M, Kvarnung 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, Jacquette 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. PMCID: 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. PMCID: 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, Hurles 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. PMCID: 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, Kukolich MK, McGaughran 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. PMCID: PMC4930086.

Rafati N, Andersson LS, Mikko S, Feng C, Raudsepp T, Pettersson J, Janecka J, Wattle O, Ameur A, Thyreen G, Eberth J, Huddleston J, Malig M, Bailey E, **Eichler EE**, Dalin G, Chowdary B, Anderssson 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. PMCID: 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. PMCID: 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. PMCID: PMC4931320.

*Nuttle X, Giannuzzi G, Duyzend MH, 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. PMCID: 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. doi: 10.1111/trf.13797. PMID: 27580710. PMCID: N/A.

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. PMCID: PMC5059695.

*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. PMCID: 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. PMCID: PMC5105161.

*Turner TN, Yi Q, Krumm N, Huddleston J, Hoekzema K, Stessman HAF, Doebley A, Bernier RA, Nickerson DA, **Eichler EE**. (2017). denovo-db: a compendium of human de novo variants. *Nucl Acids Res* Jan 4;45(D1):D804–D811. doi: 10.1093/nar/gkw865. Epub 2016 Oct 5. PMCID: PMC5210614.

Bramswig NC, Lüdecke HJ, Pettersson M, Albrecht B, Bernier RA, Cremer K, **Eichler EE**, Falkenstein D, Gerdts 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, Wieczorek 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. doi: 10.1007/s00439-016-1743-x. PMCID: PMC5821420.

Tolomeo D, Capozzi O, Stanyon RR, Archidiacono N, D'Addabbo P, Catacchio CR, Purgato S, Perini G, Schempp W, Huddleston J, Malig M, **Eichler EE**, Rocchi M. (2017). Epigenetic origin of evolutionary novel centromeres. *Sci Rep* Feb 3;7:41980. doi: 10.1038/srep41980. PMCID: PMC5290474.

*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, Nuttle X, Hoekzema K, Lindsay-Graves TA, Wilson RK, **Eichler EE**. (2017). The evolution and population diversity of human-specific segmental duplications. *Nat Ecol Evol* Feb 17;1:69. doi:10.1038/s41559-016-0069. PMCID: PMC5450946. http://hdl.handle.net/1773/38703

*Dougherty ML, Nuttle 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. PMCID: 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* Jun;174(4):381–389. PMCID: PMC5467442.

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* Jul 1;34(7):1669–1681. PMCID: PMC5722054.

*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 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nat Genet* Apr;49(4):515–526. PMCID: PMC5374041.

*Huddleston J, Chaisson MJP, 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. PMCID: PMC5411763.

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. PMCID: PMC5411779.

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* Jun 5;10(6):866–877.

*Chaisson MJ, Mukherjee S, Kannan S, **Eichler EE**. (2017). Resolving multicopy duplications de novo using polyploid phasing. In: Sahinalp S. (eds) *Res Comput Mol Biol* RECOMB 2017. Lecture Notes in Computer Science, vol 10229. Springer, Cham, May;10229: 117–133. PMCID: PMC5553120.

Xia EH, Yang DR, Jiang JJ, Zhang QJ, Liu Y, Liu YL, Zhang Y, Zhang HB, Shi C, Tong Y, Kim C, Chen H, Peng YQ, Yu Y, Zhang W, **Eichler EE**, Gao LZ. (2017). The caterpillar fungus, Ophiocordyceps sinensis, genome provides insights into highland adaptation of fungal pathogenicity. *Sci Rep* May 11;7(1):1806. PMCID: PMC5432009.

Hudac CM, Stessman HAF, DesChamps TD, Kresse A, Faja S, Neuhaus E, Webb SJ, **Eichler EE**, Bernier RA. (2017). Exploring the heterogeneity of neural social indices for genetically distinct etiologies of autism. *J Neurodev Disord* May 26;9:24. PMCID: PMC5446693.

*Geisheker MR, Heymann G, Wang T, Coe BP, Turner TN, Stessman HAF, Hoekzema K, Kvarnung M, Shaw M, Friend K, Liebelt J, Barnett C, Thompson EM, Haan E, Guo H, Anderlid BM, Nordgren A, Lindstrand A, Vandeweyer G, Alberti A, Avola E, Vinci M, Giusto S, Pramparo T, Pierce K, Nalabolu S, Michaelson JJ, Sedlacek Z, Santen GWE, Peeters H, Hakonarson H, Courchesne E,

Romano C, Kooy RF, Bernier RA, Nordenskjöld M, Gecz J, Xia K, Zweifel LS, **Eichler EE**. (2017). Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nat Neurosci* Aug;20(8):1043–1051. PMCID: PMC5539915.

*Turner TN, Coe BP, Dickel DE, Hoekzema K, Nelson BJ, Zody MC, Kronenberg ZN, Hormozdiari F, Raja A, Pennacchio LA, Darnell RB, **Eichler EE**. (2017). Genomic patterns of de novo mutation in simplex autism. *Cell* Oct 19;171(3):710–722.e12. PMCID: PMC5679715.

Prufer K, de Filippo C, Grote S, Mafessoni F, Korlevic P, Hajdinjak M, Vernot B, Skov L, Hsieh P, Peyregne S, Reher D, Hopfe C, Nagel S, Maricic T, Fu Q, Theunert C, Rogers R, Skoglund P, Chintalapati M, Dannemann M, Nelson BJ, Key FM, Rudan P, Kucan Z, Gusic I, Golovanova LV, Doronichev VB, Patterson N, Reich D, **Eichler EE**, Slatkin M, Schierup MH, Andres A, Kelso J, Meyer M, Paabo S. (2017). A high-coverage Neandertal genome from Vindija Cave in Croatia. *Science* Nov 3;358(6363):655–658. PMCID: PMC6185897.

Earl RK, Turner TN, Mefford HC, Hudac CM, Gerdts J, **Eichler EE**, Bernier RA. (2017). Clinical phenotype of ASD-associated *DYRK1A* haploinsufficiency. *Mol Autism* Oct 5;8:54. PMCID: PMC5629761.

Siper PM, De Rubeis S, Trelles MDP, Durkin A, Di Marino D, Muratet F, Frank Y, Lozano R, **Eichler EE**, Kelly M, Beighley J, Gerdts J, Wallace AS, Mefford HC, Bernier RA, Kolevzon A, Buxbaum JD. (2017). Prospective investigation of FOXP1 syndrome. *Mol Autism* Oct 24;8:57. PMCID: PMC5655854.

Kuderna LFK, Tomlinson C, Hillier LW, Tran A, Fiddes I, Armstrong J, Laayouni H, Gordon D, Huddleston J, Perez RG, Povolotskaya I, Armero AS, Garrido JG, Ho D, Ribeca P, Alioto T, Green RE, Paten B, Navarro A, Betranpetit J, Herrero J, **Eichler EE**, Sharp AJ, Feuk L, Warren WC, Marques-Bonet T. (2017). A 3-way hybrid approach to generate a new high quality chimpanzee reference genome (Pan_tro_3.0). *Gigascience* Nov 1;6(11):1–6. PMCID: PMC5714192.

Küry S, van Woerden GM, Besnard T, Proietti Onori M, Latypova X, Towne MC, Cho MT, Prescott TE, Ploeg MA, Sanders S, Stessman HAF, Pujol A, Distel B, Robak LA, Bernstein JA, Denommé-Pichon AS, Lesca G, Sellars EA, Berg J, Carré W, Busk ØL, van Bon BWM, Waugh JL, Deardorff M, Hoganson GE, Bosanko KB, Johnson DS, Dabir T, Holla ØL, Sarkar A, Tveten K, de Bellescize J, Braathen GJ, Terhal PA, Grange DK, van Haeringen A, Lam C, Mirzaa G, Burton J, Bhoj EJ, Douglas J, Santani AB, Nesbitt AI, Helbig KL, Andrews MV, Begtrup A, Tang S, van Gassen KLI, Juusola J, Foss K, Enns GM, Moog U, Hinderhofer K, Paramasivam N, Lincoln S, Kusako BH, Lindenbaum P, Charpentier E, Nowak CB, Cherot E, Simonet T, Ruivenkamp CAL, Hahn S, Brownstein CA, Xia F, Schmitt S, Deb W, Bonneau D, Nizon M, Quinquis D, Chelly J, Rudolf G, Sanlaville D, Parent P, Gilbert-Dussardier B, Toutain A, Sutton VR, Thies J, Peart-Vissers LELM, Boisseau P, Vincent M, Grabrucker AM, Dubourg C; Undiagnosed Diseases Network, Tan WH, Verbeek NE, Granzow M, Santen GWE, Shendure J, Isidor B, Pasquier L, Redon R, Yang Y, State MW, Kleefstra T, Cogné B; GEM HUGO; Deciphering Developmental Disorders Study, Petrovski S, Retterer K, **Eichler EE**, Rosenfeld JA, Agrawal PB, Bézieau S, Odent S, Elgersma Y, Mercier S. (2017). De novo mutations in protein kinase genes CAMK2A and CAMK2B cause intellectual disability. *Am J Hum Genet* Nov 2;101(5):768–788. PMCID: PMC5673671.

Luhrs K, Ward T, Hudac CM, Gerdts J, Stessman HAF, **Eichler EE**, Bernier RA. (2017). Associations between familial rates of psychiatric disorders and de novo genetic mutations in autism. *Autism Res Treat* 2017:9371964. doi: 10.1155/2017/9371964. Epub 2017 Nov 8. PMCID: PMC5698792.

Wallace AS, Hudac CM, Steinman KJ, Peterson JL, DesChamps TD, Duyzend MH, Nuttle X, **Eichler EE**, Bernier RA. (2017). Longitudinal report of child with de novo 16p11.2 triplication. *Clin Case Rep* Dec 6;6(1):147–154. doi: 10.1002/ccr3.1236. eCollection 2018 Jan. PMCID: PMC5771938.

Jansen S, Hoischen A, Coe BP, Carvill GL, Van Esch H, Bosch DGM, Andersen UA, Baker C, Bauters M, Bernier RA, van Bon BW, Claahsen-van der Grinten HL, Gecz J, Gilissen C, Grillo L, Hackett A, Kleefstra T, Koolen D, Kvarnung M, Larsen MJ, Marcelis C, McKenzie F, Monin ML, Nava C, Schuurs-Hoeijmakers JH, Pfundt R, Steehouwer M, Stevens SJC, Stumpel CT, Vansenne F, Vinci M, van de Vorst M, Vries P, Witherspoon K, Veltman JA, Brunner HG, Mefford HC, Romano C, Vissers LELM, **Eichler EE**, de Vries BBA. (2018). A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. *Eur J Hum Genet* Jan;26(1):54–63. PMCID: PMC5839042.

Smith JJ, Timoshevskaya N, Ye C, Holt C, Keinath MC, Parker HJ, Cook ME, Hess JE, Narum SR, Lamanna F, Kaessmann H, Timoshevskiy VA, Waterbury CKM, Saraceno C, Wiedemann LM, Robb SMC, Baker C, **Eichler EE**, Hockman D, Sauka-Spengler T, Yandell M, Krumlauf R, Elgar G, Amemiya CT. (2018). The sea lamprey germline genome provides insights into programmed genome rearrangement and vertebrate evolution. *Nat Genet* Feb;50(2):270–277. PMCID: PMC5805609.

Aneichyk T, Hendriks WT, Yadav R, Shin D, Gao D, Vaine CA, Collins RL, Domingo A, Currall B, Stortchevoi A, Multhaupt-Buell T, Penney EB, Cruz L, Dhakal J, Brand H, Hanscom C, Antolik C, Dy M, Ragavendran A, Underwood J, Cantsilieris S, Munson KM, **Eichler EE**, Acuna P, Go C, Jamora RDG, Rosales RL, Church DM, Williams SR, Garcia S, Klein C, Muller U, Wilhelmsen KC, Timmers HTM, Sapir Y, Wainger BJ, Henderson D, Ito N, Weisenfeld N, Jaffe D, Sharma N, Breakefield XO, Ozelius LJ, Bragg DC, Talkowski ME. (2018). Dissecting the causal mechanism of X-linked Dystonia-Parkinsonism by integrating genome and transcriptome assembly. *Cell* Feb 22;172(5):897–909.e21. PMCID: PMC5831509.

Arnett AB, Cairney BE, Wallace AS, Gerdts J, Turner TN, **Eichler EE**, Bernier RA. (2018). Comorbid symptoms of inattention, autism, and executive cognition in youth with putative genetic risk. *J Child Psychol Psychiatry* Mar;59(3):268-276. doi: 10.1111/jcpp.12815. Epub 2017 Sep 18. PMCID: PMC5812799.

Cheng H, Dharmadhikari AV, Varland S, Ma N, Domingo D, Kleyner R, Rope AF, Yoon M, Stray-Pedersen A, Posey JE, Crews SR, Eldomery MK, Akdemir ZC, Lewis AM, Sutton VR, Rosenfeld JA, Conboy E, Agre K, Xia F, Walkiewicz M, Longoni M, High FA, van Slegtenhorst MA, Mancini GMS, Finnila CR, van Haeringen A, den Hollander N, Ruivenkamp C, Naidu S, Mahida S, Palmer EE, Murray L, Lim D, Jayakar P, Parker MJ, Giusto S, Stracuzzi E, Romano C, Beighley JS, Bernier RA, Küry S, Nizon M, Corbett MA, Shaw M, Gardner A, Barnett C, Armstrong R, Kassahn KS, Van Dijck A, Vandeweyer G, Kleefstra T, Schieving J, Jongmans MJ, de Vries BBA, Pfundt R, Kerr B, Rojas SK, Boycott KM, Person R, Willaert R, **Eichler EE**, Kooy RF, Yang Y, Wu JC, Lupski JR, Arnesen T, Cooper GM, Chung WK, Gecz J, Stessman HAF, Meng L, Lyon GJ. (2018). Truncating variants in NAA15 are associated with variable levels of intellectual disability, autism spectrum disorder, and congenital anomalies. *Am J Hum Genet* May 3;102(5):985–994. PMCID: PMC5986698.

*Cantsilieris S, Nelson BJ, Huddleston J, Baker C, Harshman L, Penewit K, Munson KM, Sorensen M, Welch AE, Dang V, Grassmann F, Richardson AJ, Guymer RH, Graves-Lindsay TA, Wilson RK, Weber BHF, Baird PN, Allikmets R, **Eichler EE**. (2018). Recurrent structural variation, clustered sites of selection, and disease risk for the complement factor H (CFH) gene family. *Proc Natl Acad Sci U S A* May 8;115(19):E4433–E4442. PMCID: PMC5948961.

Fiddes IT, Lodewijk GA, Mooring M, Bosworth CM, Ewing AD, Mantalas GL, Novak AM, van den Bout A, Bishara A, Rosenkrantz JL, Lorig-Roach R, Field AR, Haeussler M, Russo L, Bhaduri A, Nowakowski TJ, Pollen AA, Dougherty ML, Nuttle X, Addor MC, Zwolinski S, Katzman S, Kriegstein A, **Eichler EE**, Salama SR, Jacobs FMJ, Haussler D. (2018). Human-specific NOTCH2NL genes affect notch signaling and cortical neurogenesis. *Cell* May 31;173(6):1356–1369.e22. PMCID: PMC5986104.

Catacchio CR, Maggiolini FAM, D'Addabbo P, Bitonto M, Capozzi O, Signorile ML, Miroballo M, Archidiacono N, **Eichler EE**, Ventura M, Antonacci F. (2018). Inversion variants in human and primate genomes. *Genome Res* Jun;28(6):910–920. PMCID: PMC5991517.

*Kronenberg ZN, Fiddes IT, Gordon D, Murali S, Cantsilieris S, Meyerson OS, Underwood JG, Nelson BJ, Chaisson MJP, Dougherty ML, Munson KM, Hastie AR, Diekhans M, Hormozdiari F, Lorusso N, Hoekzema K, Qiu R, Clark K, Raja A, Welch AE, Sorensen M, Baker C, Fulton RS, Armstrong J, Graves-Lindsay TA, Denli AM, Hoppe ER, Hsieh P, Hill CM, Pang AWC, Lee J, Lam ET, Dutcher SK, Gage FH, Warren WC, Shendure J, Haussler D, Schneider VA, Cao H, Ventura M, Wilson RK, Paten B, Pollen A, **Eichler EE**. (2018). High-resolution comparative analysis of great ape genomes. *Science* Jun 8;360(6393). pii: eaar6343. doi: 10.1126/science.aar6343. PMCID: PMC6178954.

Fiddes IT, Armstrong J, Diekhans M, Nachtweide S, Kronenberg ZN, Underwood JG, Gordon D, Earl D, Keane T, **Eichler EE**, Haussler D, Stanke M, Paten B. (2018). Comparative Annotation Toolkit (CAT)-simultaneous clade and personal genome annotation. *Genome Res* Jul;28(7):1029–1038. PMCID: PMC6028123.

Ghareghani M, Porubsk D, Sanders AD, Meiers S, **Eichler EE**, Korbel JO, Marschall T. (2018). Strand-seq enables reliable separation of long reads by chromosome via expectation maximization. *Bioinformatics* Jul 1;34(13):i115–i123. PMCID: PMC6022540.

Tucci S, Vohr SH, McCoy RC, Vernot B, Robinson MR, Barbieri C, Nelson BJ, Fu W, Purnomo GA, Sudoyo H, **Eichler EE**, Barbujani G, Visscher PM, Akey JM, Green RE. (2018). Evolutionary history and adaptation of a human pygmy population of Flores Island, Indonesia. *Science* Aug 3;361(6401):511–516. PMCID: PMC6709593.

Arnett AB, Rhoads CL, Hoekzema K, Turner TN, Gerdts J, Wallace AS, Bedrosian-Sermone S, **Eichler EE**, Bernier RA. (2018). The autism spectrum phenotype in ADNP syndrome. *Autism Res* Sep;11(9):1300–1310. PMCID: PMC6203613.

*Dougherty ML, Underwood JG, Nelson BJ, Tseng E, Munson KM, Penn O, Nowakowski TJ, Pollen AA, **Eichler EE**. (2018). Transcriptional fates of human-specific segmental duplications in brain. *Genome Res* Oct;28(10):1566–1576. PMCID: PMC6169893.

*Vollger MR, Dishuck PC, Sorensen M, Welch AE, Dang V, Dougherty ML, Graves-Lindsay TA, Wilson RK, Chaisson MJP, **Eichler EE**. (2019). Long-read sequence and assembly of segmental duplications. *Nat Methods* Jan;16(1):88–94. Epub 2018 Dec 17. PMCID: PMC6382464.

*Coe BP, Stessman HAF, Sulovari A, Geisheker MR, Bakken TE, Lake AM, Dougherty JD, Lein ES, Hormozdiari F, Bernier RA, **Eichler EE**. (2019). Neurodevelopmental disease genes implicated by de novo mutation and copy number variation morbidity. *Nat Genet* Jan;51(1):106-116. doi: 10.1038/s41588-018-0288-4. Epub 2018 Dec 17. PMCID: PMC6309590.

*Guo H, Wang T, Wu H, Long M, Coe BP, Li H, Xun G, Ou J, Chen B, Duan G, Bai T, Zhao N, Shen Y, Li Y, Wang Y, Zhang Y, Baker C, Liu Y, Pang N, Huang L, Han L, Jia X, Liu C, Ni H, Yang X, Xia L, Chen J, Shen L, Li Y, Zhao R, Zhao W, Peng J, Pan Q, Long Z, Su W, Tan J, Du X, Ke X, Yao M, Hu Z, Zou X, Zhao J, Bernier RA, **Eichler EE**, Xia K. (2018). Inherited and multiple de novo mutations in autism/developmental delay risk genes suggest a multifactorial model. *Mol Autism* Dec 13;9:64. doi: 10.1186/s13229-018-0247-z. eCollection 2018. PMCID: PMC6293633.

Ang CE, Ma Q, Wapinski OL, Fan S, Flynn RA, Lee QY, Coe B, Onoguchi M, Olmos VH, Do BT, Dukes-Rimsky L, Xu J, Tanabe K, Wang L, Elling U, Penninger JM, Zhao Y, Qu K, **Eichler EE**, Srivastava A, Wernig M, Chang HY. (2019). The novel lncRNA lnc-NR2F1 is pro-neurogenic and mutated in human neurodevelopmental disorders. *Elife* Jan 10;8. pii: e41770. PMCID: PMC6380841.

*Audano PA, Sulovari A, Graves-Lindsay TA, Cantsilieris S, Sorensen M, Welch AE, Dougherty ML, Nelson BJ, Shah A, Dutcher SK, Warren WC, Magrini V, McGrath SD, Li YI, Wilson RK, **Eichler EE**. (2019). Characterizing the major structural variant alleles of the human genome. *Cell* Jan 24;176(3):663–675. PMCID: PMC6438697.

Van Dijck A, Vulto-van Silfhout AT, Cappuyns E, van der Werf IM, Mancini GM, Tzschach A, Bernier R, Gozes I, **Eichler EE**, Romano C, Lindstrand A, Nordgren A; ADNP Consortium, Kvarnung M, Kleefstra T, de Vries BBA, Küry S, Rosenfeld JA, Meuwissen ME, Vandeweyer G, Kooy RF. (2019). Clinical presentation of a complex neurodevelopmental disorder caused by mutations in ADNP. *Biol Psychiatry* Feb 15;85(4):287–297. Epub 2018 Mar 15. PMCID: PMC6139063.

Pollen AA, Bhaduri A, Andrews MG, Nowakowski TJ, Meyerson OS, Mostajo-Radji MA, Di Lullo E, Alvarado B, Bedolli M, Dougherty ML, Fiddes IT, Kronenberg ZN, Shuga J, Leyrat AA, West JA, Bershteyn M, Lowe CB, Pavlovic BJ, Salama SR, Haussler D, **Eichler EE**, Kriegstein AR. (2019). Establishing cerebral organoids as models of human-specific brain evolution. *Cell* Feb 7;176(4):743–756.e17. PMCID: PMC6544371.

Cogne B, Ehresmann S, Beauregard-Lacroix E, Rousseau J, Besnard T, Garcia T, Petrovski S, Avni S, McWalter K, Blackburn PR, Sanders SJ, Uguen K, Harris J, Cohen JS, Blyth M, Lehman A, Berg J, Li MH, Kini U, Joss S, von der Lippe C, Gordon CT, Humberson JB, Robak L, Scott DA, Sutton VR, Skraban CM, Johnston JJ, Poduri A, Nordenskjöld M, Shashi V, Gerkes EH, Bongers EMHF, Gilissen C, Zarate YA, Kvarnung M, Lally KP, Kulch PA, Daniels B, Hernandez-Garcia A, Stong N, McGaughran J, Retterer K, Tveten K, Sullivan J, Geisheker MR, Stray-Pedersen A, Tarpinian JM, Klee EW, Sapp JC, Zyskind J, Holla OL, Bedoukian E, Filippini F, Guimier A, Picard A, Busk OL, Punetha J, Pfundt R, Lindstrand A, Nordgren A, Kalb F, Desai M, Ebanks AH, Jhangiani SN, Dewan T, Coban Akdemir ZH, Telegrafi A, Zackai EH, Begtrup A, Song X, Toutain A, Wentzensen IM, Odent S, Bonneau D, Latypova X, Deb W; CAUSES Study, Redon S, Bilan F, Legendre M, Troyer C, Whitlock K, Caluseriu O, Murphree MI, Pichurin PN, Agre K, Gavrilova R, Rinne T, Park M, Shain C, Heinzen EL, Xiao R, Amiel J, Lyonnet S, Isidor B, Biesecker LG, Lowenstein D, Posey JE, Denomme-Pichon AS; Deciphering Developmental Disorders study, Ferec C, Yang XJ, Rosenfeld JA, Gilbert-Dussardier B, Audebert-Bellanger S, Redon R, Stessman HAF, Nellaker C, Yang Y, Lupski JR, Goldstein DB, **Eichler EE**, Bolduc F, Bezieau S, Kury S, Campeau PM. (2019). Missense variants in the histone acetyltransferase complex component gene TRRAP cause autism and syndromic intellectual disability. *Am J Hum Genet* Mar 7;104(3):530–541. PMCID: PMC6407527.

Maggiolini FAM, Cantsilieris S, D'Addabbo P, Manganelli M, Coe BP, Dumont BL, Sanders AD, Pang AWC, Vollger MR, Palumbo O, Palumbo P, Accadia M, Carella M, **Eichler EE**, Antonacci F. (2019). Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. *PLoS Genet* Mar 27;15(3):e1008075. PMCID: PMC6436712.

Tilghman JM, Ling AY, Turner TN, Sosa MX, Krumm N, Chatterjee S, Kapoor A, Coe BP, Nguyen KH, Gupta N, Gabriel S, **Eichler EE**, Berrios C, Chakravarti A. (2019). Molecular genetic anatomy and risk profile of Hirschsprung's disease. *N Engl J Med* Apr 11;380(15):1421–1432. PMCID: PMC6596298.

*Chaisson MJP, Sanders AD, Zhao X, Malhotra A, Porubsky D, Rausch T, Gardner EJ, Rodriguez OL, Guo L, Collins RL, Fan X, Wen J, Handsaker RE, Fairley S, Kronenberg ZN, Kong X, Hormozdiari F, Lee D, Wenger AM, Hastie AR, Antaki D, Anantharaman T, Audano PA, Brand H, Cantsilieris S, Cao H, Cerveira E, Chen C, Chen X, Chin CS, Chong Z, Chuang NT, Lambert CC, Church DM, Clarke L, Farrell A, Flores J, Galeev T, Gorkin DU, Gujral M, Guryev V, Heaton WH, Korlach J, Kumar S, Kwon JY, Lam ET, Lee JE, Lee J, Lee WP, Lee SP, Li S, Marks P, Viaud-Martinez K, Meiers S, Munson KM, Navarro FCP, Nelson BJ, Nodzak C, Noor A, Kyriazopoulou-Panagiotopoulou S, Pang AWC, Qiu Y, Rosanio G, Ryan M, Stütz A, Spierings DCJ, Ward A, Welch AE, Xiao M, Xu W, Zhang C, Zhu Q, Zheng-Bradley X, Lowy E, Yakneen S, McCarroll S, Jun G, Ding L, Koh CL, Ren B, Flicek P, Chen K, Gerstein MB, Kwok PY, Lansdorp PM, Marth GT, Sebat J, Shi X, Bashir A, Ye K, Devine SE, Talkowski ME, Mills RE, Marschall T, Korbel JO, **Eichler EE**, Lee C. (2019). Multi-platform discovery of haplotype-resolved structural variation in human genomes. *Nat Commun* Apr 16;10(1):1784. PMCID: PMC6467913.

*Guo H, Duyzend MH, Coe BP, Baker C, Hoekzema K, Gerdts J, Turner TN, Zody MC, Beighley JS, Murali SC, Nelson BJ; University of Washington Center for Mendelian Genomics, Bamshad MJ, Nickerson DA, Bernier RA, **Eichler EE**. (2019). Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. *Genet Med* Jul;21(7):1611–1620. Epub 2018 Dec 3. PMCID: PMC6546556.

Fenckova M, Blok LER, Asztalos L, Goodman DP, Cizek P, Singgih EL, Glennon JC, IntHout J, Zweier C, **Eichler EE**, von Reyn CR, Bernier RA, Asztalos Z, Schenck A. (2019). Habituation learning is a widely affected mechanism in drosophila models of intellectual disability and autism spectrum disorders. *Biol Psychiatry* Aug 15;86(4):294–305. PMCID: PMC7053436.

Salpietro V, Dixon CL, Guo H, Bello OD, Vandrovcova J, Efthymiou S, Maroofian R, Heimer G, Burglen L, Valence S, Torti E, Hacke M, Rankin J, Tariq H, Colin E, Procaccio V, Striano P, Mankad K, Lieb A, Chen S, Pisani L, Bettencourt C, Männikkö R, Manole A, Brusco A, Grosso E, Ferrero GB, Armstrong-Moron J, Gueden S, Bar-Yosef O, Tzadok M, Monaghan KG, Santiago-Sim T, Person RE, Cho MT, Willaert R, Yoo Y, Chae JH, Quan Y, Wu H, Wang T, Bernier RA, Xia K, Blesson A, Jain M, Motazacker MM, Jaeger B, Schneider AL, Boysen K, Muir AM, Myers CT, Gavrilova RH, Gunderson L, Schultz-Rogers L, Klee EW, Dyment D, Osmond M, Parellada M, Llorente C, Gonzalez-Peñas J, Carracedo A, Van Haeringen A, Ruivenkamp C, Nava C, Heron D, Nardello R, Iacomino M, Minetti C, Skabar A, Fabretto A; SYNAPS Study Group, Raspall-Chaure M, Chez M, Tsai A, Fassi E, Shinawi M, Constantino JN, De Zorzi R, Fortuna S, Kok F, Keren B, Bonneau D, Choi M, Benzeev B, Zara F, Mefford HC, Scheffer IE, Clayton-Smith J, Macaya A, Rothman JE, **Eichler EE**, Kullmann DM, Houlden H. (2019). AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. *Nat Commun* Jul 12;10(1):3094. PMCID: PMC6626132.

*Gao LZ, Liu YL, Zhang D, Li W, Gao J, Liu Y, Li K, Shi C, Zhao Y, Zhao YJ, Jiao JY, Mao SY, Gao CW, **Eichler EE**. (2019). Evolution of Oryza chloroplast genomes promoted adaptation to diverse ecological habitats. *Commun Biol* Jul 26;2:278. PMCID: PMC6659635.

Nellåker C, Alkuraya FS, Baynam G, Bernier RA, Bernier FPJ, Boulanger V, Brudno M, Brunner HG, Clayton-Smith J, Cogné B, Dawkins HJS, deVries BBA, Douzgou S, Dudding-Byth T, **Eichler EE**, Ferlaino M, Fieggen K, Firth HV, FitzPatrick DR, Gration D, Groza T, Haendel M, Hallowell N, Hamosh A, Hehir-Kwa J, Hitz MP, Hughes M, Kini U, Kleefstra T, Kooy RF, Krawitz P, Küry S, Lees M, Lyon GJ, Lyonnet S, Marcadier JL, Meyn S, Moslerová V, Politei JM, Poulton CC, Raymond FL, Reijnders MRF, Robinson PN, Romano C, Rose CM, Sainsbury DCG, Schofield L, Sutton VR, Turnovec M, Van Dijck A, Van Esch H, Wilkie AOM; Minerva Consortium. (2019). Enabling global clinical collaborations on identifiable patient data: The Minerva Initiative. *Front Genet* Jul 29;10:611. PMCID: PMC6681681.

Feliciano P, Zhou X, Astrovskaya I, Turner TN, Wang T, Brueggeman L, Barnard R, Hsieh A, Snyder LG, Muzny DM, Sabo A; SPARK Consortium, Gibbs RA, **Eichler EE**, O'Roak BJ, Michaelson JJ, Volfovsky N, Shen Y, Chung WK. (2019). Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. *NPJ Genom Med* Aug 23;4:19. PMCID: PMC6707204.

He Y, Luo X, Zhou B, Hu T, Meng X, Audano PA, Kronenberg ZN, **Eichler EE**, Jin J, Guo Y, Yang Y, Qi X, Su B. (2019). Long-read assembly of the Chinese rhesus macaque genome and identification of ape-specific structural variants. *Nat Commun* Sep 17;10(1):4233. PMCID: PMC6749001.

*Guo H, Li Y, Shen L, Wang T, Jia X, Liu L, Xu T, Ou M, Hoekzema K, Wu H, Gillentine MA, Liu C, Ni H, Peng P, Zhao R, Zhang Y, Phornphutkul C, Stegmann APA, Prada CE, Hopkin RJ, Shieh JT, McWalter K, Monaghan KG, van Hasselt PM, van Gassen K, Bai T, Long M, Han L, Quan Y, Chen M, Zhang Y, Li K, Zhang Q, Tan J, Zhu T, Liu Y, Pang N, Peng J, Scott DA, Lalani SR, Azamian M, Mancini GMS, Adams DJ, Kvarnung M, Lindstrand A, Nordgren A, Pevsner J, Osei-Owusu IA, Romano C, Calabrese G, Galesi O, Gecz J, Haan E, Ranells J, Racobaldo M, Nordenskjold M, Madan-Khetarpal S, Sebastian J, Ball S, Zou X, Zhao J, Hu Z, Xia F, Liu P, Rosenfeld JA, de Vries BBA, Bernier RA, Xu ZD, Li H, Xie W, Hufnagel RB, **Eichler EE**, Xia K. (2019). Disruptive variants of *CSDE1* associate with autism and interfere with neuronal development and synaptic transmission. *Sci Adv* Sep 25;5(9):eaax2166. doi: 10.1126/sciadv.aax2166. eCollection 2019 Sep. PMCID: PMC6760934.

*Guo H, Bettella E, Marcogliese PC, Zhao R, Andrews JC, Nowakowski TJ, Gillentine MA, Hoekzema K, Wang T, Wu H, Jangam S, Liu C, Ni H, Willemsen MH, van Bon BW, Rinne T, Stevens SJC, Kleefstra T, Brunner HG, Yntema HG, Long M, Zhao W, Hu Z, Colson C, Richard N, Schwartz CE, Romano C, Castiglia L, Bottitta M, Dhar SU, Erwin DJ, Emrick L, Keren B, Afenjar A, Zhu B, Bai B, Stankiewicz P, Herman K; University of Washington Center for Mendelian Genomics, Mercimek-Andrews S, Juusola J, Wilfert AB, Abou Jamra R, Büttner B, Mefford HC, Muir AM, Scheffer IE, Regan BM, Malone S, Gecz J, Cobben J, Weiss MM, Waisfisz Q, Bijlsma EK, Hoffer MJV, Ruivenkamp CAL, Sartori S, Xia F, Rosenfeld JA, Bernier RA, Wangler MF, Yamamoto S, Xia K, Stegmann APA, Bellen HJ, Murgia A, **Eichler EE**. (2019). Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nat Commun* Oct 15;10(1):4679. PMCID: PMC6794285.

*Hsieh P, Vollger MR, Dang V, Porubsky D, Baker C, Cantsilieris S, Hoekzema K, Lewis AP, Munson KM, Sorensen M, Kronenberg ZN, Murali S, Nelson BJ, Chiatante G, Maggiolini FAM, Blanché H, Underwood JG, Antonacci F, Deleuze JF, **Eichler EE**. (2019). Adaptive archaic introgression of copy number variants and the discovery of previously unknown human genes. *Science* Oct 18;366(6463). PMCID: PMC6860971.

Giannuzzi G, Schmidt PJ, Porcu E, Willemin G, Munson KM, Nuttle X, Earl R, Chrast J, Hoekzema K, Risso D, Männik K, De Nittis P, Baratz ED; 16p11.2 Consortium, Herault Y, Gao X, Philpott CC, Bernier RA, Kutalik Z, Fleming MD, **Eichler EE**, Reymond A. (2019). The human-specific BOLA2 duplication modifies iron homeostasis and anemia predisposition in chromosome 16p11.2 autism individuals. *Am J Hum Genet* Nov 7;105(5):947–958. PMCID: PMC6849090.

*Sulovari A, Li R, Audano PA, Porubsky D, Vollger MR, Logsdon GA; Human Genome Structural Variation Consortium, Warren WC, Pollen AA, Chaisson MJP, **Eichler EE**. (2019). Human-specific tandem repeat expansion and differential gene expression during primate evolution. *Proc Natl Acad Sci U S A* Nov 12;116(46):23243–23253. PMCID: PMC6859368.

Wu H, Li H, Bai T, Han L, Ou J, Xun G, Zhang Y, Wang Y, Duan G, Zhao N, Chen B, Du X, Yao M, Zou X, Zhao J, Hu Z, **Eichler EE**, Guo H, Xia K. (2020). Phenotype-to-genotype approach reveals head-circumference-associated genes in an autism spectrum disorder cohort. *Clin Genet* Feb;97(2):338–346. Epub 2019 Nov 14. PMCID: PMC7307605.

*Turner TN, Wilfert AB, Bakken TE, Bernier RA, Pepper MR, Zhang Z, Torene RI, Retterer K, **Eichler EE**. (2019). Sex-based analysis of de novo variants in neurodevelopmental disorders. *Am J Hum Genet* Dec 5;105(6):1274–1285. PMCID: PMC6904808.

Li YR, Glessner JT, Coe BP, Li J, Mohebnasab M, Chang X, Connolly J, Kao C, Wei Z, Bradfield J, Kim C, Hou C, Khan M, Mentch F, Qiu H, Bakay M, Cardinale C, Lemma M, Abrams D, Bridglall-Jhingoor A, Behr M, Harrison S, Otieno G, Thomas A, Wang F, Chiavacci R, Wu L, Hadley D, Goldmuntz E, Elia J, Maris J, Grundmeier R, Devoto M, Keating B, March M, Pellagrino R, Grant SFA, Sleiman PMA, Li M, **Eichler EE**, Hakonarson H. (2020). Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. *Nat Commun* Jan 14;11(1):255. PMCID: PMC6959272.

Beighley JS, Hudac CM, Arnett AB, Peterson JL, Gerdts J, Wallace AS, Mefford HC, Hoekzema K, Turner TN, O'Roak BJ, **Eichler EE**, Bernier RA. (2020). Clinical phenotypes of carriers of mutations in CHD8 or its conserved target genes. *Biol Psychiatry* Jan 15;87(2):123–131. Epub 2019 Jul 30. PMCID: PMC6925323.

Nakamori M, Panigrahi GB, Lanni S, Gall-Duncan T, Hayakawa H, Tanaka H, Luo J, Otabe T, Li J, Sakata A, Caron MC, Joshi N, Prasolava T, Chiang K, Masson JY, Wold MS, Wang X, Lee MYWT, Huddleston J, Munson KM, Davidson S, Layeghifard M, Edward LM, Gallon R, Santibanez-Koref M, Murata A, Takahashi MP, **Eichler EE**, Shlien A, Nakatani K, Mochizuki H, Pearson CE. (2020). A slipped-CAG DNA-binding small molecule induces trinucleotide-repeat contractions in vivo. *Nat Genet* Feb;52(2):146–159. PMCID: PMC7043212.

Liu J, Shi C, Shi CC, Li W, Zhang QJ, Zhang Y, Li K, Lu HF, Shi C, Zhu ST, Xiao ZY, Nan H, Yue Y, Zhu XG, Wu Y, Hong XN, Fan GY, Tong Y, Zhang D, Mao CL, Liu YL, Hao SJ, Liu WQ, Lv MQ, Zhang HB, Liu Y, Hu-Tang GR, Wang JP, Wang JH, Sun YH, Ni SB, Chen WB, Zhang XC, Jiao YN, **Eichler EE**, Li GH, Liu X, Gao LZ. (2020). The chromosome-based rubber tree genome provides new insights into spurge genome evolution and rubber biosynthesis. *Mol Plant* Feb 3;13(2):336–350. PMID: 31838037. PMCID: N/A.

Maggiolini FAM, Mercuri L, Antonacci F, Anaclerio F, Calabrese FM, Lorusso N, L'Abbate A, Sorensen M, Giannuzzi G, **Eichler EE**, Catacchio CR, Ventura M. (2020). Evolutionary dynamics of the POTE gene family in human and nonhuman primates. *Genes* Feb 18;11(2). pii: E213. PMCID: PMC7073761.

*Vollger MR, Logsdon GA, Audano PA, Sulovari A, Porubsky D, Peluso P, Wenger AM, Concepcion GT, Kronenberg ZN, Munson KM, Baker C, Sanders AD, Spierings DCJ, Lansdorp PM, Surti U, Hunkapiller MW, **Eichler EE**. (2020). Improved assembly and variant detection of a haploid human genome using single-molecule, high-fidelity long reads. *Ann Hum Genet* Mar;84(2):125–140. doi: 10.1111/ahg.12364. Epub 2019 Nov 11. PMCID: PMC7015760.

Mirzaa GM, Chong JX, Piton A, Popp B, Foss K, Guo H, Harripaul R, Xia K, Scheck J, Aldinger KA, Sajan SA, Tang S, Bonneau D, Beck A, White J, Mahida S, Harris J, Smith-Hicks C, Hoyer J, Zweier C, Reis A, Thiel CT, Jamra RA, Zeid N, Yang A, Farach LS, Walsh L, Payne K, Rohena L, Velinov M, Ziegler A, Schaefer E, Gatinois V, Geneviève D, Simon MEH, Kohler J, Rotenberg J, Wheeler P, Larson A, Ernst ME, Akman CI, Westman R, Blanchet P, Schillaci LA, Vincent-Delorme C, Gripp KW, Mattioli F, Guyader GL, Gerard B, Mathieu-Dramard M, Morin G, Sasanfar R, Ayub M, Vasli N, Yang S, Person R, Monaghan KG, Nickerson DA, van Binsbergen E, Enns GM, Dries AM, Rowe LJ, Tsai ACH, Svihovec S, Friedman J, Agha Z, Qamar R, Rodan LH, Martinez-Agosto J, Ockeloen CW, Vincent M, Sunderland WJ, Bernstein JA; Undiagnosed Diseases Network, **Eichler EE**, Vincent JB; University of Washington Center for Mendelian Genomics (UW-CMG), Bamshad MJ. (2020). De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. *Genet Med* Mar;22(3):538–546. doi: 10.1038/s41436-019-0693-9. Epub 2019 Nov 14. PMCID: PMC7060121.

Zhang QJ, Li W, Li K, Nan H, Shi C, Zhang Y, Dai ZY, Lin YL, Yang XL, Tong Y, Zhang D, Lu C, Feng LY, Wang CF, Liu XX, Huang JA, Jiang WK, Wang XH, Zhang XC, **Eichler EE**, Liu ZH, Gao LZ. (2020). The chromosome-level reference genome of tea tree unveils recent bursts of non-autonomous LTR retrotransposons to drive genome size evolution. *Mol Plant* Jul 6;13(7):935–938. PMID: 32353626. PMCID: N/A.

Scott TM, Guo H, **Eichler EE**, Rosenfeld JA, Pang K, Liu Z, Lalani S, Bi W, Yang Y, Bacino CA, Streff H, Lewis AM, Koenig MK, Thiffault I, Bellomo A, Everman DB, Jones JR, Stevenson RE, Bernier R, Gilissen C, Pfundt R, Hiatt SM, Cooper GM, Holder JL, Scott DA. (2020). BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. *Hum Mutat* May;41(5):921–925. PMCID: PMC7262739.

Myers SM, Challman TD, Bernier R, Bourgeron T, Chung WK, Constantino JN, **Eichler EE**, Jacquemont S, Miller DT, Mitchell KJ, Zoghbi HY, Martin CL, Ledbetter DH. (2020). Insufficient evidence for "autism-specific" genes. *Am J Hum Genet* May 7;106(5):587–595. PMCID: PMC7212289.

*Porubsky D, Sanders AD, Hops W, Hsieh P, Sulovari A, Li R, Mercuri L, Sorensen M, Murali SC, Gordon D, Cantsilieris S, Pollen AA, Ventura M, Antonacci F, Marschall T, Korbel JO, **Eichler EE**. (2020). Recurrent inversion toggling and great ape genome evolution. *Nat Genet* Aug;52(8):849–858. PMCID: PMC7415573.

Hudac CM, Bove J, Barber S, Duyzend M, Wallace A, Martin CL, Ledbetter DH, Hanson E, Goin-Kochel RP, Green-Snyder L, Chung WK, **Eichler EE**, Bernier RA. (2020). Evaluating heterogeneity in ASD symptomatology, cognitive ability, and adaptive functioning among 16p11.2 CNV carriers. *Autism Res* Aug;13(8):1300–1310. PMID: 32597026. PMCID: N/A.

Miga KH, Koren S, Rhie A, Vollger MR, Gershman A, Bzikadze A, Brooks S, Howe E, Porubsky D, Logsdon GA, Schneider VA, Potapova T, Wood J, Chow W, Armstrong J, Fredrickson J, Pak E, Tigyi K, Kremitzki M, Markovic C, Maduro V, Dutra A, Bouffard GG, Chang AM, Hansen NF, Wilfert AB, Thibaud-Nissen F, Schmitt AD, Belton JM, Selvaraj S, Dennis MY, Soto DC, Sahasrabudhe R, Kaya G, Quick J, Loman NJ, Holmes N, Loose M, Surti U, Risques RA, Graves Lindsay TA, Fulton R, Hall I, Paten B, Howe K, Timp W, Young A, Mullikin JC, Pevzner PA, Gerton JL, Sullivan BA, **Eichler EE**, Phillippy AM. (2020). Telomere-to-telomere assembly of a complete human X chromosome. *Nature* Sep;585(7823):79–84. PMCID: PMC7484160.

Shafin K, Pesout T, Lorig-Roach R, Haukness M, Olsen HE, Bosworth C, Armstrong J, Tigyi K, Maurer N, Koren S, Sedlazeck FJ, Marschall T, Mayes S, Costa V, Zook JM, Liu KJ, Kilburn D, Sorensen M, Munson KM, Vollger MR, Monlong J, Garrison E, **Eichler EE**, Salama S, Haussler D, Green RE, Akeson M, Phillippy A, Miga KH, Carnevali P, Jain M, Paten B. (2020). Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. *Nat Biotechnol* Sep;38(9):1044–1053. PMCID: PMC7483855.

Cappuccio G, Sayou C, Tanno PL, Tisserant E, Bruel AL, Kennani SE, Sá J, Low KJ, Dias C, Havlovicová M, Hancarova M, **Eichler EE**, Devillard F, Moutton S, Van-Gils J, Dubourg C, Odent S, Gerard B, Piton A, Yamamoto T, Okamoto N, Firth H, Metcalfe K, Moh A, Chapman KA, Aref-Eshghi E, Kerkhof J, Torella A, Nigro V, Perrin L, Piard J, Le Guyader G, Jouan T, Thauvin-Robinet C, Duffourd Y, George-Abraham JK, Buchanan CA, Williams D, Kini U, Wilson K; Telethon Undiagnosed Diseases Program, Sousa SB, Hennekam RCM, Sadikovic B, Thevenon J, Govin J, Vitobello A, Brunetti-Pierri N. (2020). De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. *Genet Med* Oct 1;11(1):4932. PMID: 32694869. PMCID: N/A.

Course MM, Gudsnuk K, Smukowski SN, Winston K, Desai N, Ross JP, Sulovari A, Bourassa CV, Spiegelman D, Couthouis J, Yu CE, Tsuang DW, Jayadev S, Kay MA, Gitler AD, Dupre N, **Eichler EE**, Dion PA, Rouleau GA, Valdmanis PN. (2020). Evolution of a human-specific tandem repeat associated with ALS. *Am J Hum Genet* Sep 3;107(3):445–460. PMCID: PMC7477013.

*Cantsilieris S, Sunkin SM, Johnson ME, Anaclerio F, Huddleston J, Baker C, Dougherty ML, Underwood JG, Sulovari A, Hsieh P, Mao Y, Catacchio CR, Malig M, Welch AE, Sorensen M, Munson KM, Jiang W, Girirajan S, Ventura M, Lamb BT, Conlon RA, **Eichler EE**. (2020). An evolutionary driver of interspersed segmental duplications in primates. *Genome Biol* Aug 10;21(1):202. PMCID: PMC7419210.

Nurk S, Walenz BP, Rhie A, Vollger MR, Logsdon GA, Grothe R, Miga KH, **Eichler EE**, Phillippy AM, Koren S. (2020). HiCanu: accurate assembly of segmental duplications, satellites, and allelic variants from high-fidelity long reads. *Genome Res* Sep;30(9):1291–1305. PMCID: PMC7545148.

*Arnett AB, Beighley JS, Kurtz-Nelson EC, Hoekzema K, Wang T, Bernier RA, **Eichler EE**. (2020). Developmental predictors of cognitive and adaptive outcomes in genetic subtypes of autism spectrum disorder. *Autism Res* Oct;13(10):1659–1669. PMCID: PMC7861657.

*Wang T, Hoekzema K, Vecchio D, Wu H, Sulovari A, Coe BP, Gillentine MA, Wilfert AB, Perez-Jurado LA, Kvarnung M, Sleyp Y, Earl RK, Rosenfeld JA, Geisheker MR, Han L, Du B, Barnett C, Thompson E, Shaw M, Carroll R, Friend K, Catford R, Palmer EE, Zou X, Ou J, Li H, Guo H, Gerdts J, Avola E, Calabrese G, Elia M, Greco D, Lindstrand A, Nordgren A, Anderlid BM, Vandeweyer G, Van Dijck A, Van der Aa N, McKenna B, Hancarova M, Bendova S, Havlovicova M, Malerba G, Bernardina BD, Muglia P, van Haeringen A, Hoffer MJV, Franke B, Cappuccio G, Delatycki M, Lockhart PJ, Manning MA, Liu P, Scheffer IE, Brunetti-Pierri N, Rommelse N, Amaral DG, Santen GWE, Trabetti E, Sedláček Z, Michaelson JJ, Pierce K, Courchesne E, Kooy RF; SPARK Consortium, Nordenskjöld M, Romano C, Peeters H, Bernier RA, Gecz J, Xia K, **Eichler EE**. (2020). Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nat Commun* Oct 1;11(1):4932. PMCID: PMC7530681.

Rodriguez OL, Gibson WS, Parks T, Emery M, Powell J, Strahl M, Deikus G, Auckland K, **Eichler EE**, Marasco WA, Sebra R, Sharp AJ, Smith ML, Bashir A, Watson CT. (2020). A novel framework for characterizing genomic haplotype diversity in the human immunoglobulin heavy chain locus. *Front Immunol* Sep 23;11:2136. PMCID: PMC7539625.

Maggiolini FAM, Sanders AD, Shew CJ, Sulovari A, Mao Y, Puig M, Catacchio CR, Dellino M, Palmisano D, Mercuri L, Bitonto M, Porubský D, Cáceres M, **Eichler EE**, Ventura M, Dennis MY, Korbel JO, Antonacci F. (2020). Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution. *Genome Res* Nov;30(11):1680–1693. PMCID: PMC7605249.

*Guo H, Zhang Q, Dai R, Yu B, Hoekzema K, Tan J, Tan S, Jia X, Chung WK, Hernan R, Alkuraya FS, Alsulaiman A, Al-Muhaizea MA, Lesca G, Pons L, Labalme A, Laux L, Bryant E, Brown NJ, Savva E, Ayres S, Eratne D, Peeters H, Bilan F, Letienne-Cejudo L, Gilbert-Dussardier B, Ruiz-Arana IL, Merlini JM, Boizot A, Bartoloni L, Santoni F, Karlowicz D, McDonald M, Wu H, Hu Z, Chen G, Ou J, Brasch-Andersen C, Fagerberg CR, Dreyer I, Chun-Hui Tsai A, Slegesky V, McGee RB, Daniels B, Sellars EA, Carpenter LA, Schaefer B, Sacoto MJG, Begtrup A, Schnur RE, Punj S, Wentzensen IM, Rhodes L, Pan Q, Bernier RA, Chen C, **Eichler EE**, Xia K. (2020). NCKAP1 disruptive variants lead to a neurodevelopmental disorder with core features of autism. *Am J Hum Genet* Nov 5;107(5):963–976. PMCID: PMC7674997.

*Warren WC, Harris RA, Haukness M, Fiddes IT, Murali SC, Fernandes J, Dishuck PC, Storer JM, Raveendran M, Hillier LW, Porubsky D, Mao Y, Gordon D, Vollger MR, Lewis AP, Munson KM, DeVogelaere E, Armstrong J, Diekhans M, Walker JA, Tomlinson C, Graves-Lindsay TA, Kremitzki M, Salama SR, Audano PA, Escalona M, Maurer NW, Antonacci F, Mercuri L, Maggiolini FAM, Catacchio CR, Underwood JG, O'Connor DH, Sanders AD, Korbel JO, Ferguson B, Kubisch HM, Picker L, Kalin NH, Rosene D, Levine J, Abbott DH, Gray SB, Sanchez MM, Kovacs-Balint ZA, Kemnitz JW, Thomasy SM, Roberts JA, Kinnally EL, Capitanio JP, Skene JHP, Platt M, Cole SA, Green RE, Ventura M, Wiseman RW, Paten B, Batzer MA, Rogers J, **Eichler EE**. (2020). Sequence diversity analyses of an improved rhesus macaque genome enhance its biomedical utility. *Science* Dec 18;370(6523):eabc6617. PMCID: PMC7818670.

*Porubsky D, Ebert P, Audano PA, Vollger MR, Harvey WT, Marijon P, Ebler J, Munson KM, Sorensen M, Sulovari A, Haukness M, Ghareghani M; Human Genome Structural Variation Consortium, Lansdorp PM, Paten B, Devine SE, Sanders AD, Lee C, Chaisson MJP, Korbel JO, **Eichler EE**, Marschall T. (2021). Fully phased human genome assembly without parental data using single-cell strand sequencing and long reads. *Nat Biotechnol* Mar;39(3):302-308. doi: 10.1038/s41587-020-0719-5. Epub 2020 Dec 7. PMCID: PMC7954704.

Radio FC, Pang K, Ciolfi A, Levy MA, Hernández-García A, Pedace L, Pantaleoni F, Liu Z, de Boer E, Jackson A, Bruselles A, McConkey H, Stellacci E, Lo Cicero S, Motta M, Carrozzo R, Dentici ML, McWalter K, Desai M, Monaghan KG, Telegrafi A, Philippe C, Vitobello A, Au M, Grand K, Sanchez-Lara PA, Baez J, Lindstrom K, Kulch P, Sebastian J, Madan-Khetarpal S,

Roadhouse C, MacKenzie JJ, Monteleone B, Saunders CJ, Jean Cuevas JK, Cross L, Zhou D, Hartley T, Sawyer SL, Monteiro FP, Secches TV, Kok F, Schultz-Rogers LE, Macke EL, Morava E, Klee EW, Kemppainen J, Iascone M, Selicorni A, Tenconi R, Amor DJ, Pais L, Gallacher L, Turnpenny PD, Stals K, Ellard S, Cabet S, Lesca G, Pascal J, Steindl K, Ravid S, Weiss K, Castle AMR, Carter MT, Kalsner L, de Vries BBA, van Bon BW, Wevers MR, Pfundt R, Stegmann APA, Kerr B, Kingston HM, Chandler KE, Sheehan W, Elias AF, Shinde DN, Towne MC, Robin NH, Goodloe D, Vanderver A, Sherbini O, Bluske K, Hagelstrom RT, Zanus C, Faletra F, Musante L, Kurtz-Nelson EC, Earl RK, Anderlid BM, Morin G, van Slegtenhorst M, Diderich KEM, Brooks AS, Gribnau J, Boers RG, Finestra TR, Carter LB, Rauch A, Gasparini P, Boycott KM, Barakat TS, Graham JM Jr, Faivre L, Banka S, Wang T, **Eichler EE**, Priolo M, Dallapiccola B, Vissers LELM, Sadikovic B, Scott DA, Holder JL Jr, Tartaglia M. (2021). SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *Am J Hum Genet* Mar 4;108(3):502–516. PMCID: PMC8008487.

Dingemans AJM, Stremmelaar DE, Vissers LELM, Jansen S, Nabais Sá MJ, van Remortele A, Jonis N, Truijen K, van de Ven S, Ewals J, Verbruggen M, Koolen DA, Brunner HG, **Eichler EE**, Gecz J, de Vries BBA. (2021). Human disease genes website series: An international, open and dynamic library for up-to-date clinical information. *Am J Med Genet A* Apr;185(4):1039–1046. PMCID: PMC7986414.

*Ebert P, Audano PA, Zhu Q, Rodriguez-Martin B, Porubsky D, Bonder MJ, Sulovari A, Ebler J, Zhou W, Serra Mari R, Yilmaz F, Zhao X, Hsieh P, Lee J, Kumar S, Lin J, Rausch T, Chen Y, Ren J, Santamarina M, Höps W, Ashraf H, Chuang NT, Yang X, Munson KM, Lewis AP, Fairley S, Tallon LJ, Clarke WE, Basile AO, Byrska-Bishop M, Corvelo A, Evani US, Lu TY, Chaisson MJP, Chen J, Li C, Brand H, Wenger AM, Ghareghani M, Harvey WT, Raeder B, Hasenfeld P, Regier AA, Abel HJ, Hall IM, Flicek P, Stegle O, Gerstein MB, Tubio JMC, Mu Z, Li YI, Shi X, Hastie AR, Ye K, Chong Z, Sanders AD, Zody MC, Talkowski ME, Mills RE, Devine SE, Lee C, Korbel JO, Marschall T, **Eichler EE**. (2021). Haplotype-resolved diverse human genomes and integrated analysis of structural variation. *Science* Apr 2;372(6537):eabf7117. PMCID: PMC8026704.

*Gillentine MA, Wang T, Hoekzema K, Rosenfeld J, Liu P, Guo H, Kim CN, De Vries BBA, Vissers LELM, Nordenskjold M, Kvarnung M, Lindstrand A, Nordgren A, Gecz J, Iascone M, Cereda A, Scatigno A, Maitz S, Zanni G, Bertini E, Zweier C, Schuhmann S, Wiesener A, Pepper M, Panjwani H, Torti E, Abid F, Anselm I, Srivastava S, Atwal P, Bacino CA, Bhat G, Cobian K, Bird LM, Friedman J, Wright MS, Callewaert B, Petit F, Mathieu S, Afenjar A, Christensen CK, White KM, Elpeleg O, Berger I, Espineli EJ, Fagerberg C, Brasch-Andersen C, Hansen LK, Feyma T, Hughes S, Thiffault I, Sullivan B, Yan S, Keller K, Keren B, Mignot C, Kooy F, Meuwissen M, Basinger A, Kukolich M, Philips M, Ortega L, Drummond-Borg M, Lauridsen M, Sorensen K, Lehman A; CAUSES Study, Lopez-Rangel E, Levy P, Lessel D, Lotze T, Madan-Khetarpal S, Sebastian J, Vento J, Vats D, Benman LM, Mckee S, Mirzaa GM, Muss C, Pappas J, Peeters H, Romano C, Elia M, Galesi O, Simon MEH, van Gassen KLI, Simpson K, Stratton R, Syed S, Thevenon J, Palafoll IV, Vitobello A, Bournez M, Faivre L, Xia K; SPARK Consortium, Earl RK, Nowakowski T, Bernier RA, **Eichler EE**. (2021). Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Med* Apr 19;13(1):63. PMCID: PMC8056596.

*Kronenberg ZN, Rhie A, Koren S, Concepcion GT, Peluso P, Munson KM, Porubsky D, Kuhn K, Mueller KA, Low WY, Hiendleder S, Fedrigo O, Liachko I, Hall RJ, Phillippy AM, **Eichler EE**, Williams JL, Smith TPL, Jarvis ED, Sullivan ST, Kingan SB. (2021). Extended haplotype-phasing of long-read de novo genome assemblies using Hi-C. *Nat Commun* Apr 28;12(1):1935. PMCID: PMC8081726.

Zhao X, Collins RL, Lee WP, Weber AM, Jun Y, Zhu Q, Weisburd B, Huang Y, Audano PA, Wang H, Walker M, Lowther C, Fu J; Human Genome Structural Variation Consortium, Gerstein MB, Devine SE, Marschall T, Korbel JO, **Eichler EE**, Chaisson MJP, Lee C, Mills RE, Brand H, Talkowski ME. (2021). Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. *Am J Hum Genet* May 6;108(5):919–928. PMCID: PMC8206509.

*Logsdon GA, Vollger MR, Hsieh P, Mao Y, Liskovykh MA, Koren S, Nurk S, Mercuri L, Dishuck PC, Rhie A, de Lima LG, Dvorkina T, Porubsky D, Harvey WT, Mikheenko A, Bzikadze AV, Kremitzki M, Graves-Lindsay TA, Jain C, Hoekzema K, Murali SC, Munson KM, Baker C, Sorensen M, Lewis AM, Surti U, Gerton JL, Larionov V, Ventura M, Miga KH, Phillippy AM, **Eichler EE**. (2021). The structure, function and evolution of a complete human chromosome 8. *Nature* May;593(7857):101–107. PMCID: PMC8099727.

*Mao Y, Catacchio CR, Hillier LW, Porubsky D, Li R, Sulovari A, Fernandes JD, Montinaro F, Gordon DS, Storer JM, Haukness M, Fiddes IT, Murali SC, Dishuck PC, Hsieh P, Harvey WT, Audano PA, Mercuri L, Piccolo I, Antonacci F, Munson KM, Lewis AP, Baker C, Underwood JG, Hoekzema K, Huang TH, Sorensen M, Walker JA, Hoffman J, Thibaud-Nissen F, Salama SR, Pang AWC, Lee J, Hastie AR, Paten B, Batzer MA, Diekhans M, Ventura M, **Eichler EE**. (2021). A high-quality bonobo genome refines the analysis of hominid evolution. *Nature* Jun;594(7861):77–81. PMCID: PMC8172381.

Earl RK, Ward T, Gerdts J, **Eichler EE**, Bernier RA, Hudac CM. (2021). Sleep problems in children with ASD and gene disrupting mutations. *J Genet Psychol* Sep-Oct;182(5):317–334. PMCID: PMC8445595.

Coll-Tané M, Gong NN, Belfer SJ, van Renssen LV, Kurtz-Nelson EC, Szuperak M, Eidhof I, van Reijmersdal B, Terwindt I, Durkin J, Verheij MMM, Kim CN, Hudac CM, Nowakowski TJ, Bernier RA, Pillen S, Earl RK, **Eichler EE**, Kleefstra T, Kayser MS, Schenck A. (2021). The CHD8/CHD7/Kismet family links blood-brain barrier glia and serotonin to ASD-associated sleep defects. *Sci Adv* Jun 4;7(23):eabe2626. PMCID: PMC8177706.

Cousin MA, Creighton BA, Breau KA, Spillmann RC, Torti E, Dontu S, Tripathi S, Ajit D, Edwards RJ, Afriyie S, Bay JC, Harper KM, Beltran AA, Munoz LJ, Falcon Rodriguez L, Stankewich MC, Person RE, Si Y, Normand EA, Blevins A, May AS, Bier L, Aggarwal V, Mancini GMS, van Slegtenhorst MA, Cremer K, Becker J, Engels H, Aretz S, MacKenzie JJ, Brilstra E, van Gassen

KLI, van Jaarsveld RH, Oegema R, Parsons GM, Mark P, Helbig I, McKeown SE, Stratton R, Cogne B, Isidor B, Cacheiro P, Smedley D, Firth HV, Bierhals T, Kloth K, Weiss D, Fairley C, Shieh JT, Kritzer A, Jayakar P, Kurtz-Nelson E, Bernier RA, Wang T, **Eichler EE**, van de Laar IMBH, McConkie-Rosell A, McDonald MT, Kemppainen J, Lanpher BC, Schultz-Rogers LE, Gunderson LB, Pichurin PN, Yoon G, Zech M, Jech R, Winkelmann J; Undiagnosed Diseases Network; Genomics England Research Consortium, Beltran AS, Zimmermann MT, Temple B, Moy SS, Klee EW, Tan QK, Lorenzo DN. (2021). Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. *Nat Genet* Jul;53(7):1006-1021. PMCID: PMC8273149.

*Miller DE, Sulovari A, Wang T, Loucks H, Hoekzema K, Munson KM, Lewis AP, Fuerte EPA, Paschal CR, Walsh T, Thies J, Bennett JT, Glass I, Dipple KM, Patterson K, Bonkowski ES, Nelson Z, Squire A, Sikes M, Beckman E, Bennett RL, Earl D, Lee W, Alikmets R, Perlman SJ, Chow P, Hing AV, Wenger TL, Adam MP, Sun A, Lam C, Chang I, Zou X, Austin SL, Huggins E, Safi A, Iyengar AK, Reddy TE, Majoros WH, Allen AS, Crawford GE, Kishnani PS, University of Washington for Mendelian Genomics, King MC, Cherry T, Chong J, Bamshad MJ, Nickerson DA, Mefford HC, Doherty D, **Eichler EE**. (2021). Targeted long-read sequencing identifies missing disease-causing variation. *AJHG* Aug 5;108(8):1436–1449. PMCID: PMC8387463.

Course MM, Sulovari A, Gudsnuk K, **Eichler EE**, Valdmanis PN. (2021). Characterizing nucleotide variation and expansion dynamics in human-specific variable number tandem repeats. *Genome Res* Aug;31(8):1313–1324. PMCID: PMC8327921.

*Wilfert AB, Turner TN, Murali SC, Hsieh P, Sulovari A, Wang T, Coe BP, Guo H, Hoekzema K, Bakken TE, Winterkorn LH, Evani US, Byrska-Bishop M, Earl RK, Bernier RA; SPARK Consortium, Zody MC, **Eichler EE**. (2021). Recent ultra-rare inherited variants implicate new autism candidate risk genes. *Nat Genet* Aug;53(8):1125–1134. PMCID: PMC8459613.

*Hsieh P, Dang V, Vollger MR, Mao Y, Huang TH, Dishuck PC, Baker C, Cantsilieris S, Lewis AP, Munson KM, Sorensen M, Welch AE, Underwood JG, **Eichler EE**. (2021). Evidence for opposing selective forces operating on human-specific duplicated TCAF genes in Neanderthals and humans. *Nat Commun* Aug 25;12(1):5118. PMCID: PMC8387397.

Goldmann JM, Hampstead JE, Wong WSW, Wilfert AB, Turner TN, Jonker MA, Bernier R, Huynen MA, **Eichler EE**, Veltman JA, Maxwell GL, Gilissen C. (2021). Differences in the number of de novo mutations between individuals are due to small family-specific effects and stochasticity. *Genome Res* Sep;31(9):1513–1518. PMCID: PMC8415378.

Kurtz-Nelson EC, Tham SW, Ahlers K, Cho D, Wallace AS, **Eichler EE**, Bernier RA, Earl RK. (2021). Brief Report: Associations between self-injurious behaviors and abdominal pain among individuals with ASD-associated disruptive mutations. *J Autism Dev Disord* Sep;51(9):3365-3373. doi: 10.1007/s10803-020-04774-z. Epub 2020 Nov 11. PMCID: PMC8110605.

Ziffra RS, Kim CN, Ross JM, Wilfert A, Turner TN, Haeussler M, Casella AM, Przytycki PF, Keough KC, Shin D, Bogdanoff D, Kreimer A, Pollard KS, Ament SA, **Eichler EE**, Ahituv N, Nowakowski TJ. (2021). Single-cell epigenomics reveals mechanisms of human cortical development. *Nature* Oct;598(7879):205–213. PMCID: PMC8494642.

Mouakkad-Montoya L, Murata MM, Sulovari A, Suzuki R, Osia B, Malkova A, Katsumata M, Giuliano AE, **Eichler EE**, Tanaka H. (2021). Quantitative assessment reveals the dominance of duplicated sequences in germline-derived extrachromosomal circular DNA. *Proc Natl Acad Sci U S A* Nov 23;118(47):e2102842118. PMCID: PMC8617514.

Giannuzzi G, Logsdon GA, Chatron N, Miller DE, Reversat J, Munson KM, Hoekzema K, Bonnet-Dupeyron MN, Rollat-Farnier PA, Baker CA, Sanlaville D, **Eichler EE**, Schluth-Bolard C, Reymond A. (2021). Alpha satellite insertion close to an ancestral centromeric region. *Mol Biol Evol* Dec 9;38(12):5576–5587. PMCID: PMC8662618.

Johansson PA, Brattås PL, Douse CH, Hsieh P, Adami A, Pontis J, Grassi D, Garza R, Sozzi E, Cataldo R, Jönsson ME, Atacho DAM, Pircs K, Eren F, Sharma Y, Johansson J, Fiorenzano A, Parmar M, Fex M, Trono D, **Eichler EE**, Jakobsson J. (2022). A cis-acting structural variation at the ZNF558 locus controls a gene regulatory network in human brain development. *Cell Stem Cell* Jan 6;29(1):52–69.e8. PMID: 34624206. PMCID: N/A – Cell Press Open Access.

Fanjul-Fernández M, Brown NJ, Hickey P, Diakumis P, Rafehi H, Bozaoglu K, Green CC, Rattray A, Young S, Alhuzaimi D, Mountford HS, Gillies G, Lukic V, Vick T, Finlay K, Coe BP, **Eichler EE**, Delatycki MB, Wilson SJ, Bahlo M, Scheffer IE, Lockhart PJ. (2022). A family study implicates GBE1 in the etiology of autism spectrum disorder. *Hum Mutat* Jan;43(1):16–29. PMCID: PMC8720068.

*Vollger MR, Kerpedjiev P, Phillippy AM, **Eichler EE**. (2022). StainedGlass: Interactive visualization of massive tandem repeat structures with identity heatmaps. *Bioinformatics* Jan 10;38(7):2049–51. PMCID: PMC8963321.

Lin J, Yang X, Kosters W, Xu T, Jia Y, Wang S, Zhu Q, Ryan M, Guo L, Zhang C, Lee C, Devine SE, **Eichler EE**, Ye K; Human Genome Structural Variation Consortium. (2022). Mako: A graph-based pattern growth approach to detect complex structural variants. *Genomics Proteomics Bioinformatics* Feb;20(1):205–218. PMCID: PMC9510932.

Gofin Y, Wang T, Gillentine MA, Scott TM, Berry AM, Azamian MS, Genetti C, Agrawal PB, Picker J, Wojcik MH, Delgado MR, Lynch SA, Scherer SW, Howe JL, Bacino CA, DiTroia S, VanNoy GE, O'Donnell-Luria A, Lalani SR, Graf WD, Rosenfeld JA, **Eichler EE**, Earl RK, Scott DA. (2022). Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. *Hum Mutat* Apr;43(4):461–470. PMCID: PMC8960338.

*Noyes MD, Harvey WT, Porubsky D, Sulovari A, Li R, Rose NR, Audano PA, Munson KM, Lewis AP, Hoekzema K, Mantere T, Graves-Lindsay TA, Sanders AD, Goodwin S, Kramer M, Mokrab Y, Zody MC, Hoischen A, Korbel JO, McCombie WR, **Eichler EE**. (2022). Familial long-read sequencing increases yield of de novo mutations. *Am J Hum Genet.* Apr 7;109(4):631–646. PMCID: PMC9069071.

Altemose N, Logsdon GA, Bzikadze AV, Sidhwani P, Langley SA, Caldas GV, Hoyt SJ, Uralsky L, Ryabov FD, Shew CJ, Sauria MEG, Borchers M, Gershman A, Mikheenko A, Shepelev VA, Dvorkina T, Kunyavskaya O, Vollger MR, Rhie A, McCartney AM, Asri M, Lorig-Roach R, Shafin K, Lucas JK, Aganezov S, Olson D, de Lima LG, Potapova T, Hartley GA, Haukness M, Kerpedjiev P, Gusev F, Tigyi K, Brooks S, Young A, Nurk S, Koren S, Salama SR, Paten B, Rogaev EI, Streets A, Karpen GH, Dernburg AF, Sullivan BA, Straight AF, Wheeler TJ, Gerton JL, **Eichler EE**, Phillippy AM, Timp W, Dennis MY, O'Neill RJ, Zook JM, Schatz MC, Pevzner PA, Diekhans M, Langley CH, Alexandrov IA, Miga KH. (2022). Complete genomic and epigenetic maps of human centromeres. *Science* Apr;376(6588):eabl4178. PMCID: PMC9233505.

Gershman A, Sauria MEG, Guitart X, Vollger MR, Hook PW, Hoyt SJ, Jain M, Shumate A, Razaghi R, Koren S, Altemose N, Caldas GV, Logsdon GA, Rhie A, **Eichler EE**, Schatz MC, O'Neill RJ, Phillippy AM, Miga KH, Timp W. (2022). Epigenetic patterns in a complete human genome. *Science* Apr;376(6588):eabj5089. PMCID: PMC9170183.

Hoyt SJ, Storer JM, Hartley GA, Grady PGS, Gershman A, de Lima LG, Limouse C, Halabian R, Wojenski L, Rodriguez M, Altemose N, Rhie A, Core LJ, Gerton JL, Makalowski W, Olson D, Rosen J, Smit AFA, Straight AF, Vollger MR, Wheeler TJ, Schatz MC, **Eichler EE**, Phillippy AM, Timp W, Miga KH, O'Neill RJ. (2022). From telomere to telomere: The transcriptional and epigenetic state of human repeat elements. *Science* Apr;376(6588):eabk3112. PMCID: PMC9301658.

*Nurk S, Koren S, Rhie A, Rautiainen M, Bzikadze AV, Mikheenko A, Vollger MR, Altemose N, Uralsky L, Gershman A, Aganezov S, Hoyt SJ, Diekhans M, Logsdon GA, Alonge M, Antonarakis SE, Borchers M, Bouffard GG, Brooks SY, Caldas GV, Chen NC, Cheng H, Chin CS, Chow W, de Lima LG, Dishuck PC, Durbin R, Dvorkina T, Fiddes IT, Formenti G, Fulton RS, Fungtammasan A, Garrison E, Grady PGS, Graves-Lindsay TA, Hall IM, Hansen NF, Hartley GA, Haukness M, Howe K, Hunkapiller MW, Jain C, Jain M, Jarvis ED, Kerpedjiev P, Kirsche M, Kolmogorov M, Korlach J, Kremitzki M, Li H, Maduro VV, Marschall T, McCartney AM, McDaniel J, Miller DE, Mullikin JC, Myers EW, Olson ND, Paten B, Peluso P, Pevzner PA, Porubsky D, Potapova T, Rogaev EI, Rosenfeld JA, Salzberg SL, Schneider VA, Sedlazeck FJ, Shafin K, Shew CJ, Shumate A, Sims Y, Smit AFA, Soto DC, Sović I, Storer JM, Streets A, Sullivan BA, Thibaud-Nissen F, Torrance J, Wagner J, Walenz BP, Wenger A, Wood JMD, Xiao C, Yan SM, Young AC, Zarate S, Surti U, McCoy RC, Dennis MY, Alexandrov IA, Gerton JL, O'Neill RJ, Timp W, Zook JM, Schatz MC, **Eichler EE**, Miga KH, Phillippy AM. (2022). The complete sequence of a human genome. *Science* Apr;376(6588):44–53. PMCID: PMC9186530.

*Vollger MR, Guitart X, Dishuck PC, Mercuri L, Harvey WT, Gershman A, Diekhans M, Sulovari A, Munson KM, Lewis AP, Hoekzema K, Porubsky D, Li R, Nurk S, Koren S, Miga KH, Phillippy AM, Timp W, Ventura M, **Eichler EE**. (2022). Segmental duplications and their variation in a complete human genome. *Science* Apr;376(6588):eabj6965. PMCID: PMC8979283.

Ebler J, Ebert P, Clarke WE, Rausch T, Audano PA, Houwaart T, Mao Y, Korbel JO, **Eichler EE**, Zody MC, Dilthey AT, Marschall T. (2022). Pangenome-based genome inference allows efficient and accurate genotyping across a wide spectrum of variant classes. *Nat Genet* Apr;54(4):518–525. PMCID: PMC9005351.

Miller DE, Lee L, Galey M, Kandhaya-Pillai R, Tischkowitz M, Amalnath D, Vithlani A, Yokote K, Kato H, Maezawa Y, Takada-Watanabe A, Takemoto M, Martin GM, **Eichler EE**, Hisama FM, Oshima J. (2022). Targeted long-read sequencing identifies missing pathogenic variants in unsolved Werner syndrome cases. *J Med Genet* May 9;59(11):1087–1094. PMCID: PMC9613861.

Ahring PK, Liao VWY, Gardella E, Johannesen KM, Krey I, Selmer KK, Stadheim BF, Davis H, Peinhardt C, Koko M, Coorg RK, Syrbe S, Bertsche A, Santiago-Sim T, Diemer T, Fenger CD, Platzer K, **Eichler EE**, Lerche H, Lemke JR, Chebib M, Møller RS. (2022). Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. *Brain* May 24;145(4):1299–1309. PMCID: PMC9630717.

*Porubsky D, Höps W, Ashraf H, Hsieh P, Rodriguez-Martin B, Yilmaz F, Ebler J, Hallast P, Maria Maggiolini FA, Harvey WT, Henning B, Audano PA, Gordon DS, Ebert P, Hasenfeld P, Benito E, Zhu Q; Human Genome Structural Variation Consortium (HGSVC), Lee C, Antonacci F, Steinrücken M, Beck CR, Sanders AD, Marschall T, **Eichler EE**, Korbel JO. (2022). Recurrent inversion polymorphisms in humans associate with genetic instability and genomic disorders. *Cell* May 26;185(11):1986-2005.e26. PMCID: PMC9563103.

*Cingöz S, Soydemir D, Öner TÖ, Karaca E, Özden B, Kurul SH, Bayram E; University of Washington Center for Mendelian Genomics, Coe BP, Nickerson DA, **Eichler EE**. (2022). Novel biallelic variants affecting the OTU domain of the gene OTUD6B associate with severe intellectual disability syndrome and molecular dynamics simulations. *Eur J Med Genet* Jun;65(6):104497. PMCID: PMC9448893.

Giannuzzi G, Chatron N, Mannik K, Auwerx C, Pradervand S, Willemin G, Hoekzema K, Nuttle X, Chrast J, Sadler MC, Porcu E, Herault Y, Isidor B, Gilbert-Dussardier B, **Eichler EE**, Kutalik Z, Reymond A. (2022). Possible association of 16p11.2 copy number variation with altered lymphocyte and neutrophil counts. *NPJ Genom Med* Jun 17;7(1):38. PMCID: PMC9205872.

Gaik M, Kojic M, Stegeman MR, Öncü-Öner T, Kościelniak A, Jones A, Mohamed A, Chau PYS, Sharmin S, Chramiec-Głąbik A, Indyka P, Rawski M, Biela A, Dobosz D, Millar A, Chau V, Ünalp A, Piper M, Bellingham MC, **Eichler EE**, Nickerson DA, Güleryüz H, Abbassi NEH, Jazgar K, Davis MJ, Mercimek-Andrews S, Cingöz S, Wainwright BJ, Glatt S. (2022). Functional

divergence of the two Elongator subcomplexes during neurodevelopment. *EMBO Mol Med* Jul 7;14(7):e15608. PMCID: PMC9260213.

van der Sluijs PJ, Joosten M, Alby C, Attié-Bitach T, Gilmore K, Dubourg C, Fradin M, Wang T, Kurtz-Nelson EC, Ahlers KP, Arts P, Barnett CP, Ashfaq M, Baban A, van den Born M, Borrie S, Busa T, Byrne A, Carriero M, Cesario C, Chong K, Cueto-González AM, Dempsey JC, Diderich KEM, Doherty D, Farholt S, Gerkes EH, Gorokhova S, Govaerts LCP, Gregersen PA, Hickey SE, Lefebvre M, Mari F, Martinovic J, Northrup H, O'Leary M, Parbhoo K, Patrier S, Popp B, Santos-Simarro F, Stoltenburg C, Thauvin-Robinet C, Thompson E, Vulto-van Silfhout AT, Zahir FR, Scott HS, Earl RK, **Eichler EE**, Vora NL, Wilnai Y, Giordano JL, Wapner RJ, Rosenfeld JA, Haak MC, Santen GWE. (2022). Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. *Genet Med* Aug;24(8):1753–1760. PMCID: PMC9378544.

Jia X, Zhang S, Tan S, Du B, He M, Qin H, Chen J, Duan X, Luo J, Chen F, Ouyang L, Wang J, Chen G, Yu B, Zhang G, Zhang Z, Lyu Y, Huang Y, Jiao J, Chen JYH, Swoboda KJ, Agolini E, Novelli A, Leoni C, Zampino G, Cappuccio G, Brunetti-Pierri N, Gerard B, Ginglinger E, Richer J, McMillan H, White-Brown A, Hoekzema K, Bernier RA, Kurtz-Nelson EC, Earl RK, Meddens C, Alders M, Fuchs M, Caumes R, Brunelle P, Smol T, Kuehl R, Day-Salvatore DL, Monaghan KG, Morrow MM, **Eichler EE**, Hu Z, Yuan L, Tan J, Xia K, Shen Y, Guo H. (2022). De novo variants in genes regulating stress granule assembly associate with neurodevelopmental disorders. *Sci Adv* Aug 19;8(33):eabo7112. PMCID: PMC9385150.

Zhou X, Feliciano P, Shu C, Wang T, Astrovskaya I, Hall JB, Obiajulu JU, Wright JR, Murali SC, Xu SX, Brueggeman L, Thomas TR, Marchenko O, Fleisch C, Barns SD, Snyder LG, Han B, Chang TS, Turner TN, Harvey WT, Nishida A, O'Roak BJ, Geschwind DH; SPARK Consortium, Michaelson JJ, Volfovsky N, **Eichler EE**, Shen Y, Chung WK. (2022). Integrating de novo and inherited variants in 42,607 autism cases identifies mutations in new moderate-risk genes. *Nat Genet* Sep;54(9):1305–1319. PMCID: PMC9470534.

Miller DE, Hanna P, Galey M, Reyes M, Linglart A, **Eichler EE**, Jüppner H. (2022). Targeted long-read sequencing identifies a retrotransposon insertion as a cause of altered GNAS exon A/B methylation in a family with autosomal dominant pseudohypoparathyroidism type 1b (PHP1B). *J Bone Miner Res* Sep;37(9):1711–1719. PMCID: PMC9474630.

Dingemans AJM, Truijen KMG, van de Ven S, Bernier R, Bongers EMHF, Bouman A, de Graaff-Herder L, **Eichler EE**, Gerkes EH, De Geus CM, van Hagen JM, Jansen PR, Kerkhof J, Kievit AJA, Kleefstra T, Maas SM, de Man SA, McConkey H, Patterson WG, Dobson AT, Prijoles EJ, Sadikovic B, Relator R, Stevenson RE, Stumpel CTRM, Heijligers M, Stuurman KE, Löhner K, Zeidler S, Lee JA, Lindy A, Zou F, Tedder ML, Vissers LELM, de Vries BBA. (2022). The phenotypic spectrum and genotype-phenotype correlations in 106 patients with variants in major autism gene CHD8. *Transl Psychiatry* Oct 1;12(1):421. PMCID: PMC9526704.

Chen G, Yu B, Tan S, Tan J, Jia X, Zhang Q, Zhang X, Jiang Q, Hua Y, Han Y, Luo S, Hoekzema K, Bernier RA, Earl RK, Kurtz-Nelson EC, Idleburg MJ, Khetarpal SM, Clark R, Sebastian J, Fernandez-Jaen A, Alvarez S, King SD, Ramos LL, Santos MLS, Martin DM, Brooks D, Symonds JD, Cutcutache I, Pan Q, Hu Z, Yuan L, **Eichler EE**, Xia K, Guo H. (2022). GIGYF1 disruption associates with autism and impaired IGF-1R signaling. *J Clin Invest* Oct 3;132(19):e159806. PMCID: PMC9525121.

Jarvis ED, Formenti G, Rhie A, Guarracino A, Yang C, Wood J, Tracey A, Thibaud-Nissen F, Vollger MR, Porubsky D, Cheng H, Asri M, Logsdon GA, Carnevali P, Chaisson MJP, Chin CS, Cody S, Collins J, Ebert P, Escalona M, Fedrigo O, Fulton RS, Fulton LL, Garg S, Gerton JL, Ghurye J, Granat A, Green RE, Harvey W, Hasenfeld P, Hastie A, Haukness M, Jaeger EB, Jain M, Kirsche M, Kolmogorov M, Korbel JO, Koren S, Korlach J, Lee J, Li D, Lindsay T, Lucas J, Luo F, Marschall T, Mitchell MW, McDaniel J, Nie F, Olsen HE, Olson ND, Pesout T, Potapova T, Puiu D, Regier A, Ruan J, Salzberg SL, Sanders AD, Schatz MC, Schmitt A, Schneider VA, Selvaraj S, Shafin K, Shumate A, Stitziel NO, Stober C, Torrance J, Wagner J, Wang J, Wenger A, Xiao C, Zimin AV, Zhang G, Wang T, Li H, Garrison E, Haussler D, Hall I, Zook JM, **Eichler EE**, Phillippy AM, Paten B, Howe K, Miga KH; Human Pangenome Reference Consortium. (2022). Semi-automated assembly of high-quality diploid human reference genomes. *Nature* Nov;611(7936):519–531. PMCID: PMC9668749.

*Gillentine MA, Wang T, **Eichler EE**. (2022). Estimating the prevalence of de novo monogenic neurodevelopmental disorders from large cohort studies. *Biomedicines* Nov 9;10(11):2865. PMCID: PMC9687899.

*Wang T, Kim CN, Bakken TE, Gillentine MA, Henning B, Mao Y, Gilissen C; SPARK Consortium, Nowakowski TJ, **Eichler EE**. (2022). Integrated gene analyses of de novo variants from 46,612 trios with autism and developmental disorders. *Proc Natl Acad Sci U S A* Nov 15;119(46):e2203491119. PMCID: PMC9674258.

Gibson WS, Rodriguez OL, Shields K, Silver CA, Dorgham A, Emery M, Deikus G, Sebra R, **Eichler EE**, Bashir A, Smith ML, Watson CT. (2022). Characterization of the immunoglobulin lambda chain locus from diverse populations reveals extensive genetic variation. *Genes Immun* Dec 21. doi: 10.1038/s41435-022-00188-2. Online ahead of print. PMID: 36539592. PMC Journal: PMCID In Process.

*Dishuck PC, Rozanski AN, Logsdon GA, Porubsky D, **Eichler EE**. (2023). GAVISUNK: Genome assembly validation via inter-SUNK distances in Oxford Nanopore reads. *Bioinformatics* Jan 1;39(1):btac714. PMCID: PMC9805576.

Bao B, Zahiri J, Gazestani VH, Lopez L, Xiao Y, Kim R, Wen TH, Chiang AWT, Nalabolu S, Pierce K, Robasky K, Wang T, Hoekzema K, **Eichler EE**, Lewis NE, Courchesne E. (2023). A predictive ensemble classifier for the gene expression diagnosis of ASD at ages 1 to 4 years. *Mol Psychiatry* Feb;28(2):822–833. Epub 2022 Oct 20. PMCID: PMC9908553.

Jun G, English AC, Metcalf GA, Yang J, Chaisson MJ, Pankratz N, Menon VK, Salerno WJ, Krasheninina O, Smith AV, Lane JA, Blackwell T, Kang HM, Salvi S, Meng Q, Shen H, Pasham D, Bhamidipati S, Kottapalli K, Arnett DK, Ashley-Koch A, Auer PL, Beutel KM, Bis JC, Blangero J, Bowden DW, Brody JA, Cade BE, Chen YI, Cho MH, Curran JE, Fornage M, Freedman BI, Fingerlin T, Gelb BD, Hou L, Hung YJ, Kane JP, Kaplan R, Kim W, Loos RJF, Marcus GM, Mathias RA, McGarvey ST, Montgomery C, Naseri T, Nouraie SM, Preuss MH, Palmer ND, Peyser PA, Raffield LM, Ratan A, Redline S, Reupena S, Rotter JI, Rich SS, Rienstra M, Ruczinski I, Sankaran VG, Schwartz DA, Seidman CE, Seidman JG, Silverman EK, Smith JA, Stilp A, Taylor KD, Telen MJ, Weiss ST, Williams LK, Wu B, Yanek LR, Zhang Y, Lasky-Su J, Gingras MC, Dutcher SK, **Eichler EE**, Gabriel S, Germer S, Kim R, Viaud-Martinez KA, Nickerson DA; NHLBI Trans-Omics for Precision Medicine (TOPMed) Consortium; Luo J, Reiner A, Gibbs RA, Boerwinkle E, Abecasis G, Sedlazeck FJ. (2023). Structural variation across 138,134 samples in the TOPMed consortium. *bioRxiv* Jan 25:2023.01.25.525428. doi: 10.1101/2023.01.25.525428. Preprint. PMCID: PMC9900832.

Rautiainen M, Nurk S, Walenz BP, Logsdon GA, Porubsky D, Rhie A, **Eichler EE**, Phillippy AM, Koren S. (2023) Telomere-to-telomere assembly of diploid chromosomes with Verkko. *Nat Biotechnol* Feb 16. doi: 10.1038/s41587-023-01662-6. Online ahead of print. PMID: 36797493.

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), Zwieq 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, Ordonez 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. PMCID: 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. PMCID: 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. PMCID: PMC2680341.

McKernan KJ, Peckham HE, Costa G, McLaughlin S, Tsung E, Fu Y, Clouser C, Dunkan 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. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: 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, Nemesh 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. PMCID: PMC3077050.

Scally A, Dutheil 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. PMCID: 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. PMCID: PMC3498939.

1000 Genomes Project Consortium, Abecasis GR, Auton A, Brooks LD, DePristo MA, Durbin RM, Handsaker RE, Kang HM, Marth GT, McVean GA. (2012). An integrated map of genetic variation from 1,092 human genomes. *Nature* Nov 1;491(7422):56–65. §**Eichler EE** 1000 Genomes Project Consortium Steering Committee Member and Analysis and Structural Variation Groups. PMCID: PMC3498066.

Prüfer K, Racimo F, Patterson N, Jay F, Sankararaman S, Sawyer S, Heinze A, Renaud G, Sudmant PH, de Filippo C, Li H, Mallick S, Dannemann M, Fu Q, Kircher M, Kuhlwilm M, Lachmann M, Meyer M, Ongyerth M, Siebauer M, Theunert C, Tandon A, Moorjani P, Pickrell J, Mullikin JC, Vohr SH, Green RE, Hellmann I, Johnson PL, Blanche H, Cann H, Kitzman JO, Shendure J, **Eichler EE**, Lein ES, Bakken TE, Golovanova LV, Doronichev VB, Shunkov MV, Derevianko AP, Viola B, Slatkin M, Reich D, Kelso J, Pääbo S. (2014). The complete genome sequence of a Neanderthal from the Altai Mountains. *Nature* Jan 2;505(7481):43–49. Epub 2013 Dec 18. PMCID: 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. §**Eichler EE** Marmoset Genome Sequencing and Analysis Consortium Member. PMCID: 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, Nevonen KA, O'Bleness M, Ochis C, Odom 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. PMCID: PMC4249732.

EuroEPINOMICS-RES Consortium; Epilepsy Phenome/Genome Project; Epi4K Consortium. (2014). De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. *Am J Hum Genet* Oct 2;95(4):360–370. §**Eichler EE** Epi4K Consortium Member. PMCID: PMC4185114.

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, Kutyavin T, Haugen E, Dunn D, Reynolds AP, Neph S, Humbert R, Hansen RS, De Bruijn M, Selleri L, Rudensky A, Josefowicz S, Samstein R, **Eichler EE**, Orkin SH, Levasseur D, Papayannopoulou T, Chang KH, Skoultchi A, Gosh S, Disteche 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. PMCID: 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, Selleri 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. PMCID: PMC4337786.

Epilepsy Phenome/Genome Project Epi4K Consortium. (2015). Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. *Ann Neurol* Aug;78(2):323–328. §**Eichler EE** Epi4K Consortium Member; Initial Design of Epi4K & contributing author. PMCID: PMC4646089.

1000 Genomes Project Consortium, Auton A, Brooks LD, Durbin RM, Garrison EP, Kang HM, Korbel JO, Marchini JL, McCarthy S, McVean GA, Abecasis GR. (2015). A global reference for human genetic variation. *Nature* Oct 1;526(7571):68–74. §**Eichler EE** 1000 Genomes Project Consortium Steering Committee Member and Analysis and Structural Variation Groups. PMCID: PMC4750478.

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. §**Eichler EE** Epi4K Consortium Member. PMCID: PMC4974067.

Rogers J, Raveendran M, Harris RA, Mailund T, Leppala K, Athanasiadis G, Schierup MH, Cheng J, Munch K, Walker JA, Konkel MK, Jordan V, Steely CJ, Beckstrom TO, Bergey C, Burrell A, Schrempf D, Noll A, Kothe M, Kopp GH, Liu Y, Murali S, Billis K,

Martin FJ, Muffato M, Cox L, Else J, Disotell T, Muzny DM, Phillips-Conroy J, Aken B, **Eichler EE**, Marques-Bonet T, Kosiol C, Batzer MA, Hahn MW, Tung J, Zinner D, Roos C, Jolly CJ, Gibbs RA, Worley KC; Baboon Genome Analysis Consortium. (2019). The comparative genomics and complex population history of Papio baboons. *Sci Adv* Jan 30;5(1):eaau6947. PMCID: PMC6401983.

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. PMCID: PMC2756412.

*Mefford HC, **Eichler EE**. (2009). Duplication hotspots, rare genomic disorders, and common disease. *Curr Opin Genet Dev* Jun;19(3):196–204. PMCID: PMC2746670.

*Marques-Bonet T, Ryder OA, **Eichler EE**. (2009). Sequencing primate genomes: What have we learned? *Annu Rev Genomics Hum Genet* 10:355–386. PMCID: PMC6662594.

*Marques-Bonet T, **Eichler EE**. (2009). The evolution of human segmental duplications and the core duplicon hypothesis. *Cold Spring Harb Symp Quant Biol* 74:355–362. PMCID: PMC4114149.

*Marques-Bonet T, Girirajan S, **Eichler EE**. (2009). The origins and impact of primate segmental duplications. *Trends Genet* Oct;25(10):443–454. PMCID: PMC2847396.

Manolio TA, Collins FS, Cox NJ, Goldstein DB, Hindorff 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. PMCID: 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. PMCID: 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. PMCID: PMC2942068.

*Girirajan S, **Eichler EE**. (2010). Phenotypic variability and genetic susceptibility to genomic disorders. *Hum Mol Genet* Oct 15;19(R2):R176–187. PMCID: 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. PMCID: PMC4108431.

*Girirajan S, Campbell CD, **Eichler EE**. (2011). Human copy number variation and complex genetic disease. *Annu Rev Genet* 45:203–226. PMCID: PMC6662611.

*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. PMCID: PMC4114147.

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

*Campbell CD, **Eichler EE**. (2013). Properties and rates of germline mutations in humans. *Trends Genet* Oct;29(10):575–584. PMCID: 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. PMCID: 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. PMCID: 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. PMCID: PMC4077789.

*Nuttle 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. PMCID: 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. PMCID: 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. PMCID: PMC4745987.

*van Bon BWM, Coe BP, de Vries BBA, **Eichler EE**. (2015). DYRK1A-related intellectual disability syndrome. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2015.

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

*Dennis MY, **Eichler EE**. (2016). Human adaptation and evolution by segmental duplication. *Curr Opin Genet Dev* Dec;41:44–52. PMCID: 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. PMCID: PMC5484527.

*Wilfert AB, Sulovari A, Turner TN, Coe BP, **Eichler EE**. (2017). Recurrent de novo mutations in neurodevelopmental disorders: Properties and clinical implications. *Genome Med* Nov 27;9(1):101. PMCID: PMC5704398.

*Turner TN, **Eichler EE**. (2019). The role of de novo noncoding regulatory mutations in neurodevelopmental disorders. *Trends Neurosci* Feb;42(2):115-127. Epub 2018 Dec 15. PMCID: PMC6382467.

***Eichler EE**. (2019). Genetic variation, comparative genomics, and the diagnosis of disease. *N Engl J Med* Jul 4;381(1):64–74. PMCID: PMC6681822.

*Logsdon GA, Vollger MR, **Eichler EE**. (2020). Long-read human genome sequencing and its applications. *Nat Rev Genet* Oct;21(10):597–614. PMCID: PMC7877196.

Eichler EE. (2021). 2020 William Allan Award introduction: Mary-Claire King. *Am J Hum Genet* Mar 4;108(3):383–385. PMCID: PMC8175867.

*Logsdon GA, **Eichler EE**. (2021) Mining the gaps of chromosome 8. *Nature* May 14. doi: 10.1038/d41586-021-01095-8. Online ahead of print. PMCID: PMC8590702.

Arnett AB, Wang T, **Eichler EE**, Bernier RA. (2021). Reflections on the genetics-first approach to advancements in molecular genetic and neurobiological research on neurodevelopmental disorders. *J Neurodev Disord* Jun 21;13(1):24. PMCID: PMC8215789.

Turner TN and **Eichler EE**. (2022). Chapter on “Genetics and Genomics.” In: Hollander E, Hagerman R, Ferretti C, editors. Textbook of Autism Spectrum Disorders, Second Edition. American Psychiatric Association Publishing. p 155–174.

Jacquemont S, Huguet G, Klein M, Chawner SJRA, Donald KA, van den Bree MBM, Sebat J, Ledbetter DH, Constantino JN, Earl RK, McDonald-McGinn DM, van Amelsvoort T, Swillen A, O'Donnell-Luria AH, Glahn DC, Almasy L, **Eichler EE**, Scherer SW, Robinson E, Bassett AS, Martin CL, Finucane B, Vorstman JAS, Bearden CE, Gur RE; Genes to Mental Health Network. (2022). Genes To Mental Health (G2MH): A framework to map the combined effects of rare and common variants on dimensions of cognition and psychopathology. *Am J Psychiatry* Mar;179(3):189–203. PMCID: PMC9345000.

Wang T, Antonacci-Fulton L, Howe K, Lawson HA, Lucas JK, Phillippy AM, Popejoy AB, Asri M, Carson C, Chaisson MJP, Chang X, Cook-Deegan R, Felsenfeld AL, Fulton RS, Garrison EP, Garrison NA, Graves-Lindsay TA, Ji H, Kenny EE, Koenig BA, Li D, Marschall T, McMichael JF, Novak AM, Purushotham D, Schneider VA, Schultz BI, Smith MW, Sofia HJ, Weissman T, Flicek P, Li H, Miga KH, Paten B, Jarvis ED, Hall IM, **Eichler EE**, Haussler D; Human Pangenome Reference Consortium. (2022). The Human Pangenome Project: a global resource to map genomic diversity. *Nature* Apr;604(7906):437–446. PMCID: PMC9402379.

*Wang T, Zhao PA, **Eichler EE**. (2022). Rare variants and the oligogenic architecture of autism. *Trends Genet* Sep;38(9):895–903. PMCID: PMC9378350.

*Logsdon GA, **Eichler EE**. (2022). The dynamic structure and rapid evolution of human centromeric satellite DNA. *Genes (Basel)* Dec 28;14(1):92. PMCID: PMC9859433.

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).