CURRICULUM VITAE

Evan Eugene Eichler
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EDUCATION

1995	Ph.D.	Department of Human Molecular Genetics, Baylor College of Medicine, Houston, TX Thesis (David L. Nelson, Supervisor): *AGG Interspersions within the FMR1 CGG Repeat: Models and Mechanisms of Triplet Repeat Instability*
1991	–	Research Scholar, Deutscher Akademischer Austauschdienst Ludwig-Maximilians Universität, Munich, Germany
1990	B.S.	Department of Biology, University of Saskatchewan, Saskatoon, Canada Honours Program in Biology

PROFESSIONAL EXPERIENCE

2008–Present	Professor (with tenure) Department of Genome Sciences, University of Washington (UW), Seattle, WA
2005–Present	Howard Hughes Medical Institute Investigator (HHMI)
2020–Present	Member, The Brotman Baty Institute (BBI)
2015–Present	Associate Member, New York Genome Center (NYGC), New York City, NY
2021–2023	Director (interim), Northwest Genomics Center (NWGC) University of Washington (UW) School of Medicine, Seattle, WA
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 for Top 10 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	Variant Bio, 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

2023–Present	International SAB, Telethon Institute for Genetics and Medcine (TIGEM), Naples Italy
2023	Reviewer, Knut and Alice Wallenberg Foundation.
2021–Present	International SAB (Fachbeirat), Max Planck Institute for Psycholinguistics, Nijmegen, Netherlands
2021–2023	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/ National Human Genome Research Institute (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	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

2024–Present	Member, Genome Sciences Seminar Committee

2021–2023	Director, Northwest Genomics Center (NWGC)
2022–2023	Member, NWGC Director 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– Present	Director, UW Long Reads Sequencing Center
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)
2012–Present	The Washington State Academy of Sciences (WSAS)
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
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

Genes and Development
Nature Methods
Neuron
Trends in Genetics
American Journal of 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
European Journal of Human Genetics
Genomics
Mammalian Genome
Neurogenetics
Human Genetics
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 every other year)
Department of Genome Sciences, University of Washington (UW)

2006–Current GENOME 465/565 "Advanced Human Genetics"
Lecturer: Genome Structure, Disease, Diversity and Evolution
10-week course co-taught with Mary-Claire King (13 contact hours/5 weeks every year)
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

GENOME 371 "Introductory Genetics"
2008 Lecturer (25 contact hours/10 weeks)
2007 Faculty Shadow (50 contact hrs/10 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 [5 tenured/tenure-track faculty; 4 clinicians; 3 postdocs or research associates; 2 hold positions in industry])

2022–Present Elizabeth (Lizzie) Plender, UW, predoctoral candidate jointly with Dr. Jessie Bloom, advanced to candidacy, June 2023.

2021–Present Taylor Real, UW, predoctoral candidate jointly with Dr. Andrew Stergachis, advanced to candidacy June 2022.

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: Attending Physician, Division of Genetics and Genomics, 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: Professor (tenured), 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: President, BioMap US, 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. Past: Technical Director, Center for Spatial and Functional Genomics, Children's Hospital of Philadelphia, PA; Current: Sr. Research Scientist, GENEWIZ, By Azenta Life Sciences

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. Last Known: 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 (55 postdocs: 8 current; 26 tenured/tenure-track faculty; 15 hold positions in industry; remainder are clinicians, research associates, faculty instructors or scientific writers)

2023–Present Sarah Brotman, Ph.D., postdoctoral research: Structural variation contribution to genome-wide association

2023–Present Mihir Trivedi, Ph.D., postdoctoral research: Characterization of ape chromosomal rearrangements

2023–Present Luyao Ren, Ph.D., postdoctoral research: Landscape of protein coding variants in segmental duplication regions and their association with human diseases

2023–Present Lingbin Ni, Ph.D., postdoctoral research: Functional characterization of structural variations by Hi-C and long-read sequencing

2023–Present Jiadong Lin, Ph.D., postdoctoral research: Structural variants in complex and divergent regions

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. Current: Staff Scientist, Allen Institute for Cell Science

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. Past: Acting Instructor, UW; Current: Consulting Bioinformatics Scientist, 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.

2018–2023 Glennis Logsdon, Ph.D., postdoctoral research: Sequence, assembly, and variation of centromeric regions of the human genome. Current: Assistant Professor, Department of Genetics, University of Pennsylvania Perelman School of Medicine, Philadelphia, PA

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: Senior Bioinformatics Scientist, Guardant Health, Seattle, WA

2017–2020 Arvis Sulovari, Ph.D., postdoctoral research: Integrated discovery of dosage sensitivity genes in neurodevelopmental disorders. Current: Principal 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: Head of Nutrition Research, 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. Past: Staff Software Engineer, DataBricks, Seattle, WA; Current: Staff Software Engineer & Tech Lead Manager, BigQuery at Google

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: Senior 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: Associate 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: Associate 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: Associate 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; Assistant Technical Director of Clinical Development at GeneDx, St. Louis, MO; Current: Lead Genomic Data Scientist, Geisinger

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; Principal Bioinformatician, Wellcome Sanger Institute, Hinxton, Cambridge, UK; Current: Senior Principal Scientist, Artios, 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. Past: Director, Data Science, Novartis Institutes for BioMedical Research (NIBR), Boston, MA; Current: Director, Cancer Data Science, Broad Institute of MIT and Harvard

2008–2012 Santhosh Girirajan, Ph.D., postdoctoral research: Mechanisms and implications of large-scale genome rearrangements. Current: Professor (tenured), 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: 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: Professor, University of Kentucky, Lexington, KY

2007–2010 Tomas Marques-Bonet, Ph.D., Marie Curie Fellow: Evolution of human/great-ape segmental duplications. Current: ICREA Researcher Professor, 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 Incendia 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., Morehouse College
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)

2023–Present	Sophia B. Gibson	Genome Sciences, UW	Advisor: Danny Miller
2020–2023	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: 566 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
- Invited Speaker, Clemson University Distinguished Lectures in Human Genetics Series, Clemson, SC, March
- Invited Speaker, 17th Troina Meeting on Genetics of Neurodevelopmental Disorders, Troina, Italy, April
- Invited Speaker, MSTP Dinner Presentation, Seattle, WA, April
- Invited Speaker, Clinical Genomics & NGS - 35th Course Jointly Organized by ESHG & CEUB, Bertinoro, Italy, May
- Invited Speaker, CSHL: Biology of Genomes Meeting, Cold Spring Harbor, NY, May
- Invited Speaker, Brazilian Cytogenetics and Cytogenomics Meeting, Curitiba, Brazil, May
- Invited Faculty, Workshop on Genomics Application, Český Krumlov, Czech Republic, May
- Invited Seminars, Bloch Lecture Series, University of Louisville Department of Biochemistry and Molecular Genetics, Louisville, KY, June
- Invited Speaker, 40th Blankenese Conference 'Evolutionary Medicine', Hamburg, Germany, June
- Invited Presenter, XXIII International Congress of Genetics (ICG): Genetics & Genomics – Linking Life and Society, Melbourne, Australia, July
- Invited Presenter, Han Brunner Farewell Symposium, Nijmegen, Netherlands, September
- Invited Speaker, First International Forum on Autism Genetics, Bocchigliero, Italy, September

- Invited Speaker and Moderator, Human Pangenome Reference Consortium and Cross Consortium Meeting, St. Louis, MO, September
- Invited Lecturer, Duke Distinguished Lecture Series, Durham, NC, September
- Invited Presenter, Genome Institute of Singapore Speaker Series, Singapore, October
- Invited Workshop Presenter, 14th International Workshop on Advanced Genomics, Tokyo, Japan, October
- Invited Speaker, UW Institute for Public Health Genetics, Seattle, WA, October
- Invited Class Lecturer, Seattle Pacific University, Seattle, WA, October
- Invited Keynote, University of Florida Genetics Institute (UFGI) Symposium, Gainesville, FL, November
- Invited Speaker, Korea Academy of Science and Technology (KAST) 10th InterAcademy Workshop "Somatic Mosaicism in Human Development, Aging, and Diseases," Frankfurt, Germany, November
- Invited Lecturer, Stephen T. Warren Distinguished Lecture, Department of Human Genetics, Emory University School of Medicine, Atlanta, Georgia, December

2024

- Invited Faculty Talk, Human Biology Division, Fred Hutchinson Cancer Research Center, Seattle, WA, January
- Invited Faculty, Workshop on Genomics Application, Český Krumlov, Czech Republic, January
- Invited International Speaker, Indian Society of Human Genetics (ISHG) 2024, Ahmedabad, India, January
- Invited Speaker, Genomics of Rare Disease 2024, Wellcome Sanger Institute, Hinxton, UK, March
- Invited Seminar, Department of Biomedical Engineering (BME), Vanderbilt University, Nashville, TN, April
- Panel Member, Annual NHGRI/GTG Meeting panelist with Mary-Claire King, Bob Waterston, Gail Jarvik, & Mike Bamshad, Seattle, WA, April
- Invited Keynote, 9th Meeting of the Study Group on Genetics of Diabetes (SGGD), Exeter, UK, April
- Invited Speaker, Clinical Genomics & NGS - 36th Course Jointly Organized by ESHG & CEUB, Bertinoro, Italy, May
- Invited Speaker, The Jackson Laboratory: Long-Read Sequencing Workshop, Farmington, CT, May
- Invited Seminar, Portland State University Biology Department Lester Newman Seminar Series, Portland, OR, May
- Invited Concurrent Symposia Speaker, European Human Genetics Conference/ESHG, Berlin, Germany, June
- Invited Speaker, SMaHT (Somatic Mosaicism across Human Tissues) 2024 Annual Meeting, St. Louis, MO, June
- Invited Lecturer, Paris Brain Institute ICM: International Lecture Series, Paris, France, June

RESEARCH GRANTS

a) Active

National Institutes of Health/ National Institutes of Mental Health (R01 MH101221-12)
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 HG002385-22)
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
2018–2027

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, Tom Nowakowski, Huda Zoghbi
2021–2025

Weill Neurohub Family Foundation

Title: Mapping human-specific constraint and characterizing cell type specificity in recently duplicated genes during neurodevelopment
Goal: Characterize functional properties of human-specific genes associated with neurodevelopment
Multi-PI: Alex Pollen (UCSF), Peter Sudmant (UC Berkeley), Evan Eichler (UW)
2023–2025

National Institutes of Health (1U01HG010971-05)
Title: Center for Human Reference Genome Diversity
Goal: Construction of a pangenome graph based on phased assemblies from diverse human genomes.
Multi-PI: Karen Miga; Evan E. Eichler; Ira Hall; Erich Jarvis
2019–2029

National Institutes of Health (1U41HG007497-09)
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, Tobias Marschall
2013–2028

National Institutes of Health (3OT2OD002748-01S5)
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: Chia-Lin Wei, Gail Jarvik; Evan E. Eichler
2018–2024

National Institutes of Health (1U01HG011744-03)
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: Chia-Lin Wei, Evan E. Eichler; Michael Bamshad
2021–2026

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 (PI change request to Chia-Lin Wei pending)
2016–2024

National Institutes of Health (NHLBI) (5R01NS122766-02)
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

National Institutes of Health (1UM1DA058220-01)
Title: Mosaicism in Human Tissues, from Telomere to Telomere
Goal: Generate a map of somatic mosaicism in humans using deep short-read and long-read sequencing data coupled with full-length transcript RNA sequencing.
Multi-PI: James Bennett, Evan E. Eichler, Andrew Stergachis
2023–2028

b) Previous
National Institutes of Health (1U01MH119705-05)
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 (1U01HG10973-04)
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

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: ¼-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

(556 peer-reviewed publications: 237 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.

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