

BIOGRAPHICAL SKETCH

NAME Evan E. Eichler, Ph.D.	POSITION TITLE Associate Professor		
eRA COMMONS USER NAME EEICHLER			
EDUCATION/TRAINING (Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)			
INSTITUTION AND LOCATION	DEGREE	YEAR(s)	FIELD OF STUDY
University of Saskatchewan, Saskatoon, Canada	B.Sc.	1990	Biology
Ludwig-Maximilians Universitaet, Munich, Germany		1990-1991	Molecular Biology
Baylor College of Medicine, Houston, TX	Ph.D.	1995	Molecular Genetics

Positions and Honors**Professional Experience**

1995-1997	Postdoctoral Fellow , Biology and Biotechnology Research Program, Lawrence Livermore National Laboratory, Livermore, CA
1997	Research Affiliate , Department of Human Genetics, Roswell Park Cancer Institute, Buffalo, NY
1997-2003	Assistant Professor , Department of Genetics, Case Western Reserve University, Cleveland, OH
2002-2004	Director , Bioinformatics Core Facility, Department of Genetics, Case Western Reserve University, Cleveland, OH
2003-2004	Associate Professor , Department of Genetics, Case Western Reserve University, Cleveland, OH
2004-Present	Associate Professor , Department of Genome Sciences, University of Washington, Seattle, WA
2005-Present	Investigator , Howard Hughes Medical Institute

Honors, Awards and Service

1987-1988	NSERC (Canada) Research Award
1990-1991	<i>Deutscher Akademischer Austauschdienst</i> Research Scientist Award
1993-1995	National Research Service Award/Human Genome Research
1994	Predoctoral Basic American Society of Human Genetics Award
1995-1997	Alexander Hollaender Distinguished Human Genome Postdoctoral Fellowship
1998-2001	March of Dimes Basil O'Connor Scholar
1999-Present	Editorial Board of <i>Genome Research</i> . 2002 Appointed Editor.
2001-2003	Faculty of 1000
2002-2004	Editorial Board of <i>American Journal of Human Genetics</i>
2002-Present	Editorial Board of <i>BMC Genomics</i>
2002-Present	Editorial Board of <i>DNA Sequence</i>
2005-Present	HHMI Investigator
2006	AAAS Fellow

B. Selected Publications (35 of 123)

- Eichler EE**, Richards S, Gibbs RA, Nelson DL. 1993. Fine structure of the human FMR1 gene. *Hum Molec Genet* 2:1147-1153.
- Eichler EE**, Holden JJA, Popovich BW, Reiss AL, Richards CS, Snow K, Thibodeau SN, Ward PA, Nelson DL. 1994. Length of uninterrupted CGG repeats determines instability in the FMR1 gene. *Nature Genetics* 8:88-94.
- Eichler EE**, Kunst CB, Lugenbeel KA, Ryder OA, Warren ST, Nelson DL. 1995. Evolution of the FMR1 CGG Repeat. *Nature Genetics* 11:302-308.
- Eichler EE**, Lu F, Shen Y, Antonucci R, Doggett NA, Moyzis RK, Baldini A, Gibbs RA, Nelson DL. 1996. Duplication of the Xq28 CDM-CTR region to 16p11.1: A novel pericentromeric-directed mechanism for paralogous genome evolution. *Hum Molec Genet* 5:899-912.

- Eichler EE**, Budarf ML, Rocchi M, Deaven LD, Doggett NK, Nelson DL, Mohrenweiser H. 1997. Interchromosomal duplications of the adrenoleukodystrophy locus: A phenomenon of pericentromeric plasticity. *Hum Molec Genet* 6:991-1002.
- Eichler EE**. 1998. Masquerading repeats: Paralogous pitfalls of the human genome. *Genome Res* 8:758-762.
- Eichler EE**, Archidiacono N, Rocchi M. 1999. CAGGG Repeats and the pericentromeric duplication of the hominoid genome. *Genome Res* 9:1048-1058.
- Horvath JE, Viggiano L, Loftus BJ, Adams MD, Archidiacono N, Rocchi M, **Eichler EE**. 2000. Molecular structure and evolution of an alpha/non-alpha satellite junction. *Hum Molec Genet* 9:113-123.
- Horvath JE, Schwartz S, **Eichler EE**. 2000. The mosaic structure of human pericentromeric DNA: A strategy for characterizing complex regions of the human genome. *Genome Res* 10:839-852.
- Bailey JA, Carrel L, Chakravarti A, **Eichler EE**. 2000. Molecular evidence for Lyon's Second Hypothesis: The relationship between LINE-1 elements and X inactivation. *Proc Natl Acad Sci* 97:6634-6639.
- International Sequencing Consortium. 2001. Initial sequencing and analysis of the human genome. *Nature* 409:860-920. *Contributors Bailey JA, **Eichler EE** to Segmental Duplication section of the manuscript, pages 889-892.
- Eichler EE**. 2001. Segmental duplications: What's missing, misassigned, and misassembled – and should we care? *Genome Res* 11:653-656.
- Bailey JA, Yavor AM, Massa HF, Trask BJ, **Eichler EE**. 2001. Segmental duplications: Organization and impact within the current human genome project assembly. *Genome Res*.11:1005-17.
- Eichler, EE**. 2001. Recent duplication, domain accretion and the dynamic mutation of the human genome. *Trends Genet* 17:661-669.
- Johnson ME, Viggiano L, Bailey JA, Abdul-Rauf M, Goodwin G, Rocchi M, **Eichler EE**. 2001. Positive selection of a novel gene family during the emergence of humans and great apes. *Nature* 413:514-519.
- Bailey JA, Yavor AM, Viggiano L, Misceo D, Horvath JE, Archidiacono N, Schwartz S, Rocchi M, **Eichler EE**. 2002. Human specific duplication and mosaic transcripts: The paralogous structure of human chromosome 22. *Am J Hum Genet* 70:83-100.
- Bailey JA, Gu Z, Clark RA, Reinert K, Samonte RV, Schwartz SS, Adams MD, Myers EW, Li PW, **Eichler EE**. 2002. Recent segmental duplications in the human genome. *Science* 293:1003-1007.
- Horvath JE, Gulden CL, Bailey JA, Yohn C, McPherson JD, Prescott A, Roe BA, De Jong PJ, Ventura M, Misceo D, Archidiacono N, Zhao S, Schwartz S, Rocchi M, **Eichler EE**. 2003. Using a pericentromeric interspersed repeat to recapitulate the phylogeny and expansion of human centromeric segmental duplications. *Mol Biol Evol* 20(9):1463-1479.
- Locke DP, Segraves R, Carbone L, Archidiacono N, Albertson DG, Pinkel D, **Eichler EE**. 2003. Large-scale variation among human and great ape genomes determined by array comparative genomic hybridization. *Genome Res* 13:347-357.
- Bailey JA, Liu G, **Eichler EE**. 2003. An Alu transposition model for the origin and expansion of segmental duplications. *Am J Hum Genet* 73(4):823-834.
- She X, Horvath JE, Jiang Z, Liu G, Furey TS, Christ L, Clark R, Graves T, Alkan C, Bailey JA, Sahinalp C, Rocchi M, Haussler D, Wilson RK, Miller W, Schwartz S, **Eichler EE**. 2004. The structure and evolution of centromeric transition regions within the human genome. *Nature* 430:857-864.
- She X, Jiang Z, Clark RA, Liu G, Cheng Z, Tuzun E, Church DM, Sutton G, Halpern AL, **Eichler EE**. 2004. Shotgun sequence assembly and recent segmental duplications within the human genome. *Nature* 431:927-930.
- Yohn CT, Jiang Z, McGrath SD, Hayden KE, Khaitovich P, Johnson ME, Eichler MY, McPherson JD, Zhao S, Paabo S, **Eichler EE**. 2005. Lineage-specific expansions of retroviral insertions within the genomes of African great apes but not humans and orangutans. *PLoS Biol* 3(4):e110.
- Horvath JE, Gulden CL, Samonte RU, Eichler MY, Ventura M, McPherson JD, Graves T, Wilson RK, Scwartz S, Rocchi M, **Eichler EE**. 2005. Punctuated duplication events during the evolution of human chromosome 2p11. *Genome Res* 15:914-927.
- Tuzun E, Sharp AJ, Bailey JA, Kaul R, Morrison VA, Pertz LM, Haugen E, Hayden H, Albertson D, Pinkel D, Olson MV, **Eichler EE**. 2005. Fine-scale structural variation of the human genome. *Nat Genet* 37:727-732.
- Cheng Z, Ventura M, She X, Khaitovich P, Graves T, Osoegawa K, Church D, DeJong P, Wilson RK, Paabo S, Rocchi M, **Eichler EE**. 2005. A genome-wide comparison of recent chimpanzee and human segmental duplications. *Nature* 437:88-93.
- Newman TL, Tuzun E, Morrison VA, Hayden KE, Ventura M, McGrath SD, Rocchi M, **Eichler EE**. 2005. A genome-wide survey of structural variation between human and chimpanzee. *Genome Res* 15:1344-1356.

- Eichler EE.** 2006. Widening the spectrum of human genetic variation. *Nat Genet* 38(1):9-11.
- She X, Liu G, Ventura M, Zhao S, NISC Comparative Sequencing Program, Misceo D, Roberto R, Cardone MF, Rocchi M, Green ED, Archidiacono N, **Eichler EE.** 2006. A preliminary comparative analysis of primate segmental duplications shows elevated substitution rates and a great-ape expansion of intrachromosomal duplications. *Genome Res* 16:576-583.
- Bailey JA, **Eichler EE.** 2006. Primate segmental duplications: Crucibles of evolution, diversity and disease. *Nat Rev Genet* 7(7):552-564.
- Locke DP, Sharp AJ, McCarroll SA, McGrath SD, Newman TL, Cheng Z, Schwartz S, Albertson DG, Pinkel D, Altshuler DM, **Eichler EE.** 2006. Linkage disequilibrium and heritability of copy-number polymorphisms within duplicated regions of the human genome. *Am J Hum Genet* 79:275-290.
- Sharp AJ, Hansen S, Selzer RR, Cheng Z, Regan R, Hurst JA, Stewart H, Price SM, Blair E, Hennekam RC, Fitzpatrick CA, Segraves R, Richmond TA, Guiver C, Albertson DG, Pinkel D, Eis PS, Schwartz S, Knight SJ, **Eichler EE.** 2006. Discovery of previously unidentified genomic disorders from the duplication architecture of the human genome. *Nat Genet* 38:1038-1042.
- Johnson ME, NISC Comparative Sequencing Program, Cheng Z, Morrison AV, Scherer S, Ventura M, Gibbs RA, Green ED, **Eichler EE.** 2006. Recurrent duplication-driven transposition of DNA during hominoid evolution. *Proc Natl Acad Sci USA* 103:17626-17631.
- Eichler EE,** Nickerson DA, Altshuler D, Bowcock AM, Brooks LD, Carter NP, Church DM, Felsenfeld A, Guyer M, Lee C, Lupski JR, Mullikin JC, Pritchard JK, Sebat J, Sherry ST, Smith D, Valle D, Waterston RH. 2007. Completing the map of human genetic variation. *Nature* 447(7141):161-165 (10 May 2007).
- Jiang Z, Tang H, Ventura M, Cardone MF, She X, Pevzner P, **Eichler EE.** 2007. Ancestral reconstruction of segmental duplications reveals punctuated cores of human genome evolution. *Nat Genet* 39(11):1361-1368 (7 Oct 2007).

C. Research Support

Investigator Award

Period: 09/16/05-08/31/10

Source: Howard Hughes Medical Institute

No specific projects are associated with this funding. However, Dr. Eichler receives 100% of his salary and fringe benefit (FB) compensation from the Howard Hughes Medical Institute. HHMI provides support for beginning postdoctoral fellows and research assistants as well as equipment and supplies. HHMI support is being used to develop transgenic mouse models for positively selected duplicate genes, to analyze fusion transcripts as biomarkers of cancer, and to assess the association of structural variation and complex genetic disease (especially immune and autoimmunity related disease). There is no scientific overlap between the aims of this proposal and the research conducted under the auspice of the HHMI.

Project Number: (1R01 HD043569; PI)

Period: 03/31/03-12/31/08

Source: NIH

Title of Project: Segmental Aneusomy of Duplicated DNA

The aim of this grant is to assess the nature and frequency of duplication-mediated rearrangements using BAC-based array comparative genomic hybridization AND to determine if an excess of de novo events have occurred among children with mental retardation and congenital birth defects.

Project Number: (1R01 HG002385; PI)

Period: 09/21/07-7/31/10

Source: NIH

Title of Project: Sequence and Assembly of Pericentromeric Duplications (3rd cycle of funding)

The following proposal is designed to provide a systematic approach for closing DNA sequence gaps within pericentromeric regions of human chromosomes. The target of this grant will be to direct sequence closure within ten (~20 Mb) heterochromatic/euchromatic transition zones (1q12, 2p11, 2q21, 9p11, 9q12, 14q11, 15q11, 16p11, 21q11 and 22q11).

Project Number: (1R01 GM058815; PI)

Period: 04/11/07-03/31/11

Source: NIH

Title of Project: Mechanism and Instability of Segmental Duplications (3rd cycle of funding)

The aim of this grant is to investigate the evolutionary mechanism and instability of low-copy repeats on chromosome 16 among human and non-human primate species.

Project Number: (1P01 HG004120; PI) Period: 06/01/07-05/30/10

Source: NIH

Title of Project: Human Genome Structural Variation

The aim of this grant is to identify, sequence, and genotype fine-scale structural variation based on the sequencing of additional human genomes.

Project Number: (1U54 HG004592-01; Co-PI) Period: 09/29/07-6/30/11

Source: NIH

Title of Project: A Comprehensive Catalog of Human DNaseI Hypersensitive Sites

The overall aim of this proposal is to establish a comprehensive, high quality catalog of human DNaseI hypersensitive sites spanning all major tissue lineages.

Overlap: There is no scientific overlap between currently funded grants and this proposal.

COMPLETED RESEARCH

***Project Number: (U54 HG02043; Co-PI)** Period: 03/31/03-2/28/06

Source: NIH

PI: Maynard Olson (University of Washington)

Title of Project: UW Genome Center Large-Scale Sequencing Program.

The aim of this grant was to develop production sequencing capacity to target problematic euchromatic sequence regions of the human genome.

***Project Number: (1R01 ES10631; Co-PI)** Period: 4/1/01-7/31/06

Source: NIH

PI: Robert Nicholls (University of Pennsylvania)

Title of Project: Genetic and Environmental Factors in Deletion Disorders

The aim of this grant was to examine the molecular mechanisms underlying rearrangements associated with Prader-Willi and Angelman Syndromes and the role of genetic and environmental susceptibility in predisposing to this instability.

Project Number: (ER62862; PI) Period: 09/01/99-7/31/03

Source: DOE

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

The objective of this proposal was to develop and implement a novel sequence-anchor strategy to generate a contiguous BAC and cosmid map of the most proximal portion of chromosome 19p12 for the purpose of complete sequence characterization; and to assay heteromorphic variation of this pericentromeric region in the human. A supplement was issued for an additional year to assist with the analysis of segmental duplications on chromosome 15 and 16.

Project Number (FY99-0120) Period: 02/1/99-01/31/01

Source: March of Dimes

Title of Project: Chromosome Duplication and Instability in 15q11-q13

This two-year grant focused on characterization of the pericentromeric region of 15q11-q13 and the direct evaluation of duplications in this region as sites of recurrent chromosomal structural rearrangement associated with Prader-Willi, Angelman Syndrome, and 15q inversions.

Project Number: (1R01 HG01847-02; Co-PI) Period: 7/1/98-6/30/01

Source: NIH

Title of Project: Human Genomic Sequence Variation: X chromosome

Principal Investigator/Program Director (Last, First, Middle): Eichler, Evan E.

The aim of this proposal was to examine the nature and frequency of sequence variation in the human genome by studying multiple regions of the X chromosome (1 MB) in human (80 individuals) and some primate (16) samples. Dr. Eichler was Co-PI of this grant and received only salary support from this proposal.

Project Number: (1R01 MH00607-02; Co-PI)

Period: 10/1/98-9/30/01

Source: NIH

Title of Project: Human Genomic Polymorphism

This grant focused on the identification and characterization of SNPs (single-nucleotide polymorphisms) using large-scale DNA microarray technology (Affymetrix). The study targeted the analysis of sequence variation on 40 genomic segments each of 100 kilobases or greater which are known to contain candidate genes for various neuropsychiatric disorders. Dr. Eichler was Co-PI of this grant and received only salary support from this proposal.