Erika M. Kvikstad

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SUMMARY

- Over 14 years of experience in the field of computational genomics, spanning expertise in
  - genomics
  - statistical analysis of sequence data
  - software development for structural variation detection, annotation, and analysis
- 8 years of postdoctoral experience generating and analysing whole genome sequencing (WGS), whole exome sequencing (WES), and targeted sequencing data
- 2 years of translational bioinformatics experience bringing bams to benchside, to uncover disease pathogenesis across a broad range of conditions (epilepsy, immunological) and cancers
- Proficiency in computing platforms for rapid, rigorous and reproducible data-driven science
  *nix, R, HPC, git/github

EDUCATION

2003-2009 **PhD, Genetics**
The Pennsylvania State University
University Park, PA, USA

1994-1999 **B.S. Honors, Biochemistry and French**
University of Wisconsin-Madison
Madison, WI, USA

RESEARCH EXPERIENCE

2016-present **Bioinformatician,** Oxford Biomedical Research Centre
University of Oxford
Oxford, UK
Group Leader: Jenny Taylor

- Applying bioinformatics pipelines for processing of WGS data from raw data to human reference alignment and variant calling using industry tools such as BWA, SAM tools, Picard tools
- Annotating, analysing and interpreting candidate causative variants in over 100 clinical genomes from patient families (48), singletons (10), cancer-normal pairs (87)
- Collaborating with biologists, informaticians, and statisticians to deliver in-depth presentations to collaborators and stakeholders on > 20 patient cases of rare disease/cancer
- Developing novel pipelines and software for detection of viral integrations in WGS derived from FFPE, FF, and circulating cell-free (ccf) cancer DNA samples

2015 **Visiting Scientist**, Medical and Population Genetics
The Broad Institute, Inc.
Boston, Massachusetts, USA
Host: Daniel MacArthur

- Lead externally funded fellowship to generate TE-NGS from 300 clinical samples in the Exome Aggregation Consortium (ExAC)

2013-2015 **Research Associate**, The Wellcome Trust Centre for Human Genetics

University of Oxford
Oxford, UK
Group Leader: Gerton Lunter

- Designed genomics tools (experimental library preparation and computational detection algorithm) for targeted sequencing of transposable elements (TE) from NGS data: TE-NGS

2009-2012 **Postdoctoral Researcher**, Department of Genetics and Evolutionary Genomics
Université Lyon 1
Lyon, France
Advisor: Laurent Duret

- Produced first genome-wide statistical analysis of insertion/deletion (indel) site frequency spectrum from population genomic data of 179 individuals as part of 1000 Genomes Pilot
- Highlighted complex indel mutagenesis patterns and mathematically modeled frequency of cryptic indel hotspots

2003-2009 **Research Assistant**, Department of Biology
The Pennsylvania State University
University Park, PA, USA
Advisor: Kateryna Makova

- Performed multivariate modeling of publicly available TE annotations in conjunction with genome landscape features to determine sequence characteristics of TE insertions
- Conducted first genome-wide application of wavelet statistics for scale-free analysis of indel mutation patterns
- Implemented first informatics tools for genome-wide detection and annotation of indels from multiple species alignments, distributed on Galaxy bioinformatics platform

2000-2003 **Research Specialist**, Department of Genetics/Biotechnology Center
University of Wisconsin-Madison
Madison, WI, USA
Group Leader: David C. Schwartz

- Generated ordered restriction fragment digests - optical maps – of microbial genomes

1999-2000 **Associate Research Specialist**, Department of Chemistry
University of Wisconsin-Madison
Madison, WI, USA
Group Leader: Lloyd M. Smith

- Produced finished continuous and contiguous sequence as part of Human Genome Project

GRANTS AND FELLOWSHIPS

2015 **European Molecular Biology Organization**, Short-Term Fellowship
Host: The Broad Institute, Inc
Boston, Massachusetts, USA

2010-2012 **European Molecular Biology Organization,** Long-Term Fellowship
Host: Université Lyon 1
Lyon, France

2005-2008 **The Pennsylvania State University**, Academic Computing Fellowship

The Pennsylvania State University
University Park, PA, USA

PUBLICATIONS

* joint first authors

1. **Kvikstad E.M.***, Knight S.J.*, Pentony M.M., Burns A., Klintman J., Taylor J.C., Schuh A. (in preparation) Viral detection and impact of EBV in a cohort of patients with chronic lymphocytic leukaemia and Richters Syndrome.
2. Anna Schuh, Helene Dreau, Samantha JL Knight, Rosanna T Mizani, Richard Colling, Dimitris Vavoulis, Kate Ridout, Pavlos Antoniou**, Erika M. Kvikstad**, Melissa M. Pentony, Angela Hamblin, Andrew Protheroe, Marina Parton, Ketan A Shah, Orosz Zsolt, Nick Athanasou, Bass Hassan, Adrienne M Flanagan, Ahmed Ahmed, Stuart Winter, Adrian Harris, I Tomlinson, Niko Popitsch, David Church, Jenny C Taylor. (2018) Whole Genome Sequencing reveals clinically actionable mutation profiles in patients with cancer. *Molecular Case Studies* (in review).
3. **Kvikstad E.M**., Piazza P., Taylor J.C., Lunter, G. (2018) A high throughput screen for active human transposable elements. *BMC Genomics* 19:115.
4. **Kvikstad E.M.,** Duret L. (2014) Strong heterogeneity in mutation rate causes misleading hallmarks of natural selection on indel mutations. *Molecular Biology and Evolution* 31(1):23-36.
5. **Kvikstad E.M**., Makova K.D. (2013) Rapid evolution of genes on the human X-chromosome version 2.0. In: *Encyclopedia Of Life Sciences*. John Wiley & Sons, Ltd: Chichester.
6. Montgomery S.B.*, Goode D*, **Kvikstad E***, Albers K, Zhang Z, Mu XJ, Ananda G, Howie B, The 1000 Genomes Pilot Project Consortium, Sidow A, Duret L, Gerstein M, Makova K, Marchini J, McVean G, Lunter G. (2013) The origin, evolution, and functional impact of short insertion-deletion variants identified in 179 human genomes. *Genome Research* 23(5): 749-61.
7. **Kvikstad E.M.**, Makova K.D. (2010) The (r)evolution of SINE vs. LINE distributions in primate genomes: Sex chromosomes are important. *Genome Research* 20: 600-613. **Featured cover.**
8. **Kvikstad E.M**., Chiaromonte F., Makova K.D. (2009) Ride the wavelet: Scale specific dynamics of genomic contexts flanking small insertions and deletions. *Genome Research* 19: 1153-1164.
9. **Kvikstad E.M**., Makova K.D. (2008) Rapid evolution of genes on the human X-chromosome. In: *Encyclopedia Of Life Sciences*. John Wiley & Sons, Ltd: Chichester.
10. **Kvikstad E.M**., Tyekucheva S., Chiaromonte F., Makova K.D. (2007) A macaque's-eye view of human insertions and deletions: Differences in mechanisms. *PLoS Comp Bio* 3(9): e176.
11. Zhou S, Kile A, Bechner M, Place M, **Kvikstad E**, Deng W, Wei J, Severin J, Runnheim R, Churas C, Forrest D, Dimalanta ET, Lamers C, Burland V, Blattner FR, Schwartz DC. (2004) Single-molecule approach to bacterial genomic comparisons via optical mapping. *J Bacteriol.* **186**(22): 7773-82.
12. Zhou S, Kile A, **Kvikstad E**, Bechner M, Severin J, Forrest D, Runnheim R, Churas C, Anantharaman TS, Myler P, Vogt C, Ivens A, Stuart K, Schwartz DC. (2004) Shotgun optical mapping of the entire *Leishmania major* Friedlin genome. *Mol Biochem Parasitol.* **138**(1):97-106.
13. Zhou S, **Kvikstad E**, Kile A, Severin J, Forrest D, Runnheim R, Churas C, Hickman JW, Mackenzie C, Choudhary M, Donohue T, Kaplan S, Schwartz DC. (2003) Whole-genome shotgun optical mapping of *Rhodobacter sphaeroides* strain 2.4.1 and its use for whole genome shotgun sequence assembly. *Genome Research* **13**(9): 2142-51.
14. Zhou S, Deng W, Anantharaman TS, Lim A, Dimalanta ET, Wang J, Wu T, Chunhong T, Creighton R, Kile A, **Kvikstad E**, Bechner M, Yen G, Garic-Stankovic A, Severin J, Forrest D, Runnheim R, Churas C, Lamers C, Perna NT, Burland V, Blattner FR, Mishra B, Schwartz DC.

(2002) A Whole Genome Shotgun Optical Map of *Yersinia Pestis* Strain KIM. *Applied and Environmental Microbiology* **68**: 6321-6331.

PRESENTATIONS

Invited Seminars

2018	Translational Bioinformatics Unit: Bristol-Myers Squibb, Hopewell, NJ, USA. January 2018.
2018	Leonard Davis School of Gerontology: USC, USA. January 2018.
2017	Molecular Diagnostics Centre: University of Oxford, UK. October 2017.
2017	School of Biological Sciences: University of Canterbury, Christchurch, NZ. March 2017.
2017	Nevada Institute Personalized Medicine. March 2017.
2017	School of Pharmacy and Biomedical Sciences: University of Portsmouth, UK. January 2017.
2016	Wolfson Wohl Research Centre: University of Glasgow, UK. December 2016.
2015	Analytic and Translational Genetics Unit: Massachusetts General Hospital, USA. September 2015.
2015	Center for Medical Genomics: The Pennsylvania State University-Research Park, PA, USA. August 2015.
2015	Department of Genetics: University of Cambridge, UK. July 2015.
2011	Center for Comparative Genomics and Bioinformatics: The Pennsylvania State University-Research Park, PA, USA. May 2011.
2010	Hubricht Institute: Utrecht, The Netherlands. April 2010.
2009	Laboratoire Biométrie et Biologie Évolutive: Université Claude Bernard-Lyon 1, France. January 2009.
2009	Centre Integratif de Genomique: University Lausanne-Lausanne, Switzerland. January 2009.

Contributed Talks at Conferences and Meetings

2014	Kvikstad E.M., Lunter G. A cost-effective screen for identifying novel transposable element insertions in human genomes. American Society for Human Genetics Annual Meeting: San Diego, CA, USA. October 2014.
2014	Kvikstad E.M., Duret L. Strong heterogeneity in mutation rate causes misleading hallmarks of natural selection on indel mutations. EMBO Long Term Fellows' Meeting 2014: EMBL, Heidelberg, Germany. June 2014.
2012	Kvikstad E.M., Duret L. Give a little, take a little: Indels and the evolution of genome architecture. Genome Informatics: Cambridge, UK. September 2012.
2011	Kvikstad E.M., Duret L. Indels and the evolution of genome architecture: the long and short of it. Groupement de Recherche Européen –AR "Comparative Genomics": Université Claude Bernard-Lyon 1, France. November 2011.
2010	Kvikstad, E.M., Duret L., 1000 Genomes Consortium Indel Analysis Subgroup. Heterogeneity in patterns of insertions and deletions segregating in 1000 Genomes. Integrative Post-Genomics: Université Claude Bernard-Lyon 1, France. November 2010.

2009 Kvikstad E.M., Makova K.D. The (r)evolution of SINE vs. LINE distributions in primate genomes: Do sex chromosomes matter? Groupement de Recherche Européen –AR "Comparative Genomics": Université Claude Bernard-Lyon 1, France. October 2009.

2007 Kvikstad E.M., Tyekucheva, S., Chiaromonte, F., Makova K.D. A macaque's-eye view of human insertions and deletions: Regional rate variation and mechanisms of mutagenesis. Society for Molecular Biology and Evolution Annual Meeting: Halifax, Nova Scotia, Canada. June 2007.

2006 Kvikstad E.M., Makova K.D. Contribution of insertion/deletion events to sex chromosome architecture as inferred from human-chimpanzee genomic alignments. Society for Molecular Biology and Evolution Annual Meeting: Tempe, AZ, USA. May 2006.

POSTERS

2015 Kvikstad E.M., Lunter G. Towards quantifying the disease impact of transposable element (TE) insertions in humans. EMBO/EMBL Symposium "The Mobile Genome: Genetic and physiological impacts of transposable elements": Heidelberg, Germany. September 2015.

2015 Kvikstad E.M., Lunter G. A cost-effective whole-genome screen for identifying novel transposable element (TE) insertions. Society for Molecular Biology and Evolution Annual Meeting 2015: Vienna, Austria. July 2015.

2012 Kvikstad E.M., Duret L. Indels and the evolution of genome architecture. Society for Molecular Biology and Evolution Annual Meeting 2012: Dublin, Ireland. June 2012.

2011 Kvikstad E.M., Duret L., 1000 Genomes Consortium Indel Analysis Subgroup. Born this way: Insertions and deletions are equally deleterious. The EMBO Meeting: Vienna, Austria. September 2011.

2011 Kvikstad E.M., Duret L., 1000 Genomes Consortium Indel Analysis Subgroup. Born this way: Insertions and deletions are equally deleterious. The Biology of Genomes: Cold Spring Harbor Laboratory, NY, USA. May 2011.

2008 Kvikstad E.M., Chiaromonte, F., Makova K.D. Ride the wavelet: Scale specific dynamics of genomic contexts flanking small insertions and deletions. Society for Molecular Biology and Evolution 2008: Barcelona, Spain. June 2008.

2008 Kvikstad E.M., Chiaromonte, F., Makova K.D. Ride the wavelet: Scale specific dynamics of genomic contexts flanking small insertions and deletions. The Biology of Genomes: Cold Spring Harbor Laboratory, NY, USA. May 2008.

2007 Kvikstad E.M., Tyekucheva, S., Chiaromonte, F., Makova K.D. A macaque's-eye view of human insertions and deletions: Regional rate variation and mechanisms of mutagenesis. The Biology of Genomes: Cold Spring Harbor Laboratory, NY, USA. May 2007.

2006 Kvikstad E.M., Makova K.D. Contribution of insertion/deletion events to sex chromosome architecture as inferred from human-chimpanzee genomic alignments. The Biology of Genomes: Cold Spring Harbor Laboratory, NY, USA. May 2006.

2006 Kvikstad E.M., Makova K.D. Contribution of insertion/deletion events to sex chromosome architecture as inferred from human-chimpanzee genomic alignments. The Pennsylvania State University Genetics Symposium 2006: The Pennsylvania State University-Research Park, PA, USA. April 2006.

2004	Goetting-Minesky P, **Kvikstad E**, Makova K. Male mutation bias in Perissodactyla and Cetacea. Society for Molecular Biology and Evolution Annual Meeting 2004: The Pennsylvania State University-Research Park, PA, USA. June 2004.

TEACHING AND MENTORING EXPERIENCE

2017	"Introduction to Genomics" School of Biological Sciences University of Canterbury Guest Lecturer
2014	Galaxy Demo The Wellcome Trust Centre for Human Genetics University of Oxford Instructor
2011-2012	Eugénie Pessia Université Lyon 1 PhD Thesis Committee Member
2010	Galaxy Demo PRABI Université Claude Bernard-Lyon 1 Instructor
2009	Evolution of Developmental Mechanisms The Pennsylvania State University Teaching Assistant
2008	Molecular Evolution The Pennsylvania State University Teaching Assistant
2006-2008	"Evolution of Sex Chromosomes", Molecular Evolution The Pennsylvania State University Guest Lecturer
2005	3rd Bioinformatics Workshop The Pennsylvania State University Teaching Assistant

HONORS AND AWARDS

2015	The Wellcome Trust Centre for Human Genetics: Carer's Fund Award
2015	Society for Molecular Biology and Evolution: Childcare Travel Award
2008	Jeanette Ritter Mohnkern Graduate Student Scholarship in Biology
2006-2007	Institute for Molecular Evolutionary Genetics: Travel Award
2006	Intercollege Graduate Degree Program in Genetics: Travel Award
2005-2006	Women in Science and Engineering (WISE): Lockheed Martin Travel Grant
2004-2005	The Pennsylvania State University: Graham Endowed Fellowship
2004-2005	The Pennsylvania State University: J. Ben and Helen D. Hill Memorial Award
2003-2004	The Pennsylvania State University: Braddock Research Award
2003-2004	The Pennsylvania State University: Graduate Fellowship, university-wide recognition of highly recruited students
1999	University of Wisconsin-Madison: Graduation with Honors

PROFESSIONAL SERVICE

Professional Membership

2014-present	Gender Equality Committee, The Wellcome Trust Centre for Human Genetics
2013-present	Oxford Females in Engineering, Science and Technology (OxFEST)
2006-present	Society for Molecular Biology and Evolution
2014	American Society of Human Genetics
2003-2009	Institute of Molecular Evolutionary Genetics

Conferences and Symposia

2010	Technical staff, Society for Molecular Biology and Evolution Annual Meeting (Lyon, France)
2006	Technical support, CSHL Biology of Genomes (Cold Spring Harbor Laboratory, NY)
2005	Technical support, Penn State 24th Summer Symposium in Molecular Biology "Comparative and Functional Genomics" (University Park, PA)

Review of manuscripts

- *Bioinformatics*
- *BMC Evolutionary Biology*
- *Genome Biology*
- *Human Mutation*
- *Journal of Molecular Evolution*
- *Molecular Biology and Evolution*
- *Molecular Genetics and Genomics*
- *Nature Scientific Reports*
- *Nucleic Acids Research*
- *PLoS Genetics*

Outreach

2016	Volunteer, Oxford Biomedical Research Centre Open Day, John Radcliffe Hospital, Oxford, UK
2015-2016	Volunteer, Question of Taste DNA Workshop, The Museum of Natural History, Oxford UK
2014	Volunteer, Science in Your World, Oxfordshire Science Festival, Oxford, UK
2010	Volunteer**,** 19 édition de la Fête de la Science "Biodiversité, biotechnologie et bioéthique, quels défis pour l'avenir ?" Lyon, France

COMPUTATIONAL SKILLS

Languages

- R
- Perl
- AWK
- MySQL
- HTML

Bioinformatics tools/packages

- BWA, Stampy
- SAM tools, Picard tools
- BED tools
- VCF tools, BCF tools
- MEGA, HyPHy, PAML
- CLUSTAL, MUSCLE, BLAST, BlastZ
- Galaxy, EMBOSS suite
- UCSC Table Browser/Genome Browser, IGV

- Plink

Operating Systems

- Linux/Unix
- OSX, Windows

Other

- Git/github version control
- HPC cluster job management (PBS, SGE, Torque)
- BASH shell scripting
- *nix utilities