

Guillaume Bourque
Dept. Human Genetics, McGill University
McGill University and Genome Quebec Innovation Centre

WORK EXPERIENCE

2010 – ...	Associate Professor Department of Human Genetics, McGill University	<i>Montréal, Qc</i>
2010 – ...	Director of Bioinformatics McGill University and Génome Québec Innovation Center	<i>Montréal, Qc</i>
2007 – 2010	Senior Group Leader & Assoc Director Computational & Mathematical Biology, Genome Institute of Singapore	<i>Singapore</i>
2004 – 2007	Group Leader Computational & Mathematical Biology, Genome Institute of Singapore	<i>Singapore</i>

ACADEMIC AND TRAINING BACKGROUND

2002 – 2004	Postdoctoral researcher Centre de Recherches Mathématiques	<i>Montréal, QC</i>
2000 – 2002	Ph.D. in Applied Mathematics Université de Montréal	<i>Los Angeles, CA</i>
1998 – 2000	M.A. in Applied Mathematics University of Southern California	<i>Los Angeles, CA</i>
1995 – 1998	B.Sc. in Computer Science and Mathematics Université de Montréal	<i>Montréal, QC</i>

SPECIAL ACCOMPLISHMENTS AND HONOURS

2015 – 2017	Distinguished Visiting Professor, Kyoto University, Japan
2012 – 2016	Chercheur-boursier Junior 2, Fonds de recherche Santé Québec (FRSQ)

MOST SIGNIFICANT CONTRIBUTIONS

1. Lu X, Sachs F, Ramsay L, Jacques PÉ, Göke J, **Bourque G@**, Ng HH. The retrovirus HERVH is a long noncoding RNA required for human embryonic stem cell identity. *Nat Struct Mol Biol.* 2014 Apr;21(4):423-5. @co-corresponding

In this study we show that the long terminal repeats of the endogenous retrotransposon HERVH function as enhancers and that HERVH is a nuclear long noncoding RNA required to maintain human embryonic stem cells (hESCs) identity. Furthermore, HERVH is associated with OCT4, coactivators and Mediator subunits. Together, these results uncover a new role of species-specific transposable elements in hESCs.

2. Jacques PE, Jeyakani J, **Bourque G@**. 2013. The majority of primate-specific regulatory sequences are derived from transposable elements. *PLoS Genet.* 9(5):e1003504. There is growing evidence showing that some of these transposon-derived sequences have been a source of new binding sites for various mammalian transcription factors. To systematically survey this contribution, we used datasets generated by the international Encyclopedia of DNA Elements (ENCODE) consortium, identifying the location of active regulatory elements in more than 40 distinct human cell types. Our results demonstrate that transposon-derived sequences have contributed hundreds of thousands of novel regulatory elements to the primate lineage and reshaped the human transcriptional landscape.
3. Kunarso G, Chia NY, Jeyakani J, Hwang C, Lu X, Chan YS, Ng H.H., **Bourque G@**. (2010) Transposable Elements have Rewired the Core Regulatory Network of Human Embryonic Stem Cells. *Nat. Genet.* 42:631-634. We studied the genomic locations of three key regulatory proteins (OCT4, NANOG and CTCF) in human and mouse embryonic stem (ES) cells. In contrast to CTCF, we found that the binding profiles of OCT4 and NANOG are drastically different with only ~5% of the regions homologously occupied. We showed that transposable elements have contributed up to 25% of the bound sites in both species and have wired new genes into the core regulatory network of ES cells. These data indicate that species-specific transposable elements have profoundly altered the transcriptional circuitry of stem cells.

Guillaume Bourque
Dept. Human Genetics, McGill University
McGill University and Genome Quebec Innovation Centre

4. Establishment of the Canadian Center for Computational Genomics (C3G), a Genome Canada platform that provides bioinformatics services to the genomics community. Recruit and manage a team of 25 bioinformaticians, software developers and IT specialists and plan a compute infrastructure to support the McGill Genome Center with a number of heterogeneous high-throughput platforms (microarray, sequencing, genotyping, etc.) including 12 HiSeqs next-generation sequencers, 1 PacBio, 1 IonTorrent. Develop open-source tools to support the genomics community such as the Genetics and genomics Analysis Platform (GenAP, www.genap.ca), which includes open-source data analysis pipelines for whole-genome sequencing, exome sequencing, transcriptome sequencing and metagenomics.
5. Head the Integrative Epigenomic Data Coordination Center at McGill and co-Investigator of the Epigenomic Mapping Center at McGill. These two initiatives are part of the Canadian Epigenetics, Environment and Health Research Consortium (CEEHRC) funded by CIHR to support the International Human Epigenome Consortium (IHEC). The objective of the Data Coordination Center is to develop a framework that leverages Compute Canada national resources to support large-scale processing, sharing and visualization of epigenomics data. With the IHEC Data Portal that we have developed (see epigenomesportal.ca/ihec), national and international epigenetic researchers can now query and exploit epigenome reference maps that have been produced by IHEC.

SELECTED PEER-REVIEWED PUBLICATIONS

1. * Hocking TD, Rigai G, **Bourque G**@. 2015. PeakSeg: constrained optimal segmentation and supervised penalty learning for peak detection in count data. *Proceedings of the 32nd International Conference on Machine Learning*. ICML-15 324-332.
2. * Hocking TD, **Bourque G**. 2015. PeakSegJoint: fast supervised peak detection via joint segmentation of multiple count data samples. *arXiv* arXiv:1506.01286.
3. Aarabi M, San Gabriel MC, Chan D, Behan NA, Caron M, Pastinen T, **Bourque G**, MacFarlane AJ, Zini A, Trasler J. 2015. High dose folic acid supplementation alters the human sperm methylome and is influenced by the MTHFR C677T polymorphism. *Hum Mol Genet*. Aug 24. pii: ddv338.
4. Hoen DR, Hickey G, **Bourque G**, Casacuberta J, Cordaux R, Feschotte C, Fiston-Lavier AS, Hua-Van A, Hubley R, Kapusta A, Lerat E, Maumus F, Pollock DD, Quesneville H, Smit A, Wheeler TJ, Bureau TE, Blanchette M. 2015. A call for benchmarking transposable element annotation methods. *Mob DNA*. Aug 4;6:13.
5. Sayadi A, Jeyakani J, Seet SH, Wei CL, **Bourque G**, Bard FA, Jenkins NA, Copeland NG, Bard-Chapeau EA. 2015. Functional features of EVI1 and EVI1 Δ 324 isoforms of MECOM gene in genome-wide transcription regulation and oncogenicity. *Oncogene*. Aug 3.
6. Dyke SO, Cheung WA, Joly Y, Ammerpohl O, Lutsik P, Rothstein MA, Caron M, Busche S, **Bourque G**, Rönblom L, Flicek P, Beck S, Hirst M, Stunnenberg H, Siebert R, Walter J, Pastinen T. 2015. Epigenome data release: a participant-centered approach to privacy protection. *Genome Biol*. Jul 17;16:142.
7. McGraw S, Zhang JX, Farag M, Chan D, Caron M, Konermann C, Oakes CC, Mohan KN, Plass C, Pastinen T, **Bourque G**, Chaillet JR, Trasler JM. 2015. Transient DNMT1 suppression reveals hidden heritable marks in the genome. *Nucleic Acids Res*. Jan 10. pii: gku1386
8. * Scelo G, Riazalhosseini Y, Greger L, Letourneau L, et al. 2014. Whole-genome sequencing reveals variation in the genomic landscape of clear cell renal cell carcinoma in Europe. *Nat Commun*. 29;5:5135.
9. * Hocking TD, Goerner-Potvin P, Morin A, Shao X, **Bourque G**. 2015. Visual annotations and a supervised learning approach for evaluating and calibrating ChIP-seq peak detectors. *arXiv* arXiv:1409.6209.

Guillaume Bourque
Dept. Human Genetics, McGill University
McGill University and Genome Quebec Innovation Centre

10. Chen ES, Gigeck CO, Rosenfeld JA, Diallo AB, Maussion G, Chen GG, Vaillancourt K, Lopez JP, Crapper L, Poujol R, Shaffer LG, **Bourque G**, Ernst C. 2014. Molecular convergence of neurodevelopmental disorders. *Am J Hum Genet.* 95(5):490-508.
11. Lu X, Sachs F, Ramsay L, Jacques PE, Göke J, **Bourque G@**, Ng HH. 2014. The retrovirus HERVH is a long noncoding RNA required for human embryonic stem cell identity. *Nat Struct Mol Biol.* 21(4):423-5.
12. Salem S, Langlais D, Lefebvre F, **Bourque G**, Bigley V, Haniffa M, Casanova JL, Burk D, Berghuis A, Butler KM, Leahy TR, Hambleton S, Gros P. 2014. Functional characterization of the human dendritic cell immunodeficiency associated with the IRF8K108E mutation. *Blood.* blood-2014-04-570879.
13. * Jacques PE, Jeyakani J, **Bourque G@**. 2013. The majority of primate-specific regulatory sequences are derived from transposable elements. *PLoS Genet.* 9(5):e1003504.
14. Kapusta A, Kronenberg Z, Lynch VJ, Zhuo X, Ramsay L, **Bourque G**, Yandell M, Feschotte C. 2013. Transposable elements are major contributors to the origin, diversification, and regulation of vertebrate long noncoding RNAs. *PLoS Genet.* 9(4):e1003470.
15. Lu X, Göke J, Sachs F, Jacques PE, Liang H, Feng B, **Bourque G**, Bubulya PA, Ng HH. 2013. SON connects the splicing-regulatory network with pluripotency in human embryonic stem cells. *Nat Cell Biol.* 15(10):1141-1152.
16. Chaveroux C, Eichner LJ, Dufour CR, Shatnawi A, Khoutorsky A, **Bourque G**, Sonenberg N, Giguère V. 2013. Molecular and genetic crosstalks between mTOR and ERR α are key determinants of rapamycin-induced nonalcoholic fatty liver. *Cell Metab.* 17(4):586-98.
17. Nagarajan N, Bertrand D, Hillmer AX, Zang ZJ, Yao F, Jacques PE, Teo ASM, Cutcutache I, Zhang Z, Lee WH, Sia YY, Gao S, Ariyaratne PN, Ho A, Woo XY, Veeravali L, Ong CK, Deng N, Desai KV, Khor CC, Hibberd ML, Shahab A, Rao J, Wu M, The M, Zhu F, Chin SY, Pang B, So JBY, **Bourque G**, Soong R, Sung WK, Teh BT, Rozen S, Ruan X, Yeoh KG, Tan PBO and Ruan Y. 2013. Whole-genome reconstruction of host and pathogen sequences and mutational signatures in gastric cancer. *Genome Biology.* 13(12):R115.
18. Khuong-Quang DA, Buczkowicz P, Rakopoulos P, Liu XY, Fontebasso AM, Bouffet E, Bartels U, Albrecht S, Schwartzenruber J, Letourneau L, Bourgey M, **Bourque G**, Montpetit A, Bourret G, Lepage P, Fleming A, Lichter P, Kool M, von Deimling A, Sturm D, Korshunov A, Faury D, Jones DT, Majewski J, Pfister SM, Jabado N, Hawkins C. 2012. K27M mutation in histone H3.3 defines clinically and biologically distinct subgroups of pediatric diffuse intrinsic pontine gliomas. *Acta Neuropathol.* 124(3):439-47.
19. Pott S, Kamrani NK, **Bourque G**, Pettersson S, Liu ET. 2012. PPARG Binding Landscapes in Macrophages Suggest a Genome-Wide Contribution of PU.1 to Divergent PPARG Binding in Human and Mouse. *PLoS One.* 7(10):e48102.
20. Bard-Chapeau EA, Jeyakani J, Kok CH, Muller J, Chua BQ, Gunaratne J, Batagov A, Jenjaroenpun P, Kuznetsov VA, Wei CL, D'Andrea RJ, **Bourque G**, Jenkins NA, Copeland NG. 2012. Ecotopic viral integration site 1 (EV1) regulates multiple cellular processes important for cancer and is a synergistic partner for FOS protein in invasive tumors. *Proc Natl Acad Sci U S A.* 109(6):2168-73.
21. Yao F, Ariyaratne PN, Hillmer AM, Lee WH, Li G, Teo AS, Woo XY, Zhang Z, Chen JP, Poh WT, Zawack KE, Chan CS, Leong ST, Neo SC, Choi PS, Gao S, Nagarajan N, Thoreau H, Shahab A, Ruan X, Cacheux-Rataboul V, Wei CL, **Bourque G**, Sung WK, Liu ET, Ruan Y. 2012. Long span DNA paired-end-tag (DNA-PET) sequencing strategy for the interrogation of genomic structural mutations and fusion-point-guided reconstruction of amplicons. *PLoS One.* 7(9):e46152.
22. Handoko L, Xu H, Li G, Ngan CY, Chew E, Schnapp M, Lee CW, Ye C, Ping JL, Mulawadi F, Wong E, Sheng J, Zhang Y, Poh T, Chan CS, Kunarso G, Shahab A, **Bourque G**, Cacheux-Rataboul V, Sung WK,

Guillaume Bourque
 Dept. Human Genetics, McGill University
 McGill University and Genome Quebec Innovation Centre

- Ruan Y, Wei CL. 2011. CTCF-mediated functional chromatin interactome in pluripotent cells. *Nat Genet.* 43(7):630-8.
23. Zemojtel T, Kielbasa SM, Arndt PF, Behrens S, **Bourque G**, Vingron M. 2011. CpG deamination creates transcription factor binding sites with high efficiency. *Genome Biol Evol.* 3:1304-11.
24. Chatterjee S, **Bourque G**, Lufkin T. 2011. Conserved And Non-Conserved Enhancers Direct Tissue Specific Transcription In Ancient Germ Layer Specific Developmental Control Genes. *BMC Dev Biol.* 11(1):63.
25. Hillmer AM, Yao F, Inaki K, Lee WH, Ariyaratne PN, Teo AS, Woo XY, Zhang Z, Zhao H, Ukil L, Chen JP, Zhu F, So JB, Salto-Tellez M, Poh WT, Zawack KF, Nagarajan N, Gao S, Li G, Kumar V, Lim HP, Sia YY, Chan CS, Leong ST, Neo SC, Choi PS, Thoreau H, Tan PB, Shahab A, Ruan X, Bergh J, Hall P, Cacheux-Rataboul V, Wei CL, Yeoh KG, Sung WK, **Bourque G**, Liu ET, Ruan Y. 2011. Comprehensive long-span paired-end-tag mapping reveals characteristic patterns of structural variations in epithelial cancer genomes. *Genome Res.* 21:665-675.
26. Inaki K, Hillmer AM, Ukil L, Yao F, Woo XY, Vardy LA, Zawack KF, Lee CW, Ariyaratne PN, Chan YS, Desai KV, Bergh J, Hall P, Putti TC, Ong WL, Shahab A, Cacheux-Rataboul V, Karuturi RK, Sung WK, Ruan X, **Bourque G**, Ruan Y, Liu ET. 2011. Transcriptional consequences of genomic structural aberrations in breast cancer. *Genome Res.* 21:676-687.
27. Kunarso G, Chia NY, Jeyakani J, Hwang C, Lu X, Chan YS, Ng HH, **Bourque G@**. 2010. Transposable Elements have Rewired the Core Regulatory Network of Human Embryonic Stem Cells. *Nat Genet.* 42:631-634.
28. Lin CH, Zhao H, Lowcay SH, Shahab A, **Bourque G@**. 2010. webMGR: an online tool for multiple genome rearrangement. *Bioinformatics.* 26(3):408-10.

SELECTED RESEARCH FUNDING (as PI first)

2015 – 2017	Genome Canada, Genomics Innovation Network Role: Project Lead Title: Canadian Center for Computational Genomics (C3G)	\$ 1,050,000
2015 – 2017	Genome Canada, Genomics Innovation Network Role: Project Lead Title: Canadian Center for Computational Genomics (C3G) – Tech. Development	\$ 527,000
2012 – 2017	CIHR, CEEHRC Epigenomics Platform Role: Principal Investigator Title: Integrative Epigenomic Data Coordination Centre (EDCC) at McGill	\$ 1,500,000
2011 – 2016	CIHR, Operating Grant Role: Principal Investigator Title: Functional characterization of the endogenous retrovirus HERV-H family in hESCs	\$ 426,482
2015 – 2019	CIHR, CEEHRC Consortium Network Role: Principal Applicant (NPA Hirst, share 7%) Title: Canadian Epigenetics, Environment and Health Research Consortium Network	\$ 2,000,000
2015 – 2019	CFI, Innovation Fund Role: Principal User (PL Jones, share 5%) Title: Canada's Genomics Enterprise (CGEn): A national genomic tools network	\$ 58,435,000
2015 – 2017	Genome Canada, Genomics Innovation Network Role: co-Lead (PL Lathrop, share 20%) Title: McGill University and G�enome Qu�ebec Innovation Center	\$ 2,000,000
2014 – 2018	NSERC, Discovery Frontiers Role: co-Investigator (PI Stein, share 7%) Title: The Cancer Genome Collaboratory	\$ 6,000,000
2012 – 2017	CIHR, CEEHRC Epigenomics Platform Role: co-Investigator (PI Lathrop, share 15%) Title: Multidimensional Epigenomics Mapping Centre (EMC) at McGill	\$ 6,000,000