

Orion Buske

buske@cs.toronto.edu
www.cs.toronto.edu/~buske

686 Bay St, Room 12.9840
Toronto, ON M5G 0A4
Canada

Education

- **PhD, Computer Science** *Sep 2016*
University of Toronto, Toronto, Canada. Advisor: Michael Brudno.
- **MSc, Computer Science** *Jan 2012*
University of Toronto, Toronto, Canada.
- **BSc, Computer Engineering, magna cum laude** *Jun 2009*
University of Washington, Seattle, USA. Advisor: Martin Tompa.
- **BSc, Bioengineering, magna cum laude** *Jun 2009*
University of Washington, Seattle, USA. Advisors: Chris Neils, Michael Regnier.

Research

Theses

- “Computational methods for predicting and validating the causes of Mendelian disease.” *University of Toronto, Department of Computer Science*. 2017. Supervisor: Michael Brudno. Chair: Deborah Zamble. Committee: Anna Goldenberg, Gary Bader, Quaid Morris, Stephen Meyn, Shamil Sunyaev.
- “Identification of deleterious synonymous variants in human genomes.” *University of Toronto, Department of Computer Science*. 2012. Supervisor: Michael Brudno. Reader: Anna Goldenberg.
- “AutoOrthoGen: Multiple genome alignment and comparison.” *University of Washington, Computer Science & Engineering*. 2009. Supervisor: Martin Tompa.
- “HeartBeat: Design and development of a headphone-mounted infrared heart rate monitor.” *University of Washington, Bioengineering*. 2009. Supervisors: Chris Neils, Michael Regnier.

Refereed Publications

- 17 Wang Z, Brudno M, **Buske O**. 2017. Towards a Directory of Rare Disease Specialists: Identifying Experts from Publication History *Proceedings of Machine Learning Research*, 68
- 16 Sobreira NLM, Arachchi H, **Buske OJ**, Chong JX, Hutton B, *et al.* 2017. Matchmaker Exchange. *Current Protocols in Human Genetics*, 95
- 15 Köhler S, Vasilevsky NA, Engelstad M, Foster E, McMurry J, *et al.* 2017. The human phenotype ontology in 2017. *Nucleic Acids Research*, 45
- 14 Labrie V*, **Buske OJ***, Oh E, Jeremian R, Ptak C, *et al.* 2016. Lactase non-persistence is directed by DNA variation-dependent epigenetic aging. *Nature Structural and Molecular Biology*, 23
- 13 Bone WP, Washington NL, **Buske OJ**, Adams DR, Davis J, *et al.* 2015. Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. *Genetics in Medicine*, 6
- 12 Smedley D, Jacobsen J, Jäger M, Köhler S, *et al.* 2015. Next-generation diagnostics and disease gene discovery with the Exomiser. *Nature Protocols*, 10

- 11 **Buske OJ***, Girdea M*, Dumitriu S, Gallinger B, Hartley T, *et al.* 2015. PhenomeCentral: a Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. *Human Mutation*, 36
- 10 **Buske OJ**, Schiettecatte F, Hutton B, Dumitriu S, Misyura A, *et al.* 2015. The Matchmaker Exchange API: automating patient matching through the exchange of structured phenotypic and genotypic profiles. *Human Mutation*, 36
- 9 Philippakis AA, Azzariti DR, Beltran S, Brookes AJ, Brownstein CA, *et al.* 2015. The Matchmaker Exchange: a platform for rare disease gene discovery. *Human Mutation*, 36
- 8 Krawitz P, **Buske O**, Zhu Na, Brudno M, Robinson PN. 2015. The Genomic Birthday Paradox: How Much is Enough? *Human Mutation*, 36
- 7 **Buske OJ**, Manickaraj A, Mital S, Ray PN, Brudno M. 2013. Identification of deleterious synonymous variants in human genomes. *Bioinformatics*, 29(15):1843–1850
- 6 Mezlini AZ, Smith EJM, Fiume M, **Buske O**, Savich G, *et al.* 2012. iReckon: Simultaneous isoform discovery and abundance estimation from RNA-seq data. *Genome Research*, 23(3):519–529
- 5 The ENCODE Project Consortium. 2012. An integrated encyclopedia of DNA elements in the human genome. *Nature*, 489:57-74
- 4 Hoffman MM, **Buske OJ**, Wang J, Weng Z, Bilmes JA, Noble WS. 2012. Unsupervised pattern discovery in human chromatin structure through genomic segmentation. *Nature Methods*, 9(5):473–476
- 3 **Buske OJ**, Hoffman MM, Ponts N, Le Roch KG, Noble WS. 2011. Exploratory analysis of genomic segmentations with Segtools. *BMC Bioinformatics*, 12(1):415
- 2 The ENCODE Project Consortium. 2011. A User’s Guide to the Encyclopedia of DNA Elements (ENCODE). *PLoS Biology*, 9(4):e1001046
- 1 Hoffman MM, **Buske OJ**, Noble WS. 2010. The Genomedata format for storing large-scale functional genomics data. *Bioinformatics*, 26(11):1458–1459

Invited Talks

- 18 “Towards patient self-phenotyping with RareConnect and PhenoTips.” *European Human Genetics Conference (ESHG)*, Stockholm, Sweden. 2017.
- 17 “Structured data for patient matchmaking.” *10th Annual Biohackathon*, Tokyo, Japan. 2017.
- 16 “Looking towards patient self-phenotyping on RareConnect in combination with PhenoTips.” *European Human Genetics Conference (ESHG)*, Copenhagen, Denmark. 2017.
- 15 “RareConnect: A network of global rare disease communities.” *E-Rare Data Workshop*, Berlin, Germany. 2017.
- 13 “RareConnect: A network of global rare disease communities.” *E-Rare Workshop*, Berlin, Germany. 2017.
- 12 “The evolving world of patient discovery platforms.” *Canadian Expert Patients in Health Technology Conference*, Toronto, Canada. 2016.
- 11 “The Matchmaker Exchange: a federated platform for discovering similar patients & rare disease genes.” *Festival of Genomics*, San Diego, CA. 2016.
- 10 “Patient matchmaking over a federated network.” *9th Annual BioHackathon*, Tsuruoka, Japan. 2016.
- 9 “Making undiagnosed patients discoverable with PhenomeCentral and the Matchmaker Exchange.” *5th Annual Canadian Human and Statistical Genetics Meeting*, Halifax, Canada. 2016.

- 8 “Connecting Rare Disease Patient Databases with the Matchmaker Exchange API.” *Bio-IT World Conference and Expo*, Boston, MA. 2016.
- 7 “Exchanging case summaries to discover rare disease cohorts across organizations.” *2nd RDF Summit for Individual Genomics*, Sendai, Japan. 2016.
- 6 “PhenomeCentral: Canadian-made rare disease patient registry.” *CORD Rare Disease Day Conference*, Toronto, Canada. 2015.
- 5 “PhenomeCentral: an integrated portal for sharing and searching patient data for rare genetic disorders.” *Hospital for Sick Children, Genetics and Genome Biology Retreat*, Toronto, Canada. Best Presentation Award. 2014.
- 4 “Identification of deleterious synonymous variants in human genomes.” *HiTSeq: Conference on High Throughput Sequencing Methods and Applications*, Berlin, Germany. 2013.
- 3 “Variant detection and the Autism sequencing project.” *HiTSeq: Conference on High Throughput Sequencing Methods and Applications*, Vienna, Austria. 2011.
- 2 “Variant detection and the Autism sequencing project.” *7th ISCB Student Council Symposium*, Vienna, Austria. *BMC Bioinformatics*, 12(Suppl 11):A4 . Best Presentation Award: Third Place. 2011.
- 1 “Semi-supervised enhancer prediction using the Segway framework.” *ENCODE Project Consortium Conference*, Bethesda, MD. 2010.

Posters

- 3 **Buske OJ**, Girdea M, Dumitriu S, Gallinger B, Hartley T, *et al.* 2015. PhenomeCentral: a portal for phenotypic and genotypic matchmaking of patients with rare genetic diseases. *ASHG*, Baltimore, MD.
- 2 **Buske O**, Manickaraj A, Mital S, Brudno M. 2012. Identification of deleterious synonymous variants in human genomes. *RECOMB*, Barcelona, Spain.
- 1 **Buske O**, Hoffman M, Noble W. 2009. Exploratory analysis of a genomic segmentation with segtools. *Biomedical Computation at Stanford Symposium*, Stanford University.

Teaching

Instructor

- 1 Computer Programming II (CSC148), University of Toronto *Sum 2012*
Lectured weekly, designed assignments and labs, managed discussion board, and administered introductory computer science course in Python.

Teaching Assistant

- 11 “Computing for Medicine,” University of Toronto *Spr 2016*
Helped students during software development bootcamps.
- 10 Computer Science Undergraduate Help Centre, University of Toronto *Aut 2014*
Helped students from all undergraduate courses.
- 9 Software Tools and Systems Programming (CSC209), University of Toronto *Win 2014*
Ran tutorials, marked assignments, and held office hours.
- 8 Ramp-up sessions for Computer Programming II, University of Toronto *2012–2014*
Developed slide deck and taught two six-hour ramp-up sessions per term. *(3 terms)*
- 7 Computer Programming II (CSC148), University of Toronto *Aut 2013*
Taught lab sections for introductory computer science course in Python.

6	“Learn to Program: The Fundamentals,” University of Toronto/Coursera Monitored discussion board and helped administer massive open online course.	<i>Sum 2013</i>
5	“The why and how of computing” (CSC104), University of Toronto Taught lab sections and office hours, marked assignments, and helped design projects for introductory computing course.	<i>Win 2013</i>
4	Software Design (CSC207), University of Toronto Taught lab sections for undergraduate Java course.	<i>Aut 2011</i>
3	Software Carpentry, Toronto Designed assignments and slide decks for online programming course for scientists and engineers.	<i>Win 2011</i>
2	Computer Programming I (CSC108), University of Toronto Taught lab sections for introductory Python course.	<i>Aut 2010</i>
1	Computer Programming I (CSE142), University of Washington Taught tutorials, supervised programming lab, managed discussion board, and marked homework for introductory Java course.	<i>2006–2008 (5 terms)</i>

Undergraduate Student Mentorship

I’ve been honoured to work with the following talented undergraduates on independent research and development projects:

·	Julian Mazzitelli	<i>2017</i>
·	Zihan Wang	<i>2016</i>
·	Faye Feng	<i>2016</i>
·	Nick Frosst (to Google Brain)	<i>2015</i>
·	Tal Friedman (to UCLA)	<i>2014</i>
·	David Madras (to University of Toronto)	<i>2014</i>
·	Jonathan Zung (to Princeton)	<i>2013</i>

Awards

·	Hospital for Sick Children Restracom Scholarship, “ <i>funds exceptional researchers working under the supervision of a SickKids scientist</i> ”	<i>2013–2016</i>
·	University of Toronto McLaughlin Centre Training Award	<i>2015</i>
·	Hospital for Sick Children GGB Retreat, Best Presentation Award	<i>2014</i>
·	C.C. Gotlieb Graduate Fellowship in the Department of Computer Science, “ <i>recognizing an outstanding graduate student</i> ”	<i>2013</i>
·	Enbridge Gas Distribution Student Bursary, “ <i>to support one of our highest ranked graduate students</i> ”	<i>2013</i>
·	University of Toronto SGS Conference Grant	<i>2013</i>
·	National Science Foundation Travel Award, RECOMB 2012	<i>2012</i>
·	Ontario Graduate Scholarship, “ <i>to Ontario’s best graduate students in all disciplines</i> ”	<i>2011</i>
·	Bob Bandes Memorial Award for Excellence in Teaching, Honorable Mention “ <i>to recognize exceptional undergraduate and graduate teaching assistants</i> ”	<i>2008</i>
·	University of Washington Undergraduate Scholar Award	<i>2005</i>
·	Spokane Rotary West Scholarship	<i>2005</i>

Industry

·	Biotech Research Assistant, Amgen Inc., Seattle, Washington	<i>Sum 2005</i>
---	---	-----------------

Side projects

- Co-founder, StagePage: Audience engagement platform for the performing arts. *2014–present*
- Co-founder, EasyType French Accents: French typing software. *2011–2014*
- Co-founder, Map What’s Up: Visualizer for geo-located Twitter data. *2011*
- UI lead, Banda: Peer-to-peer paid media application. *2008*

Community and Outreach

- Panelist, “Designing Rare Alliance Canada: Opportunities Created.”
CORD Conference. Toronto, Canada. *Sep 2017*
- Panelist, “Panel discussion on data sharing.” 5th International Conference on
Rare and Undiagnosed Diseases. Stockholm, Sweden. *Aug 2017*
- Panelist, “Integrating, tools, platforms and patients participation: what is
desirable and how to get there?” E-Rare Data Workshop. Berlin, Germany. *May 2017*
- Panelist, “Vision for Canadian Rare Disease Networks.” CORD Conference.
Vancouver, Canada. *Mar 2017*
- Mentor, Ladies Learning Code, Toronto. *(3x) 2015–2016*
- Hacking Health for Kids, SickKids, Toronto. *May 2014*
- Graduate Representative, Computer Science Department Social Committee. *2013–2014*
- Weekly social organizer, Computer Science Graduate Student Society. *2011–2014*
- Invited lecture, “An introduction to computational biology.” Appleby College. *(3x) 2011–2013*
- Presentation, “Digital DNA: Bringing computational biology into high school
computer science.” 12th Annual Association for Computer Studies Educators
(ACSE) Conference. *Nov 2011*
- Teaching assistant, Software Carpentry and the.hacker.within bootcamp. *Nov 2011*
- Ran workshop, “Digital DNA.” Computer Science for High School (CS4HS)
Summer Program, University of Toronto. *Jul, Oct 2011*
- Poster, “The Genetics of Autism.” Research in Action, University of Toronto. *Apr 2011*
- Treasurer, ACM Student Chapter, University of Washington. *2009–2010*
- Weekly social co-organizer, ACM Student Chapter, University of Washington. *2009–2010*

Reviewer

- Oxford Bioinformatics *2012–2016*
- PLOS ONE *2016*
- European Journal of Human Genetics *2015*