Orion Buske

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

24 Cohen ASA, *et al.* 2022. Genomic answers for children: Dynamic analyses of > 1000 pediatric rare disease genomes. *Genetics in Medicine*

23 Driver HG, *et al.* 2022. Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery. *Human Mutation*

22 Laurie S, *et al.* 2022. The RD-Connect Genome-Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. *Human Mutation*

21 Osmond M, *et al.* 2022. Outcome of over 1500 matches through the Matchmaker Exchange for rare disease gene discovery: The 2-year experience of Care4Rare Canada. *Genetics in Medicine*, 24

20 Rehm HL, *et al.* 2021. GA4GH: International policies and standards for data sharing across genomic research and healthcare. *Cell genomics*, 1

19 Köhler S, Øien NC, **Buske OJ**, *et al.* 2019. Encoding clinical data with the human phenotype ontology for computational differential diagnostics. *Current protocols in human genetics*, 103

18 Fujiwara T, Yamamoto Y, Kim JD, **Buske O**, Takagi T. 2018. PubCaseFinder: A case-report-based, phenotype-driven differential-diagnosis system for rare diseases. *Proceedings of Machine Learning Research*, 68

17 Wang Z, Brudno M, **Buske O**. 2017. Towards a Directory of Rare Disease Specialists: Identifying Experts from Publication History. *The American Journal of Human Genetics*, 103

16 Sobreira NLM, Arachchi H, **Buske OJ**, *et al.* 2017. Matchmaker Exchange. *Current Protocols in Human Genetics*, 95

15 Köhler S, *et al.* 2017. The human phenotype ontology in 2017. *Nucleic Acids Research*, 45

14 Labrie V*, **Buske OJ***, *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**, *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, *et al.* 2015. Next-generation diagnostics and disease gene discovery with the Exomiser. *Nature Protocols*, 10

11 **Buske OJ***, Girdea M*, *et al.* 2015. PhenomeCentral: a Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. *Human Mutation*, 36

10 **Buske OJ**, *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, *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**, *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

22 "How structured data is streamlining genomic diagnosis as part of the first pan-Canadian rare disease data repository, Genomics4RD." *A Rare International Dialogue*, Toronto, Canada. 2019.

21 "Using structured data to streamline genomic diagnosis in the first Canadian rare disease research platform, Genomics4RD." *7th Internation Conference on Rare & Undiagnosed Diseases*, Toronto, Canada. 2019.

20 "Helping doctors collect structured data to better diagnose genetic conditions." *11th Annual Biohackathon*, Tokyo, Japan. 2018.

19 "Why is sharing data with researchers so important?" *European Conference on Rare Diseases (ECRD)*, Vienna, Austria. 2018.

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 *Sum 2013*
Monitored discussion board and helped administer massive open online course.

5 "The why and how of computing" (CSC104), University of Toronto *Win 2013*
Taught lab sections and office hours, marked assignments, and helped design projects for introductory computing course.

4 Software Design (CSC207), University of Toronto *Aut 2011*
Taught lab sections for undergraduate Java course.

3 Software Carpentry, Toronto *Win 2011*
Designed assignments and slide decks for online programming course for scientists and engineers.

2 Computer Programming I (CSC108), University of Toronto *Aut 2010*
Taught lab sections for introductory Python course.

1 Computer Programming I (CSE142), University of Washington *2006–2008*
Taught tutorials, supervised programming lab, managed discussion board, and marked homework for introductory Java course. *(5 terms)*

Undergraduate Student Mentorship

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

· Julian Mazzitelli (to BioBox Analytics) *2017*
· Zihan Wang (to Stanford) *2016*
· Faye Feng *2016*
· Nick Frosst (to Google Brain) *2015*
· Tal Friedman (to UCLA) *2014*
· David Madras (to University of Toronto) *2014*
· Jonathan Zung (to Princeton) *2013*

Awards

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

Industry

- Chief Executive Officer, PhenoTips, Toronto, Canada *2018–2023*
- Senior Computer Specialist, Genome Sciences, University of Washington, Seattle, USA *2008–2009*
- Biotech Research Assistant, Amgen Inc., Seattle, USA *Sum 2005*

Side projects

- Co-founder, StagePage: Audience engagement platform for the performing arts. *2014–2020*
- 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

- Session Chair, "Developing tools to empower patient experts." European Conference on Rare Diseases (ECRD). Vienna, Austria *May 2018*
- 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*

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