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Webpage: https://jmonlong.github.io/

Education

2012 - 2018	McGill University, Montreal, Canada.
	PhD in the Human Genetics Department in Guillaume Bourque's Lab.
2010-2011	Universitat Politècnica de Catalunya, Barcelona, Spain.
	Master in Statistic and Operations Research as an exchange student in the
	Faculty of Mathematics and Statistics.
2008 - 2010	Ensimag, Grenoble, France.
	Computer Science and Mathematics course with specialization in Bioinfor-
	matics.
2006 - 2008	Lycée Michel Montaigne, Bordeaux, France.
	Preparatory classes for entrance to the Grandes Écoles. Mathematics and Physics.

Professional Experiences and Projects

2018 - 2012 - 2018	University of California Santa Cruz, Santa Cruz, USA. Postdoctoral researcher with Benedict Paten in the Computational Genomics Lab, analyzing structural variation using variation graphs. Bourque lab, McGill Genome Centre, Montreal, Canada. PhD project. Implementation of a CNV detection method for whole-genome se-
	quencing that can deal with repeat-rich regions. Application to large cohorts of
	normal and disease genomes.
2017	CENTER FOR GENOMIC MEDICINE, Kyoto, Japan.
(3 months)	As part of an exchange program, I visited Dr. Matsuda's group and contributed
	to their ongoing sequencing projects to characterize Japanese genomes.
2017	GENOMICS ENGLAND, London, UK.
(1.5 month)	Visiting the bioinformatics team lead by Dr. Rendon, I contributed to the curation
	of structural variant calls for their genome analysis pipeline.
2011 - 2012	CENTER FOR GENOMIC REGULATION(CRG), Barcelona, Spain.
(1 year)	Graduation project in Roderic Guigó's group. Comparison of splicing activity
	and detection of splicing QTLs from RNA-Seq experiments. Participation in the
	Geuvadis and GTEx projects.
2011	Universitat Politècnica de Catalunya, Barcelona, Spain.
(3 months)	Study of the regularization of the generalized canonical correlation analysis.
2010	Neomades (Mobile software development), Bidart, France.
(3 months)	Implementation of a Java module of their principal product.
2010	Ensimag, Grenoble, France.
(1 month)	Breast cancer modelization and Bayesian estimation of the overdiagnosis rate.
	Collaboration with La Tronche's hospital (Grenoble).

Expertise

tomics.

R, Java, C, Bash, Python, WDL, Snakemake, AWK, Perl, D3/Javascript/HTML/CSS. Git, Linux, High Performance Computing, LaTeX, R/Markdown, Emacs. Structural Variation, Bioinformatics, Genomics, Pangenomics, Transcrip-

${\bf Scholarships} \ {\bf and} \ {\bf Awards}$

2019-2020	BioData Catalyst Fellow. National Heart, Lung, and Blood Institute, USA.
2018	Department Activity Award. Human Genetics Dept, McGill University.
2017	Student Exchange Support Program. JASSO, Japan.
2017	Graduate Mobility Award. Graduate and Postdoctoral Studies, McGill University.
2017	Oral Presentation, Honorable Mention. Human Genetics Dept's Research Day,
	McGill University.
2017	Excellence Award. Human Genetics Dept, McGill University.
2013-2016	Graduate Research Enhancement and Travel Award funding attendance to five
	international conferences. Human Genetics Dept, McGill University.
2015	Excellence Award. Human Genetics Dept, McGill University.
2014	Best oral presentation. Human Genetics Dept's Research Day, McGill University.
Publications	
2023	R Lorig-Roach, M Meredith, J Monlong ,, P Carnevali, K Miga, and B Paten. Phased nanopore assembly with shasta and modular graph phasing with gfase.
	bioRxiv,2023.
2023	M Kolmogorov, KJ Billingsley, M Mastoras, M Meredith, J Monlong ,
	C Blauwendraat, and B Paten. Scalable nanopore sequencing of human genomes
	provides a comprehensive view of haplotype-resolved variation and methylation. bioRxiv, 2023.
2022	G Hickey*, J Monlong *, A Novak, JM Eizenga, Human Pangenome Refer-
	ence Consortium, H Li, and B Paten. Pangenome graph construction from genome
	alignment with minigraph-cactus. bioRxiv, 2022.
2022	W Liao, M Asri, J Ebler, D Doerr, M Haukness, G Hickey, S Lu, JK Lucas, J Monlong ,, G Bourque, MJP Chaisson, P Flicek, AM Phillippy, JM Zook, EE Eichler, D Haussler, ED Jarvis, KH Miga, T Wang, E Garrison, T Marschall, IM Hall, H Li, and B Paten. A Draft Human Pangenome Reference. <i>bioRxiv</i> , 2022.
2022	CP Couturier, J Nadaf, Z Li, S Baig, G Riva, P Le, DJ Kloosterman, J Monlong , A Nkili Meyong, R Allache, T Degenhard, M Al-Rashid, M Guiot, G Bourque, J Ragoussis, L Akkari, FJ Quintana, and K Petrecca. Glioblastoma scRNAseq Shows Treatment-induced, Immune-dependent Rise In Mesenchymal Cancer Cells,
2022	and Structural Variants in Distal Neural Stem Cells. Neuro-Oncology, 2022. SD Goenka, JE Gorzynski, K Shafin, DG Fisk, T Pesout, TD Jensen, J Monlong , P Chang, G Baid,, B Paten, and EA Ashley. Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing. Nature Biotechnology, 2022.
2022	JE Gorzynski, SD Goenka, K Shafin, TD Jensen, DG Fisk, ME Grove, E Spiteri, T Pesout, J Monlong , JA Bernstein,, B Paten, and EA Ashley. Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock. <i>Circulation: Genomic and Precision Medicine</i> , 2022.
2022	JE Gorzynski, SD Goenka, K Shafin, TD Jensen, DG Fisk, ME Grove, E Spiteri, T Pesout, J Monlong ,, B Paten, and EA Ashley. Ultrarapid Nanopore Genome
2021	Sequencing in a Critical Care Setting. New England Journal of Medicine, 2022. J Sirén*, J Monlong*, X Chang*, AM Novak*, JM Eizenga*, C Markello, JA Sibbesen, G Hickey, P Chang, A Carroll, N Gupta, S Gabriel, TW Blackwell, A Ratan, KD Taylor, SS Rich, JI Rotter, D Haussler, E Garrison, and B Paten. Pangenomics enables genotyping of known structural variants in 5202 diverse genomes. Science, 2021.

- E Kuzmin, **J Monlong**, C Martinez, H Kuasne, CL Kleinman, J Ragoussis, G Bourque, and M Park. Inferring Copy Number from Triple-Negative Breast Cancer Patient Derived Xenograft scRNAseq Data Using scCNA. In *Mapping Genetic Interactions*, volume 2381, pages 285–303. Springer US, 2021.
- M Dankner, M Caron, T Al-Saadi, W Yu, V Ouellet, R Ezzeddine, SM Maritan, MG Annis, PU Le, J Nadaf, NS Neubarth, P Savage, D Zuo, CP Couturier, J Monlong, H Djambazian, H Altoukhi, G Bourque, J Ragoussis, RJ Diaz, M Park, M Guiot, S Lam, K Petrecca, and PM Siegel. Invasive growth associated with cold-inducible RNA-binding protein expression drives recurrence of surgically resected brain metastases. Neuro-Oncology, 2021.
- Y Wang, JYP Park, A Pacis, RE Denroche, GH Jang, A Zhang, A Cuggia, C Domecq, **J Monlong**, ..., and G Zogopoulos. A Preclinical Trial and Molecularly Annotated Patient Cohort Identify Predictive Biomarkers in Homologous Recombination—deficient Pancreatic Cancer. *Clinical Cancer Research*, 2020.
- 2020 CP Couturier, S Ayyadhury, PU Le, J Nadaf, **J Monlong**, ..., J Antel, G Bourque, J Ragoussis, and K Petrecca. Single-cell RNA-seq reveals that glioblastoma recapitulates a normal neurodevelopmental hierarchy. *Nature Communications*, 2020.
- 2020 K Shafin, T Pesout, R Lorig-Roach, M Haukness, HE Olsen, ..., J Monlong, ..., KH Miga, P Carnevali, M Jain, and B Paten. Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. Nature Biotechnology, 2020.
- 2020 G Hickey*, D Heller*, **J Monlong***, JA Sibbesen, J Siren, J Eizenga, ET Dawson, E Garrison, AM Novak, and B Paten. Genotyping structural variants in pangenome graphs using the vg toolkit. *Genome Biology*, 2020.
- B Rodriguez-Martin, EG Alvarez, A Baez-Ortega, ..., J Monlong, ..., HH Kazazian, KH Burns, PJ Campbell, and JMC Tubio. Pan-cancer analysis of whole genomes identifies driver rearrangements promoted by LINE-1 retrotransposition. *Nature Genetics*, 2020.
- S Jessa, A Blanchet-Cohen, B Krug, M Vladoiu, M Coutelier, D Faury, B Poreau, N De Jay, S Hébert, **J Monlong**, ..., G Bourque, J Ragoussis, L Garzia, MD Taylor, N Jabado, and CL Kleinman. Stalled developmental programs at the root of pediatric brain tumors. *Nature Genetics*, 2019.
- J Grajcarek, J Monlong, Y Nishinaka-Arai, M Nakamura, M Nagai, S Matsuo, D Lougheed, H Sakurai, MK Saito, G Bourque, and K Woltjen. Genome-wide microhomologies enable precise template-free editing of biologically relevant deletion mutations. *Nature Communications*, 2019.
- **2018 J Monlong**, P Cossette, C Meloche, G Rouleau, SL Girard, and G Bourque. Human copy number variants are enriched in regions of low mappability. *Nucleic Acids Research*, 2018.
- **J Monlong***, SL Girard*, ..., JL Michaud, G Rouleau, BA Minassian, G Bourque, and P Cossette. Global characterization of copy number variants in epilepsy patients from whole genome sequencing. *PLOS Genetics*, 2018.
- 2017 M Arseneault*, J Monlong*, ..., M Lathrop, G Bourque, and Y Riazalhosseini. Loss of chromosome Y leads to down regulation of KDM5D and KDM6C epigenetic modifiers in clear cell renal cell carcinoma. *Scientific Reports*, 2017.
- F Hamdan, CT Myers, P Cossette, P Lemay, D Spiegelman, A Dionne-Laporte, C Nassif, O Diallo, **J Monlong**, ..., BA Minassian, and JL Michaud. High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies.

 American journal of human genetics, 2017.
- **The GTEx Consortium**. Genetic effects on gene expression across human tissues. *Nature*, 2017.

A Saha, et al., and **The GTEx Consortium**. Co-expression networks reveal the 2017 tissue-specific regulation of transcription and splicing. Genome research, 2017. 2017 F Yang, et al., and The GTEx Consortium. Identifying cis-mediators for transeQTLs across many human tissues using genomic mediation analysis. Genome research, 2017. 2017 X Li, et al., and The GTEx Consortium. The impact of rare variation on gene expression across tissues. *Nature*, 2017. 2017MH Tan, et al., and The GTEx Consortium. Dynamic landscape and regulation of RNA editing in mammals. Nature, 2017. 2017 T Tukiainen, et al., and The GTEx Consortium. Landscape of X chromosome inactivation across human tissues. Nature, 2017. 2015 M Melé, PG Ferreira, F Reverter, DS DeLuca, J Monlong, M Sammeth, ..., and The GTEx Consortium. The human transcriptome across tissues and individuals. Science, 2015. 2015 The GTEx Consortium. The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans. Science, 2015. D Pervouchine, S Djebali, A Breschi, CA Davis, PP Barja, A Dobin, A Tanzer, 2015 J Lagarde, C Zaleski, L See, M Fastuca, J Drenkow, H Wang, G Bussotti, B Pei, S Balasubramanian, J Monlong, ..., C Notredame, R Guigo, and TR Gingeras. Enhanced transcriptome maps from multiple mouse tissues reveal evolutionary constraint in gene expression. Nature Communications, 2015. 2014 J Monlong, M Calvo, PG Ferreira, and R Guigó. Identification of genetic variants associated with alternative splicing using sQTLseekeR. Nature Communications, 2014. 2014PG Ferreira, P Jares, D Rico, G Gomez-Lopez, A Martinez-Trillos, N Villamor, S Ecker, A Gonzalez-Perez, DG Knowles, J Monlong, ..., E Campo, and R Guigo. Transcriptome characterization by RNA sequencing identifies a major molecular and clinical subdivision in chronic lymphocytic leukemia. Genome research, 2014. 2014 L Greger, J Su, J Rung, PG Ferreira, The Geuvadis Consortium, T Lappalainen, ET Dermitzakis, and A Brazma. Tandem RNA chimeras contribute to transcriptome diversity in human population and are associated with intronic genetic variants. PloS one, 2014. 2013 T Lappalainen, M Sammeth*, MR Friedlander*, PA t Hoen*, J Monlong*, MA Rivas*, and et al. Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, 2013. 2013 PC t Hoen, et al., and The Geuvadis Consortium. Reproducibility of highthroughput mRNA and small RNA sequencing across laboratories. Nature biotechnology, 2013. 2013 The GTEx Consortium. The Genotype-Tissue Expression (GTEx) project. Nat Genet, 2013. 2011 A Seigneurin, O François, J Labarere, P Oudeville, **J Monlong**, and M Colonna. Overdiagnosis from non-progressive cancer detected by screening mammography: stochastic simulation study with calibration to population based registry data.

Selected Scientific Presentations

2019	NΥ,	USA,	Cold	Spring	Harbor	Laboratory.
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Oral presentation, *Genome Informatics* conference.

2017 QUEBEC, CANADA, Quebec.

BMJ, 2011.

Oral presentation, 6th Annual Canadian Human and Statistical Genetics Meeting.

2016 Japan, Kyoto.

Oral presentation, 13th International Congress of Human Genetics.

2015 BC, CANADA, Vancouver.

Oral presentation, 4rd Annual Canadian Human and Statistical Genetics Meeting.

2014 UK, Cambridge.

Oral presentation, *Genome Informatics* conference.

Other Scientific Presentations

2020	NY, USA, New York City.
	Oral presentation, GSP-TOPMed Analysis Workshop.
2019	TX, USA, Houston.
	Oral presentation, GRC-GIAB Workshop at ASHG.
2019	CA, USA, UC Davis.
	Oral presentation, Northern California Computational Biology Symposium.
2017	Quebec, Canada, Montreal.
	Oral presentation, Human Genetics Dept. Research Day.
2016	Spain, Barcelona.
	Poster presentation, European Society of Human Genetics conference. Best Poster
	Award candidate.
$\boldsymbol{2015}$	Scotland, Glasgow.
	Poster presentation, European Society of Human Genetics conference.
2014	Quebec, Canada, Montreal.
	Oral presentation, Human Genetics Dept. Research Day.
2014	British Columbia, Canada, Victoria.
	Poster presentation at the 3rd Annual Canadian Human and Statistical Genetics
	Meeting.
2014	NEW MEXICO, USA, Santa Fe.
	Poster presentation at the Mobile Genetic Elements and Genome Evolution Key-
	stone conference.
2013	Quebec, Canada, Esterel.
	Poster presentation at the 2nd Annual Canadian Human and Statistical Genetics
	Meeting.
2013	Montana, USA, Big Sky.
	Poster presentation at the Mobile DNA in Mammalian Genomes FASEB confer-
	${ m ence}.$
2011	Spain, Barcelona.
	Poster presentation at the XIIIth Spanish Biometry Conference and 3rd Ibero-

Other Activities

2019-present	University of California, Santa Cruz.
	Representative for the UCSC STEM Postdoc Association. Co-chair of the 2020
	USPA Symposium.
2018	IRCM, Montreal.
	Gave a short seminar about R Markdown at the Montreal Bioinformatics User
	Group.
2013-2018	McGill University.
	Organizer and instructor of eight full-day Introduction to R and Bioinformatics
	workshops for Human Genetics graduate students.

 ${\bf 2014\text{-}2017} \qquad \quad \text{McGill University}.$

American Biometry Meeting.

VP Communications at the Human Genetics Student Society.

2017 Université de Montreal.

Participated to a Bio-Hackaton. Our 24h project was to detect extending filaments

from 3D cell imaging.

2017 IRCM, MONTREAL.

Gave a short seminar about the Gviz package at the Montreal Bioinformatics User

Group.

2016 Kyoto, Japan.

Instructor at the 4th Kyoto Course and Symposium on Bioinformatics for Next

Generation Sequencing with Applications in Human Genetics.

2015 Université de Montreal.

Participated to a Bio-Hackaton. Our 24h project was to develop interactive visu-

alization of gene expression in cancer samples.

2014 McGill University.

Teaching Assistant for the Advanced Statistical Concepts in Genetic and Genomic

Analysis course.

2014 McGill University.

Organizer of a four-days R and Bioinformatics workshop for System Biology grad-

uate students.