INSERM IRSD U1220, CHU PURPAN

31024 Toulouse, FRANCE Email: jean.monlong@inserm.fr Webpage: https://jmonlong.github.io/

Education

2012 - 2018McGill University, Montreal, Canada.

PhD in the Human Genetics Department in Guillaume Bourque's Lab.

2010 - 2011Universitat Politècnica de Catalunya, Barcelona, Spain.

> Master in Statistic and Operations Research as an exchange student in the Faculty of Mathematics and Statistics.

2008 - 2010Ensimag, Grenoble, France.

> Computer Science and Mathematics course with specialization in Bioinformatics.

2006 - 2008LYCÉE MICHEL MONTAIGNE, Bordeaux, France.

Preparatory classes for entrance to the Grandes Écoles. Mathematics and Physics.

Professional Experiences and Projects

2023 -Institut de Recherche en Santé Digestive, Toulouse, France.

Researcher CRCN at INSERM. Genomics research on pangenomes and structural

2018 - 2023University of California Santa Cruz, Santa Cruz, USA.

> Postdoctoral researcher with Benedict Paten in the Computational Genomics Lab, analyzing structural variation with pangenomes.

2012 - 2018Bourque Lab, McGill Genome Centre, Montreal, Canada.

> PhD project. Implementation of a CNV detection method for whole-genome sequencing 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 - 2012CENTER 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 Geu-

> vadis and GTEx projects. 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. Col-

laboration with La Tronche's hospital (Grenoble).

Expertise

2011

R, Java, C, Bash, Python, WDL, Snakemake, AWK, Perl, D3/Javascript/HTML/CSS.

Git, Linux, High Performance Computing, LaTeX, R/Markdown, Emacs.

Genomics, Structural Variation, Pangenomics, Transcriptomics, Bioinformatics.

Scholarships and 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.
$\boldsymbol{2017}$	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.
$\boldsymbol{2015}$	Excellence Award. Human Genetics Dept, McGill University.
$\boldsymbol{2014}$	Best oral presentation. Human Genetics Dept's Research Day, McGill University.

Publications

J Siren, P Eskandar, MT Ungaro, G Hickey, JM Eizenga, AM Novak, X Chang, PC Chang, M Kolmogorov, A Carroll, **J Monlong**, and B Paten. Personalized pangenome references. *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. *Nature Methods*, 2023.

2023

G Hickey*, **J Monlong***, J Ebler, AM Novak, JM Eizenga, Y Gao, Human Pangenome Reference Consortium, H Li, and B Paten. Pangenome graph construction from genome alignments with Minigraph-Cactus. *Nature Biotechnology*, 2023.

2023

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, T Wang, ED Jarvis, KH Miga, E Garrison, T Marschall, IM Hall, H Li, and B Paten. A draft human pangenome reference. *Nature*, 2023.

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.

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, and Structural Variants in Distal Neural Stem Cells. *Neuro-Oncology*, 2022.

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. *Circulation: Genomic and Precision Medicine*, 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 Sequencing in a Critical Care Setting. *New England Journal of Medicine*, 2022.

2021

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.

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.

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.

2020

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.

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.

2019

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.

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

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.

from whole genome sequencing. PLOS Genetics, 2018.

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 tissue-specific regulation of transcription and splicing. *Genome research*, 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.

X Li, et al., and **The GTEx Consortium**. The impact of rare variation on gene expression across tissues. *Nature*, 2017.

MH Tan, et al., and **The GTEx Consortium**. Dynamic landscape and regulation of RNA editing in mammals. *Nature*, 2017.

T Tukiainen, et al., and **The GTEx Consortium**. Landscape of X chromosome inactivation across human tissues. *Nature*, 2017.

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.

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, 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.

2018

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J Monlong, M Calvo, PG Ferreira, and R Guigó. Identification of genetic variants associated with alternative splicing using sQTLseekeR. *Nature Communications*,

2014.

2013

PG 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.

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.

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.

PC t Hoen, et al., and **The Geuvadis Consortium**. Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. *Nature biotechnology*, 2013.

2013
The GTEx Consortium. The Genotype-Tissue Expression (GTEx) project. Nat Genet, 2013.

A Seigneurin, O Francois, 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. *BMJ*, 2011.

Selected Scientific Presentations

2020	NY, USA, New York City.
	Oral presentation, GSP-TOPMed Analysis Workshop.
2019	NY, USA, Cold Spring Harbor Laboratory.
	Oral presentation, Genome Informatics conference.
2017	Quebec, Canada, Quebec.
	Oral presentation, 6th Annual Canadian Human and Statistical Genetics Meeting.
2016	Japan, Kyoto.
	Oral presentation, 13th International Congress of Human Genetics.
$\boldsymbol{2015}$	BC, Canada, Vancouver.
	Oral presentation, 4rd Annual Canadian Human and Statistical Genetics Meeting.
$\boldsymbol{2014}$	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.
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
201.1	Meeting.
2014	NEW MEXICO, USA, Santa Fe.
	Poster presentation at the Mobile Genetic Elements and Genome Evolution Key-
0010	stone conference.
2013	QUEBEC, CANADA, Esterel.
	Poster presentation at the 2nd Annual Canadian Human and Statistical Genetics
2019	Meeting.
2013	MONTANA, USA, Big Sky. Poster presentation at the Makile DNA in Mammalian Consense FASER conformation.
	Poster presentation at the Mobile DNA in Mammalian Genomes FASEB confer-
2011	ence.
2011	Spain, Barcelona. Poster presentation at the XIIIth Spanish Biometry Conference and 3rd Ibero-
	1 Oster presentation at the Allin Spanish Diometry Conjerence and Sta Deto-

Other Activities

2019-	University of California, Santa Cruz.
${f present}$	Representative for the UCSC STEM Postdoc Association. Co-chair of the 2020
	USPA Symposium.

American Biometry Meeting.

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.

2014-2017 McGill University.

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 visual-

ization of gene expression in cancer samples.

2014 McGill University.

 ${\it 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.