

Jean MONLONG  
INSERM IRSD U1220, CHU PURPAN  
31024 Toulouse, FRANCE  
Email: [jean.monlong@inserm.fr](mailto:jean.monlong@inserm.fr)  
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 **Bioinformatics**.
- 2006 – 2008** [LYCÉE 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 variants.
- 2018 – 2023** [UNIVERSITY OF CALIFORNIA SANTA CRUZ](#), Santa Cruz, USA.  
Postdoctoral researcher with Benedict Paten in the [Computational Genomics Lab](#), analyzing structural variation with pangenomes.
- 2012 – 2018** [BOURQUE 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 – 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

---

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

---

<b>2019-2020</b>	<a href="#">BioData Catalyst Fellow</a> . National Heart, Lung, and Blood Institute, USA.
<b>2018</b>	Department Activity Award. Human Genetics Dept, McGill University.
<b>2017</b>	Student Exchange Support Program. JASSO, Japan.
<b>2017</b>	Graduate Mobility Award. Graduate and Postdoctoral Studies, McGill University.
<b>2017</b>	Oral Presentation, Honorable Mention. Human Genetics Dept's Research Day, McGill University.
<b>2017</b>	Excellence Award. Human Genetics Dept, McGill University.
<b>2013-2016</b>	Graduate Research Enhancement and Travel Award funding attendance to five international conferences. Human Genetics Dept, McGill University.
<b>2015</b>	Excellence Award. Human Genetics Dept, McGill University.
<b>2014</b>	Best oral presentation. Human Genetics Dept's Research Day, McGill University.

## Publications

---

<b>2023</b>	M Kolmogorov, KJ Billingsley, M Mastoras, M Meredith, <b>J Monlong</b> , ..., C Blauwendraat, and B Paten. Scalable Nanopore sequencing of human genomes provides a comprehensive view of haplotype-resolved variation and methylation. <a href="#">Nature Methods</a> , 2023.
<b>2023</b>	G Hickey*, <b>J Monlong*</b> , 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. <a href="#">Nature Biotechnology</a> , 2023.
<b>2023</b>	W Liao, M Asri, J Ebler, D Doerr, M Haukness, G Hickey, S Lu, JK Lucas, <b>J Monlong</b> , ..., 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. <a href="#">Nature</a> , 2023.
<b>2023</b>	R Lorig-Roach, M Meredith, <b>J Monlong</b> , ..., P Carnevali, K Miga, and B Paten. Phased nanopore assembly with shasta and modular graph phasing with gfase. <a href="#">bioRxiv</a> , 2023.
<b>2022</b>	CP Couturier, J Nadaf, Z Li, S Baig, G Riva, P Le, DJ Kloosterman, <b>J Monlong</b> , 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. <a href="#">Neuro-Oncology</a> , 2022.
<b>2022</b>	SD Goenka, JE Gorzynski, K Shafin, DG Fisk, T Pesout, TD Jensen, <b>J Monlong</b> , P Chang, G Baid, ..., B Paten, and EA Ashley. Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing. <a href="#">Nature Biotechnology</a> , 2022.
<b>2022</b>	JE Gorzynski, SD Goenka, K Shafin, TD Jensen, DG Fisk, ME Grove, E Spiteri, T Pesout, <b>J Monlong</b> , JA Bernstein, ..., B Paten, and EA Ashley. Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock. <a href="#">Circulation: Genomic and Precision Medicine</a> , 2022.
<b>2022</b>	JE Gorzynski, SD Goenka, K Shafin, TD Jensen, DG Fisk, ME Grove, E Spiteri, T Pesout, <b>J Monlong</b> , ..., B Paten, and EA Ashley. Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting. <a href="#">New England Journal of Medicine</a> , 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 Domezq, **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 Ayyadury, 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.
- 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.
- 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.

- 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.
- 2017 **The GTEx Consortium**. Genetic effects on gene expression across human tissues. *Nature*, 2017.
- 2017 A Saha, et al., and **The GTEx Consortium**. Co-expression networks reveal the tissue-specific regulation of transcription and splicing. *Genome research*, 2017.
- 2017 F Yang, et al., and **The GTEx Consortium**. Identifying cis-mediators for trans-eQTLs 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.
- 2017 MH 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.
- 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.
- 2014 **J Monlong**, M Calvo, PG Ferreira, and R Guigó. Identification of genetic variants associated with alternative splicing using sQTLseeker. *Nature Communications*, 2014.
- 2014 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.
- 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 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.
- 2011 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*.
- 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.
- 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* Keystone 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 conference.
- 2011** SPAIN, Barcelona.  
**Poster** presentation at the *XIIIth Spanish Biometry Conference and 3rd Ibero-American Biometry Meeting*.

## 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.
- 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 visualization 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 graduate students.