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Ph.D. in Complex Systems

Personal information

Name Alex Humberto Di Genova Bravo.
Nationality Chilean.
Position Postdoctoral researcher.

Academic degrees

2003–2007 **Bioinformatics Engineering**, University of Talca, Chile, *Engineer.*

Undergraduate Thesis

Title *Structural study of K⁺ channels sensed by pH TASK-2 and TASK-3.*
Tutor Fernando Gonzalez-Nilo.

2013–2017 **PhD in Complex Systems**, University Adolfo Ibáñez, Chile, *Conicyt scholarship; PhD defense 14 November 2017.*

PhD Thesis

Title *Assembly of complex genomes: Algorithms and Applications.*
Tutor Alejandro Maass, Gonzalo Ruz and Marie-France Sagot.

Research experience

2008–2011 **Research engineer**, Mathomics, University of Chile, Center for Mathematical Modeling (CMM) , Santiago, Chile.

2011–2015 **Leader of genomic unit**, Mathomics, University of Chile, CMM, Santiago, Chile.

Participation in the following projects:

- SALMONID GENOMICS: IDENTIFICATION OF GENES RELATED TO PROTEIN AND VEGETAL OIL USAGE IN ATLANTIC SALMON AND RAINBOW TROUT NUTRITION, University of Chile, Corfo-INNOVA-07CN13PBT-41 (2008 - 2011).
- INFORMATION BIOMINING LABORATORY: ORGANIZATION AND STATISTICAL PROCESSING OF BIO-MINING DATA, GENOME ANNOTATION AND PLATFORM DEVELOPMENT, Funded by BioSigma S.A. (2009 - 2010)
- IDENTIFICATION OF GENES RELATED TO BERRY DEVELOPMENT AND GROWTH IN SEEDLESS TABLE GRAPES BY MEANS OF FUNCTIONAL GENOMICS, Instituto de Investigaciones Agropecuarias. (2009 - 2012).
- POTATO GENOME SEQUENCING CONSORTIUM (PGSC) (2009 - 2011).

- INTERNATIONAL ATLANTIC SALMON SEQUENCING CONSORTIUM, CHILE, CANADA AND NORWAY (2010 - 2016).
- CENTER OF GENOME REGULATION, University of Chile, Fonad-Conicyt (2011 - 2016).
- COMMUNICATION AND INFORMATION RESEARCH AND INNOVATION CENTER (CIRIC-INRIA), University of Chile, Corfo (2012 - 2012).
- THE CHILEGENOMICO PROJECT, University of Chile, Fondef (2012 - 2016).

2017–2019 **Postdoctoral researcher**, *Alignment-free algorithms for genome assembly.*, BAOBAB/LBBE team directed by Marie-France Sagot.
INRIA, Lyon, France

2020–present **Postdoctoral researcher**, *Understanding the role of structural variants in rare cancers genomes.*, Rare Cancers Genomics team directed by Lynnette Fernandez-Cuesta and Matthieu Foll.
IARC, Lyon, France

Languages

Spanish	Native speaker.
English	Proficient in both spoken and written English.
French	Proficient in spoken French.

Teaching

Course Bioinformatic *Four years teacher in Master of Medical informatics, Faculty of Medicine, University of Chile (2011-2014).*

Scientific programming

Programming languages	C++, PERL, BioPerl, BASH, R, SQL, GIT, Latex, HTML	High Performance Computing	Expert user/admin of Linux clusters and a proficient coder of concurrent software on shared memory systems.
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Software

FAST-SG	FAST-SG is an alignment-free algorithm for ultrafast scaffolding graph construction from short or long reads.	WENGAN	WENGAN is an accurate and ultra-fast hybrid genome assembler. WENGAN generates high quality human genome reconstructions using less than one day of computing time on an average server (20 cores, \leq 50Gb RAM).
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Research Interests

Algorithms, genome assembly, computational analysis of next generation sequencing data, and cancer genomics.

Publications

Selected publications

Di Genova, A., Buena-Atienza, E., Ossowski, S. & Sagot, M.-F. **Efficient hybrid de novo assembly of human genomes with WENGAN.** *Nature Biotechnology*, 2020.

Lien, S., Koop, B. F., Sandve, S. R., Miller, J. R., Kent, M. P., Nome, T., Hvidsten, T. R., Leong, J. S., Minkley, D. R., Zimin, A., Grammes, F., Grove, H., Gjuvsland, A., Walenz, B., Hermansen, R. A., von Schalburg, K., Rondeau, E. B., Di Genova, A., Samy, J. K., Olav Vik, J., Vigeland, M. D., Caler, L., Grinholt, U., Jentoft, S., Inge Vage, D., de Jong, P., Moen, T., Baranski, M., Palti, Y., Smith, D. R., et al. **The Atlantic salmon genome provides insights into rediploidization.** *Nature*, 2016.

Xu, X., Pan, S., Cheng, S., Zhang, B., Mu, D., Ni, P., Zhang, G., Yang, S., Li, R., Wang, J., Orjeda, G., Guzman, F., Torres, M., Lozano, R., Ponce, O., Martinez, D., De la Cruz, G., Chakrabarti, S. K., Patil, V. U., Skryabin, K. G., Kuznetsov, B. B., Ravin, N. V., Kolganova, T. V., Beletsky, A. V., Mardanov, A. V., Di Genova, A., Bolser, D. M., Martin, D. M., Li, G., Yang, Y., et al. **Genome sequence and analysis of the tuber crop potato.** *Nature*, 2011.

Di Genova, A., Ruz, G. A., Sagot, M.-F. & Maass, A. **Fast-SG: an alignment-free algorithm for hybrid assembly.** *GigaScience*, 2018.

Narum, S. R., Di Genova, A., Micheletti, S. J. & Maass, A. **Genomic variation underlying complex life-history traits revealed by genome sequencing in Chinook salmon.** *Proceedings of the Royal Society B: Biological Sciences*, 2018.

Di Genova, A., Almeida, A. M., Muñoz-Espinoza, C., Vizoso, P., Travisany, D., Moraga, C., Pinto, M., Hinrichsen, P., Orellana, A. & Maass, A. **Whole genome comparison between table and wine grapes reveals a comprehensive catalog of structural variants.** *BMC plant biology*, 2014.

Papers in Refereed Journals (WoS)

- 2020 32 Di Genova, A., Buena-Atienza, E., Ossowski, S. & Sagot, M.-F. **Efficient hybrid de novo assembly of human genomes with WENGAN.** *Nature Biotechnology*, 1–9.
- 31 Mucha, S. G., Ferrarini, M. G., Moraga, C., Di Genova, A., Guyon, L., Tardy, F., Rome, S., Sagot, M.-F. & Zaha, A. **Mycoplasma hyopneumoniae J elicits an antioxidant response and decreases the expression of ciliary genes in infected swine epithelial cells.** *Scientific Reports*, 13707.
- 30 Muñoz-Espinoza, C., Di Genova, A., Sánchez, A., Correa, J., Espinoza, A., Meneses, C., Maass, A., Orellana, A. & Hinrichsen, P. **Identification of SNPs and InDels associated with berry size in table grapes integrating genetic and transcriptomic approaches.** *BMC Plant Biology* **20**, 1–21.
- 29 Rezende, J. C., Di Genova, A., Nahat, R. A., Steinbüchel, A., Sagot, M.-F., Costa, R. S., Oliveira, H. C., Taciro, M. K., Silva, L. F. & Gomez, J. G. C. **The relevance of enzyme specificity for coenzymes and the presence of 6-phosphogluconate dehydrogenase for polyhydroxyalkanoates production in the metabolism of Pseudomonas sp. LFM046.** *International Journal of Biological Macromolecules*.

- 28 Verdugo, R. A., Di Genova, A., Herrera, L., Moraga, M., Acuña, M., Berrios, S., Llop, E., Valenzuela, C. Y., Bustamante, M. L., Digman, D., *et al.* Development of a small panel of SNPs to infer ancestry in Chileans that distinguishes Aymara and Mapuche components. *Biological research* **53**, 1–11.
- 2019**
- 27 Travisany, D., Ayala-Raso, A., Di Genova, A., Monsalve, L., Bernales, M., Martinez, J. P., González-Agüero, M., Defilippi, B., Cherian, S., Maass, A., *et al.* RNA-Seq analysis and transcriptome assembly of raspberry fruit (*Rubus idaeus* "Heritage") revealed several candidate genes involved in fruit development and ripening. *Scientia Horticulturae* **254**, 26–34.
- 26 Vidal, E. A., Moyano, T. C., Bustos, B. I., Perez-Palma, E., Moraga, C., Riveras, E., Montecinos, A., Azocar, L., Soto, D. C., Vidal, M., Di Genova, A., Puschel, K., Nurnberg, P., Buch, S., Hampe, J., Allende, M. L., Cambiazo, V., Gonzalez, M., Hodar, C., Montecino, M., Munoz-Espinoza, C., Orellana, A., Reyes-Jara, A., Travisany, D., Vizoso, P., Moraga, M., Eyheramendy, S., Maass, A., De Ferrari, G. V., Miquel, J. F. & Gutierrez, R. A. Whole Genome Sequence, Variant Discovery and Annotation in Mapuche-Huilliche Native South Americans. *Sci Rep* **9**, 2132.
- 2018**
- 25 Di Genova, A., Mardones, W., Cortés, M. P., Travisany, D., Maass, A. & Eyzaguirre, J. The genome sequence of the softrot fungus *Penicillium purpurogenum* reveals a high gene dosage for lignocellulolytic enzymes. *Mycology* **9**, 59–69.
 - 24 Di Genova, A., Ruz, G. A., Sagot, M.-F. & Maass, A. Fast-SG: an alignment-free algorithm for hybrid assembly. *GigaScience* **7**, giy048.
 - 23 López, M. E., Benestan, L., Moore, J.-S., Perrier, C., Gilbey, J., Di Genova, A., Maass, A., Diaz, D., Lhorente, J. P., Correa, K., Neira, R., Bernatchez, L. & Yáñez, J. M. Comparing genomic signatures of domestication in two Atlantic salmon (*Salmo salar* L.) populations with different geographical origins. *Evolutionary Applications*.
 - 22 Narum, S. R., Di Genova, A., Micheletti, S. J. & Maass, A. Genomic variation underlying complex life-history traits revealed by genome sequencing in Chinook salmon. *Proc. R. Soc. B* **285**, 20180935.
- 2017**
- 21 Loira, N., Mendoza, S., Cortés, M. P., Rojas, N., Travisany, D., Di Genova, A., Gajardo, N., Ehrenfeld, N. & Maass, A. Reconstruction of the microalga *Nannochloropsis salina* genome-scale metabolic model with applications to lipid production. *BMC Systems Biology* **11**, 66.
 - 20 Pastenes, L., Valdivieso, C., Di Genova, A., Travisany, D., Hart, A., Montecino, M., Orellana, A., Gonzalez, M., Gutiérrez, R. A., Allende, M. L., *et al.* Global gene expression analysis provides insight into local adaptation to geothermal streams in tadpoles of the Andean toad *Rhinella spinulosa*. *Scientific Reports* **7**.
- 2016**
- 19 Correa, K., Lhorente, J. P., Bassini, L., López, M. E., Di Genova, A., Maass, A., Davidson, W. S. & Yáñez, J. M. Genome wide association study for resistance to *Caligus rogercresseyi* in Atlantic salmon (*Salmo salar* L.) using a 50K SNP genotyping array. *Aquaculture*.

- 18 Latorre, M., Cortes, M. P., Travisany, D., Di Genova, A., Budinich, M., Reyes-Jara, A., Hodar, C., Gonzalez, M., Parada, P., Bobadilla-Fazzini, R. A., Cambiazo, V. & Maass, A. **The bioleaching potential of a bacterial consortium.** *Bioresource Technology* **218**, 659–666.
- 17 Lien, S., Koop, B. F., Sandve, S. R., Miller, J. R., Kent, M. P., Nome, T., Hvidsten, T. R., Leong, J. S., Minkley, D. R., Zimin, A., Grammes, F., Grove, H., Gjuvsland, A., Walenz, B., Hermansen, R. A., von Schalburg, K., Rondeau, E. B., Di Genova, A., Samy, J. K., Olav Vik, J., Vigeland, M. D., Caler, L., Grimholt, U., Jentoft, S., Inge Vage, D., de Jong, P., Moen, T., Baranski, M., Palti, Y., Smith, D. R., *et al.* **The Atlantic salmon genome provides insights into rediploidization.** *Nature* **533**, 200–205.
- 16 Muñoz-Espinoza, C., Di Genova, A., Correa, J., Silva, R., Maass, A., González-Agüero, M., Orellana, A. & Hinrichsen, P. **Transcriptome profiling of grapevine seedless segregants during berry development reveals candidate genes associated with berry weight.** *BMC plant biology* **16**, 104.
- 15 Yáñez, J. M., Naswa, S., López, M., Bassini, L., Correa, K., Gilbey, J., Bernatchez, L., Norris, A., Neira, R., Lhorente, J., Di Genova, A. & Maass, A. **Genomewide single nucleotide polymorphism discovery in Atlantic salmon (*Salmo salar*): validation in wild and farmed American and European populations.** *Molecular ecology resources* **16**, 1002–1011.
- 2015**
- 14 Arismendi, M. J., Almada, R., Pimentel, P., Bastias, A., Salvatierra, A., Rojas, P., Hinrichsen, P., Pinto, M., Di Genova, A., Travisany, D., *et al.* **Transcriptome sequencing of *Prunus* sp. rootstocks roots to identify candidate genes involved in the response to root hypoxia.** *Tree Genetics & Genomes* **11**, 1–16.
- 13 Correa, K., Lhorente, J. P., López, M. E., Bassini, L., Naswa, S., Deeb, N., Di Genova, A., Maass, A., Davidson, W. S. & Yáñez, J. M. **Genome-wide association analysis reveals loci associated with resistance against *Piscirickettsia salmonis* in two Atlantic salmon (*Salmo salar* L.) chromosomes.** *BMC genomics* **16**, 1.
- 12 Smedley, D., Haider, S., Durinck, S., Pandini, L., Provero, P., Allen, J., Arnaiz, O., Awedh, M. H., Baldock, R., Barbiera, G., Bardou, P., Beck, T., Blake, A., Bonierbale, M., Brookes, A. J., Bucci, G., Buetti, I., Burge, S., Cabau, C., Carlson, J. W., Chelala, C., Chrysostomou, C., Cittaro, D., Collin, O., Cordova, R., Cutts, R. J., Dassi, E., Di Genova, A., Djari, A., Esposito, A., *et al.* **The BioMart community portal: an innovative alternative to large, centralized data repositories.** *Nucleic Acids Res.* **43**, W589–598.
- 2014**
- 11 Di Genova, A., Almeida, A. M., Muñoz-Espinoza, C., Vizoso, P., Travisany, D., Moraga, C., Pinto, M., Hinrichsen, P., Orellana, A. & Maass, A. **Whole genome comparison between table and wine grapes reveals a comprehensive catalog of structural variants.** *BMC plant biology* **14**, 1.
- 10 Travisany, D., Cortés, M. P., Latorre, M., Di Genova, A., Budinich, M., Bobadilla-Fazzini, R. A., Parada, P., González, M. & Maass, A. **A new genome of *Acidithiobacillus thiooxidans* provides insights into adaptation to a bi-oleaching environment.** *Research in microbiology* **165**, 743–752.

- 2013**
- 9 Bórquez, D. A., Olmos, C., Álvarez, S., Di Genova, A., Maass, A. & González-Billault, C. **Bioinformatic survey for new physiological substrates of Cyclin-dependent kinase 5.** *Genomics* **101**, 221–228.
 - 8 González-Agüero, M., García-Rojas, M., Di Genova, A., Correa, J., Maass, A., Orellana, A. & Hinrichsen, P. **Identification of two putative reference genes from grapevine suitable for gene expression analysis in berry and related tissues derived from RNA-Seq data.** *BMC genomics* **14**, 1.
 - 7 Sharma, S. K., Bolser, D., de Boer, J., Sanderker, M., Amoros, W., Carboni, M. F., D'Ambrosio, J. M., de la Cruz, G., Di Genova, A., Douches, D. S., Eguiluz, M., Guo, X., Guzman, F., Hackett, C. A., Hamilton, J. P., Li, G., Li, Y., Lozano, R., Maass, A., Marshall, D., Martinez, D., McLean, K., Mejia, N., Milne, L., Munive, S., Nagy, I., Ponce, O., Ramirez, M., Simon, R., Thomson, S. J., *et al.* **Construction of reference chromosome-scale pseudomolecules for potato: integrating the potato genome with genetic and physical maps.** *G3 (Bethesda)* **3**, 2031–2047.
 - 6 Utreras, E., Henriquez, D., Contreras-Vallejos, E., Olmos, C., Di Genova, A., Maass, A., Kulkarni, A. B. & Gonzalez-Billault, C. **Cdk5 regulates Rap1 activity.** *Neurochemistry international* **62**, 848–853.
- 2012**
- 5 Hödar, C., Moreno, P., Di Genova, A., Latorre, M., Reyes-Jara, A., Maass, A., González, M. & Cambiazo, V. **Genome wide identification of Acidithiobacillus ferrooxidans (ATCC 23270) transcription factors and comparative analysis of ArsR and MerR metal regulators.** *BioMetals* **25**, 75–93.
 - 4 Travisany, D., Di Genova, A., Sepúlveda, A., Bobadilla-Fazzini, R. A., Parada, P. & Maass, A. **Draft genome sequence of the Sulfovibacillus thermosulfidooxidans Cutipay strain, an indigenous bacterium isolated from a naturally extreme mining environment in Northern Chile.** *Journal of bacteriology* **194**, 6327–6328.
- 2011**
- 3 Di Genova, A., Aravena, A., Zapata, L., Gonzalez, M., Maass, A. & Iturra, P. **SalmonDB: a bioinformatics resource for *Salmo salar* and *Oncorhynchus mykiss*.** *Database* **2011**, bar050.
 - 2 Guberman, J. M., Ai, J., Arnaiz, O., Baran, J., Blake, A., Baldock, R., Chelala, C., Croft, D., Cros, A., Cutts, R. J., Di Genova, A., Forbes, S., Fujisawa, T., Gadaleta, E., Goodstein, D. M., Gundem, G., Haggarty, B., Haider, S., Hall, M., Harris, T., Haw, R., Hu, S., Hubbard, S., Hsu, J., Iyer, V., Jones, P., Katayama, T., Kinsella, R., Kong, L., Lawson, D., *et al.* **BioMart Central Portal: an open database network for the biological community.** *Database (Oxford)* **2011**, bar041.
 - 1 Xu, X., Pan, S., Cheng, S., Zhang, B., Mu, D., Ni, P., Zhang, G., Yang, S., Li, R., Wang, J., Orjeda, G., Guzman, F., Torres, M., Lozano, R., Ponce, O., Martinez, D., De la Cruz, G., Chakrabarti, S. K., Patil, V. U., Skryabin, K. G., Kuznetsov, B. B., Ravin, N. V., Kolganova, T. V., Beletsky, A. V., Mardanov, A. V., Di Genova, A., Bolser, D. M., Martin, D. M., Li, G., Yang, Y., *et al.* **Genome sequence and analysis of the tuber crop potato.** *Nature* **475**, 189–195.

Additional Information

My publication record is available in GoogleScolar and ResearchGate.