

## BORIS E. REBOLLEDO JARAMILLO

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### Summary

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Researcher experienced in the integrative analysis of large genomic datasets. Strong background in molecular biology and statistics. Proficient in Python, R, and Shell scripting. Currently, focused on clinical genomics. Particularly, the clinical consequences of variants affecting the mitochondrial-nuclear genetic coordination.

### Education

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| 2016 | <b>Ph.D. in Bioinformatics and Genomics.</b><br>Pennsylvania State University, University Park, PA, USA.     |
| 2012 | <b>MSc. In Biochemistry and Bioinformatics.</b><br>Universidad de Concepción, Concepción, Chile.             |
| 2009 | <b>Bioengineering, minor in Molecular and Cell Biology.</b><br>Universidad de Concepción, Concepción, Chile. |

### Work experience

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| 2016 - present   | <b>Assistant Professor and Bioinformatics Core coordinator</b><br>Universidad del Desarrollo, Santiago, Chile. |
| 2016 (May - Jul) | <b>Research assistant.</b><br>Pennsylvania State University, University Park, PA, USA.                         |
| 2010 - 2016      | <b>Graduate research assistant.</b><br>Pennsylvania State University, University Park, PA, USA.                |
| 2008 - 2010      | <b>M.Sc. candidate.</b><br>Universidad de Concepción, Concepción, Chile.                                       |

### Teaching experience

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| 2017/2018 | <b>Topics in Computational Research for Genetics and Genomics.</b><br>Tutor. ICBM, Faculty of Medicine.<br>Universidad de Chile, Santiago, Chile.                       |
| 2017/2018 | <b>Advanced topics in Bioinformatics and Biostatistics.</b><br>Instructor. Ph.D. in Science and Innovation in Medicine.<br>Universidad del Desarrollo, Santiago, Chile. |
| 2017      | <b>Biostatistics.</b><br>Instructor. Ph.D. in Science and Innovation in Medicine.<br>Universidad del Desarrollo, Santiago, Chile.                                       |

### Projects

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| 2017 - 2020 | <b>FONDECYT (postdoctoral) N°3170280.</b><br>Contribution of mitochondrial DNA heteroplasmy to the phenotype of patients with maternally transmitted 22q11.2 deletion syndrome. |
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## Peer reviewed publications (\* equal contribution)

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1. Farkas C, Fuentes-Villalobos F, **Rebolledo-Jaramillo B.**, Benavides F, Castro AF, and Pincheira R. (2019). Streamlined computational pipeline for genetic background characterization of genetically engineered mice based on next generation sequencing data. *BMC genomics*, 20(1), 131. doi:10.1186/s12864-019-5504-9.
2. Schwieger-Briel A, Fuentes I, Castiglia D, Barbato A, Greutmann M, Leppert J, Duchatelet S, Hovnanian A, Burattini S, Yubero MJ, Ibañez-Arenas R, **Rebolledo-Jaramillo B.**, Gräni C, Ott H, Theiler M, Weibel L, Paller AS, Zambruno G, Fischer J, Palisson F, Has C.(2019). Epidermolysis bullosa simplex with KLHL24 mutations is associated with dilated cardiomyopathy. *J. Invest. Dermatol.* 139(1):244-249. doi: 10.1016/j.jid.2018.07.022.
3. **Rebolledo-Jaramillo B.**, Ziegler A. (2018). Teneurins: An integrative molecular, functional and biomedical overview of their role in cancer. *Front. Neurosci.*, 12:937. <https://doi.org/10.3389/fnins.2018.00937>.
4. Gruning BA, Rasche E, **Rebolledo-Jaramillo B.**, Eberhard C, Houwaart T, Chilton J, Coraor N, Backofen R, Taylor J, Nekrutenko Anton. (2017). Jupyter and Galaxy: Easing entry barriers into complex data analyses for biomedical researchers. *PLoS Computational Biology*. 13(5):e1005425.
5. Hasbún R, Iturra C, Bravo S, **Rebolledo-Jaramillo B.**, Valledor L. (2016). Differential Methylation of Genomic Regions Associated with Heteroblasty Detected by M&M Algorithm in the Nonmodel Species *Eucalyptus globulus* Labill. *Int. J. Genomics*, vol. 2016, 4395153. doi:10.1155/2016/4395153.
6. **Rebolledo-Jaramillo B\***, Su MS\*, McElhoe J, Stoller N, Dickins B, Korneliussen T, Nielsen R, Holland M, Paul I, Nekrutenko A, Makova KD. (2014) Maternal Age Effect and Severe Germline Bottleneck in the Inheritance of Human Mitochondrial DNA. *PNAS* 111(43):15474–15479.
7. **Rebolledo-Jaramillo B.**, Alarcon RA, Fernandez VI, Gutierrez SE. (2014). Cis-regulatory elements are harbored in Intron5 of the RUNX1 gene. *BMC Genomics* 15:225.
8. Dickins B\*, **Rebolledo-Jaramillo B\***, Shu-Wei S, Paul IM, Blankenberg D, Stoler N, Makova KD, Nekrutenko A. (2014). Controlling for contamination in resequencing studies with a reproducible web-based phylogenetic approach. *BioTechniques*, 56(3):134–141.
9. Zheng R, **Rebolledo-Jaramillo B.**, Zong Y, Wang L, Russo P, Hancock W, Stanger B, Hardison R, Blobel G (2013). Function of GATA factors in the adult mouse liver. *PLoS One*. 18;8(12):e83723.
10. Bar-Yaacov D, Avital G, Levin L, Richards A, Hachen N, **Rebolledo-Jaramillo B.**, Nekrutenko A, Zarivach R, Mishmar D. (2013). RNA-DNA differences in human mitochondria restore ancestral form of 16S ribosomal RNA. *Genome Res.* 23(11):1789-96.

## Book chapters

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1. Gutierrez S, Javed A, Stein J, Stein G, Nicovani S, Fernandez V, Alarcon R, Stuardo M, Martinez M, Hinojosa M and **Rebolledo Jaramillo B** (2011). Epigenetic Changes Associated with Chromosomal Translocation in Leukemia, Myeloid Leukemia - Basic Mechanisms of Leukemogenesis, Steffen Koschmieder and Utz Krug (Ed.), ISBN: 978-953-307-789-5, InTech.

## Conference presentations

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Latest talks:

1. Symposia: *Paving the road towards collaborative genomic research in Chile*. **51° Annual meeting of the Chilean Genetics Society; 2018 Nov 20-22. Puerto Varas, Chile.**
2. *Mitochondrial Genomics in 22q deletion syndrome*. **50° Annual meeting of the Chilean Genetics Society; 2017 Nov 9-11. Puerto Varas, Chile.**
3. *Maternal Age Effect and Severe Germline Bottleneck in the Inheritance of Human Mitochondrial DNA*. **39° Annual meeting of the Chilean Society for Biochemistry and Molecular Biology; 2016 Sept 27-30. Puerto Varas, Chile.**

## References

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**Gabriela Repetto, M.D.** e-mail: [gretto@udd.cl](mailto:gretto@udd.cl)  
Professor. Center for Genetics and Genomics. Universidad del Desarrollo.  
**Anton Nekrutenko, Ph.D.** e-mail: [aun1@psu.edu](mailto:aun1@psu.edu)  
Professor of Biochemistry and Molecular Biology. The Pennsylvania State University.