BORIS E. REBOLLEDO-JARAMILLO

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Summary

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

2016 **Ph.D. in Bioinformatics and Genomics.**
Pennsylvania State University, University Park, PA, USA.

2012 **MSc. In Biochemistry and Bioinformatics.**
Universidad de Concepción, Concepción, Chile.

2009 **Bioengineering, minor in Molecular and Cell Biology.**
Universidad de Concepción, Concepción, Chile.

Work experience

2019 - present **Assistant Professor and Bioinformatics Core coordinator.**
Universidad del Desarrollo, Santiago, Chile.

2016 - 2019 **Postdoctoral research assistant.**
Universidad del Desarrollo, Santiago, Chile.

2010 - 2016 **Graduate research assistant.**
Pennsylvania State University, University Park, PA, USA.

2008 - 2010 **M.Sc. candidate.**
Universidad de Concepción, Concepción, Chile.

Teaching experience

2017 - present **Biostatistics.**
Instructor. Ph.D. in Science and Innovation in Medicine.
Universidad del Desarrollo, Santiago, Chile.

2017 - present **Advanced topics in Bioinformatics and Biostatistics.**
Instructor. Ph.D. in Science and Innovation in Medicine.
Universidad del Desarrollo, Santiago, Chile.

Funded research projects

2017 - 2020 **FONDECYT (postdoctoral) Nº3170280.**
Contribution of mitochondrial DNA heteroplasmy to the phenotype of patients with maternally transmitted 22q11.2 deletion syndrome.

Peer reviewed publications (* equal contribution)

1. Durán A*, **Rebolledo-Jaramillo B***, Olguín V, Rojas-Herrera M, Las Heras M, Calderón JF, Zanlungo S, Priestman D, Platt FM, Klein AD (2021). Identification of genetic modifiers of murine hepatic β-glucocerebrosidase activity. Biochem. Biophys. Rep. 28(101105). https://doi.org/10.1016/j.bbrep.2021.101105.
2. **Rebolledo-Jaramillo B,** Obregón MG, Huckstadt V, Gómez A, Repetto GM (2021). Contribution of Mitochondrial DNA Heteroplasmy to the Congenital Cardiac and Palatal Phenotypic Variability in Maternally Transmitted 22q11.2 Deletion Syndrome. Genes 12(1), 92; https://doi.org/10.3390/genes12010092.
3. Fuentes I, Guttmann-Gruber C, Tockner B, Diem A, Klausegger A, Cofré-Araneda G, Figuera O, Hidalgo Y, Morandé P, Palisson F, **Rebolledo-Jaramillo B**, Yubero MJ, Cho RJ, Rishel HI, Marinkovich MP, Teng J, Webster TG, Prisco M, Eraso LH, Piñon Hofbauer J, South, AP (2020). Cells from discarded dressings differentiate chronic from acute wounds in patients with Epidermolysis Bullosa. Scientific reports, 10(1), 15064. https://doi.org/10.1038/s41598-020-71794-1.
4. 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.
5. 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.
6. **Rebolledo-Jaramillo B**, Ziegler A. (2018). Teneurins: An integrative molecular, functional and biomedical overview of their role in cancer. Front. Neurosci., 11 December 2018 https://doi.org/10.3389/fnins.2018.00937.
7. 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.
8. 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.
9. **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.
10. **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.
11. 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.
12. 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.
13. 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

1. Repetto G and **Rebolledo Jaramillo B** (2019). Rare diseases: Genomics and public health - Applied Genomics And Public Health, George Patrinos (Ed.), ISBN: 9780128136966, Academic Press.
2. 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.

Latest presentations (latest talks)

1. *Contribution of Mitochondrial Genomics to the Congenital Cardiac and Palatal Phenotypic Variability in 22q11.2 Deletion Syndrome*. **Weekly Wartik Genomics Lecture Series; 2020 Nov 11. Pennsylvania State University, USA.**
2. *Bioinformagic in the study of rare diseases*. I**naugural address for 2020 MSc. in Biochemistry and Bioinformatics University of Concepción program, Chile.**
3. Symposia: P*aving the road towards collaborative genomic research in Chile*. **51º Annual meeting of the Chilean Genetics Society; 2018 Nov 20-22. Puerto Varas, Chile.**
4. *Mitochondrial Genomics in 22q deletion syndrome*. **50º Annual meeting of the Chilean Genetics Society; 2017 Nov 9-11. Puerto Varas, Chile.**
5. *Maternal Age Effect and Severe Germline Bottleneck in the Inheritance of Human Mitochondrial DNA*. **39º Annual meeting of the Chilean Society for Biochemistry and Molecular Biology; 2016 Sept 27-30. Puerto Varas, Chile.**

References

Gabriela Repetto, M.D. Universidad del Desarrollo, e-mail: grepetto@udd.cl
Anton Nekrutenko, Ph.D. Pennsylvania State University, e-mail: aun1@psu.edu