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 Universidad de Concepción, Concepción, Chile.

Work experience

Years	Position
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.

Teaching experience

Years	Position
2020 - present	Guest Lecturer and thesis reviewer. MSc. in Biochemistry and Bioinformatics, Universidad de Concepción, Concepción, Chile.
2017 - present	Biostatistics. Ph.D. in Science and Innovation in Medicine. Universidad del Desarrollo, Santiago, Chile.
2017 - present	Advanced topics in Bioinformatics and Biostatistics. Ph.D. in Science and Innovation in Medicine. Universidad del Desarrollo, Santiago, Chile.

Funding

Years	Project	Title
2022 - 2024	FONDECYT (Initiation in research) N°11220642.	Contribution of intergenerational mitonuclear mismatch to disease in Latin-American admixed patients.
2017 - 2020	FONDECYT (postdoctoral) N°3170280.	Contribution of mitochondrial DNA heteroplasmy to the phenotype of patients withmaternally transmitted 22q11.2 deletion syndrome.

Collaborations

Years	Project	Title
2022 - 2025	FONDECYT regular No1221802.	“Functional genomics in autoinflammatory diseases”. (PI: C. Poli)
2018 - 2021	FONDECYT regular No1171014.	“Prodromal manifestations of Parkinson’s disease in a high risk population: 22q11.2 midrodeletion syndrome”. (PI: G. Repetto)

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Peer reviewed publications (*Equal contribution)

1. Durán A, Priestman DA, Las Heras M, Rebolledo-Jaramillo B, Olguín V, Calderón JF, Zanlungo S, Gutiérrez J, Platt FM, Klein AD. A Mouse Systems Genetics Approach Reveals Common and Uncommon Genetic Modifiers of Hepatic Lysosomal Enzyme Activities and Glycosphingolipids. International Journal of Molecular Sciences. 2023; 24(5):4915. doi:10.3390/ijms24054915

2. Fuentes I, Yubero MJ, Morandé P, Varela C, Oróstica K, Acevedo F, Rebolledo-Jaramillo B, Arancibia E, Porte L, Palisson F. (2022) Longitudinal study of wound healing status and bacterial colonisation of Staphylococcus aureus and Corynebacterium diphtheriae in epidermolysis bullosa patients. Int Wound J. 2022; 1- 10. doi:10.1111/iwj.13922

3. Cárdenas GV, Iturriaga C, Hernández CD, Tejos-Bravo M, Pérez-Mateluna G, Cabalin C, Urzúa M, Venegas-Salas LF, Fraga JP, Rebolledo B, Poli MC, Repetto GM, Casanello P, Castro-Rodríguez JA, Borzutzky A. (2021) Prevalence of filaggrin loss-of-function variants in Chilean population with and without atopic dermatitis. Int J Dermatol. doi: 10.1111/ijd.15887

4. Balboa E, Marín T, Oyarzún JE, Contreras PS, Hardt R, van den Bosch T, Alvarez AR, Rebolledo-Jaramillo B, Klein AD, Winter D, Zanlungo S. (2021). Proteomic Analysis of Niemann-Pick Type C Hepatocytes Reveals Potential Therapeutic Targets for Liver Damage. Cells. 10(8):2159. doi: 10.3390/cells10082159

5. 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). doi:10.1016/j.bbrep.2021.101105

6. 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; doi:10.3390/genes12010092

7. 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. doi:10.1038/s41598-020-71794-1

8. 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

9. 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

10. Rebolledo-Jaramillo B, Ziegler A. (2018). Teneurins: An integrative molecular, functional and biomedical overview of their role in cancer. Front. Neurosci., 11 December 2018 doi:10.3389/fnins.2018.00937

11. 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. doi:10.1371/journal.pcbi.1005425

12. 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

13. 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. doi:10.1073/pnas.140932811

14. 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. doi:10.1186/1471-2164-15-225

15. 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 doi:10.2144/000114146

16. 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 doi:10.1371/journal.pone.0083723

17. 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 doi:10.1101/gr.161265.113