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› Summary

Director of the Bioinformatics Core Facility and lead bioinformatician for the Rare Diseases Program at Universidad del Desarrollo, Chile. With seven years of experience in the clinical genomics field, I bring expertise in research, teaching bioinformatics and biostatistics, and developing bioinformatics software.

› Academic degrees

Year	Degree	University
2016	Ph.D. in Bioinformatics and Genomics	Pennsylvania State University, USA
2012	M.Sc. in Biochemistry and Bioinformatics	University of Concepcion, Chile
2009	Bioengineering	University of Concepcion, Chile

› Work experience

Year	Position	Location
2019 – present	Assistant research professor	Universidad del Desarrollo, Chile
2016 – 2019	Postdoctoral researcher	Universidad del Desarrollo, Chile

› Grants

Year	Role	Grant number	Title
2022 – 2024	PI	FONDECYT 11220642	Contribution of intergenerational mitonuclear mismatch to disease in Latin-American admixed patients
2022 – 2025	Co-PI	FONDECYT 1221802	Functional genomics in autoinflammatory diseases
2018 – 2021	Co-PI	FONDECYT 1171014	Prodromal manifestations of Parkinson's disease in a high risk population: 22q11.2 microdeletion syndrome
2017 – 2019	PI	FONDECYT 3170280	Contribution of mitochondrial DNA heteroplasmy to the phenotype of patients with maternally transmitted 22q11.2 deletion syndrome

› Teaching experience

1. Graduate

Year	Course/Activity	Program	University
2017 – present	R for Biostatistics	Ph.D. in Science and Innovation in Medicine	Universidad del Desarrollo, Chile

Year	Course/Activity	Program	University
2017 – present	Bioinformatics	Ph.D. in Science and Innovation in Medicine	Universidad del Desarrollo, Chile
2022 – 2024	Thesis co-advisor	M.Sc. in Neuroscience	Universidad de Chile, Chile
2023	Thesis external reviewer	M.Sc. in Biochemistry and Bioinformatics	Universidad de Concepcion, Chile
2021	Thesis external reviewer	Ph.D. in Nutrition and Food	Universidad de Chile, Chile
2021	Research rotation advisor	M.Sc. in Biological Sciences	Universidad de Chile, Chile
2020	Research rotation advisor	Ph.D. in Science and Innovation in Medicine	Universidad del Desarrollo, Chile
2018	Research rotation advisor	Ph.D. in Science and Innovation in Medicine	Universidad del Desarrollo, Chile

2. Undergraduate

Year	Course/Activity	Program	University
2020	Thesis advisor	Medical Technology	Universidad del Desarrollo, Chile
2019	Thesis co-advisor	Biotechnology	Universidad Mayor, Chile

Memberships

- Sociedad de Genética de Chile (SOCHIGEN, Secretary General 2022-2024)
- Sociedad Chilena de Evolución (SOCEVOL)
- Sociedad Chilena de Bioinformática (SCB)
- Sociedad Española de Bioinformática y Biología Computacional (SEBiBC)
- American Society of Human Genetics

Peer reviewed publications

(*) equal contribution

1. Poli MC, **Rebolledo-Jaramillo B**, Lagos C, Orellana J, Moreno G, Martín LM, Encina G, Böhme D, Faundes V, Zavala MJ, Hasbún T, Fischer S, Brito F, Araya D, Lira M, de la Cruz J, Astudillo C, Lay-Son G, Cares C, Aracena M, Martin ES, Coban-Akdemir Z, Posey JE, Lupski JR, Repetto GM. Decoding complex inherited phenotypes in rare disorders: the DECIPHERD initiative for rare undiagnosed diseases in Chile. *Eur J Hum Genet.* 2024 Jan 4. [doi:10.1038/s41431-023-01523-5](https://doi.org/10.1038/s41431-023-01523-5)
2. 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](https://doi.org/10.3390/ijms24054915)
3. 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](https://doi.org/10.1111/iwj.13922)
4. 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](https://doi.org/10.1111/ijd.15887)
5. 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](https://doi.org/10.3390/cells10082159)

6. 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
7. **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
8. Fuentes I, Guttman-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
9. 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
10. 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
11. **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
12. 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
13. 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
14. **Rebolledo-Jaramillo B***, Su MS*, McElhoe J, Stoller N, Dickins B, Korneliusen 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.1409328111
15. **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
16. 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
17. 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
18. 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

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. doi:10.1016/B978-0-12-813695-9.00003-0
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. DOI: 10.5772/28486