



## Mauro Lago Docampo

Postdoctoral Scholar, Cardiology

### Bio

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#### HONORS AND AWARDS

- Postdoctoral Fellowship, American Heart Association (01/01/2025)
- Postdoctoral Fellowship, Fundación Ramón Areces (10/01/2022)

#### STANFORD ADVISORS

- Marlene Rabinovitch, Postdoctoral Faculty Sponsor

### Research & Scholarship

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#### CURRENT RESEARCH AND SCHOLARLY INTERESTS

I have always been fascinated by the process of genetic penetrance, how two people can carry the same mutation but only one of them may be affected.

My main project focuses on understanding the role of TBX4 in the development of Pulmonary Arterial Hypertension (PAH). Mutations in this gene are related to very different diseases. In my research, I mix cellular models, gene editing, and high throughput assays to find the determinants of penetrance in the development of PAH under these condition...

#### LAB AFFILIATIONS

- Marlene Rabinovitch, Rabinovitch lab (3/21/2022)

### Publications

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#### PUBLICATIONS

- **First Genotype-Phenotype Study in TBX4 Syndrome Gain-of-Function Mutations Causative for Lung Disease** *AMERICAN JOURNAL OF RESPIRATORY AND CRITICAL CARE MEDICINE*  
Prapa, M., Lago-Docampo, M., Swietlik, E. M., Montani, D., Eyries, M., Humbert, M., Welch, C. L., Chung, W. K., Berger, R. M. F., Bogaard, H., Danhaive, O., Escribano-Subias, P., Gall, et al  
2022; 206 (12): 1522-1533
- **Reduced FOXF1 links unrepaired DNA damage to pulmonary arterial hypertension.** *Nature communications*  
Isobe, S., Nair, R. V., Kang, H. Y., Wang, L., Moonen, J. R., Shinohara, T., Cao, A., Taylor, S., Otsuki, S., Marciano, D. P., Harper, R. L., Adil, M. S., Zhang, et al  
2023; 14 (1): 7578
- **Novel Loss of Function KCNA5 Variants in Pulmonary Arterial Hypertension.** *American journal of respiratory cell and molecular biology*  
Vera-Zambrano, A., Lago-Docampo, M., Gallego, N., Franco-Gonzalez, J. F., Morales-Cano, D., Cruz-Utrilla, A., Villegas-Esguevillas, M., Fernandez-Malave, E., Escribano-Subias, P., Tenorio-Castano, J. A., Perez-Vizcaino, F., Valverde, D., Gonzalez, et al

2023

- **Common Variation in EDN1 Regulatory Regions Highlights the Role of PPAR gamma as a Key Regulator of Endothelin in vitro** *FRONTIERS IN CARDIOVASCULAR MEDICINE*  
Lago-Docampo, M., Solarat, C., Mendez-Martinez, L., Baloira, A., Valverde, D.  
2022; 9: 823133
- **Novel Genetic and Molecular Pathways in Pulmonary Arterial Hypertension Associated with Connective Tissue Disease** *CELLS*  
Hernandez-Gonzalez, I., Tenorio-Castano, J., Ochoa-Parra, N., Gallego, N., Perez-Olivares, C., Lago-Docampo, M., Palomino Doza, J., Valverde, D., Lapunzina, P., Escribano-Subias, P.  
2021; 10 (6)
- **ALMS1 Regulates TGF-beta Signaling and Morphology of Primary Cilia** *FRONTIERS IN CELL AND DEVELOPMENTAL BIOLOGY*  
Alvarez-Satta, M., Lago-Docampo, M., Bea-Mascato, B., Solarat, C., Castro-Sanchez, S., Christensen, S. T., Valverde, D.  
2021; 9: 623829
- **Customized Massive Parallel Sequencing Panel for Diagnosis of Pulmonary Arterial Hypertension** *GENES*  
Castano, J., Hernandez-Gonzalez, I., Gallego, N., Perez-Olivares, C., Ochoa Parra, N., Arias, P., Granda, E., Acebo, G., Lago-Docampo, M., Palomino-Doza, J., Lopez Meseguer, M., del Cerro, M., Valverde, et al  
2020; 11 (10)
- **Characterization of rare ABCC8 variants identified in Spanish pulmonary arterial hypertension patients** *SCIENTIFIC REPORTS*  
Lago-Docampo, M., Tenorio, J., Hernandez-Gonzalez, I., Perez-Olivares, C., Escribano-Subias, P., Pousada, G., Baloira, A., Arenas, M., Lapunzina, P., Valverde, D.  
2020; 10 (1): 15135
- **Clinical heterogeneity of Pulmonary Arterial Hypertension associated with variants in TBX4** *PLOS ONE*  
Hernandez-Gonzalez, I., Tenorio, J., Palomino-Doza, J., Martinez Menaca, A., Morales Ruiz, R., Lago-Docampo, M., Valverde Gomez, M., Gomez Roman, J., Enguita Valls, A., Perez-Olivares, C., Valverde, D., Gil Carbonell, J., Garrido-Lestache Rodriguez-Monte, et al  
2020; 15 (4): e0232216
- **Functional assessment of the BMPR2 gene in lymphoblastoid cell lines from Graves' disease patients** *JOURNAL OF CELLULAR AND MOLECULAR MEDICINE*  
Pousada, G., Lago-Docampo, M., Prado, S., Varela-Calvino, R., Mantinan, B., Valverde, D.  
2018; 22 (3): 1538-1547