Stanford

Hector Rodrigo Mendez

Postdoctoral Scholar, Cardiovascular Medicine

Bio

BIO

Dr. Hector Rodrigo Mendez is a Medical Geneticist from Argentina. Rodrigo completed a residency program in Medical Genetics at Centro Nacional de Genetica Medica – ANLIS (Buenos Aires, Argentina) and a Master's program in Medical Molecular Biology at Buenos Aires University.

Rodrigo continued his scientific career at a German Genomic Start-up, working as a human geneticist and providing his experience in rare disorders, genomic data (WGS/WES/gene panels) analysis, variant interpretation, and its integration with a deep focus on genotype-phenotype correlation.

Rodrigo's areas of expertise are rare disorders, NGS technology, Whole Genome Sequencing analysis, and ACMG interpretation guidelines, and his research aims are:

- Collection and analysis of clinical data through deep-learning phenotyping approaches.
- Multi-omic data integration to elucidate complex and rare genetic disorders.
- International collaborations to break down barriers to research participation amongst those who have been under-represented.

At Stanford University, under the supervision of Dr. Matthew Wheeler, he is conducting his postdoctoral research studies to achieve his scientific goals.

HONORS AND AWARDS

- Advocacy Certificate for Human Genetics and Genomics Program, American Society of Human Genetics (2024)
- The Milagros para Niños Observership Scholarship Award in Genetics, Boston Children´s Hospital – Harvard Medical School (2019)
- The International School on Inherited Ataxias: from genetics to clinics Scholarship Award, IBRO (International Brain Research Organization) (2019)
- The 13th International Congress of Inborn Errors of Metabolism Young Delegate Scholarship Award 2017, Latin American Society of Inborn Errors of Metabolism and Neonatal Screening. (2017)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Member, American College of Medical Genetics & Genomics (2023 - present)
- Member, American Society of Human Genetics (2023 - present)
- Member, European Society of Human Genetics (2023 - present)

STANFORD ADVISORS

- Matthew Wheeler, Postdoctoral Faculty Sponsor

LINKS

- My Linkedin Profile: https://www.linkedin.com/in/rmendezgen/

Publications

PUBLICATIONS

- **De novo variants in the non-coding spliceosomal snRNA gene RNU4-2 are a frequent cause of syndromic neurodevelopmental disorders.** *medRxiv : the preprint server for health sciences*

  Chen, Y., Dawes, R., Kim, H. C., Stenton, S. L., Walker, S., Ljungdahl, A., Lord, J., Ganesh, V. S., Ma, J., Martin-Geary, A. C., Lemire, G., D'Souza, E. N., Dong, et al

  2024

- **Genetic Analysis Algorithm for the Study of Patients with Multiple Congenital Anomalies and Isolated Congenital Heart Disease.** *Genes*

  Delea, M., Massara, L. S., Espeche, L. D., Bidondo, M. P., Barbero, P., Oliveri, J., Brun, P., Fabro, M., Galain, M., Fernández, C. S., Taboas, M., Bruque, C. D., Kolomenski, et al

  2022; 13 (7)

- **Oculocutaneous albinism type 1B associated with a functionally significant tyrosinase gene polymorphism detected with Whole Exome Sequencing.** *Ophthalmic genetics*

  Mendez, R., Iqbal, S., Vishnopolska, S., Martinez, C., Dibner, G., Aliano, R., Zaiat, J., Biagioli, G., Fernandez, C., Turjanski, A., Campbell, A. J., Mercado, G., Marti, et al

  2021; 42 (3): 291-295

- **A novel pathogenic frameshift variant of KAT6B identified by clinical exome sequencing in a newborn with the Say-Barber-Biesecker-Young-Simpson syndrome.** *Clinical dysmorphology*

  Mendez, R., Delea, M., Dain, L., Rittler, M.

  2020; 29 (1): 42-45