



## Helio Costa

Adjunct Clinical Assistant Professor, Pathology

### CLINICAL OFFICE (PRIMARY)

- **Molecular Genetic Pathology Laboratory**

3375 Hillview Ave Rm 2201

MC 5563

Palo Alto, CA 94304

**Tel** (650) 723-6574      **Fax** (650) 724-1567

### Bio

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#### BIO

Helio Costa, PhD, is a medical geneticist with expertise in oncology, medical genetics and genomics, computational biology, data science, software engineering, and product development. He is passionate about leveraging his interdisciplinary skillset to build and develop commercial-grade cancer diagnostic products and medical software that aid in patient care and clinical decision support. Currently he is Medical Director of Oncology at Natera, and an Adjunct Clinical Assistant Professor in the Department of Pathology at Stanford Medical School.

Dr. Costa's research focuses on developing and implementing new medical diagnostic genetic tests and software for use in patient care. His research group developed DNA and RNA cancer diagnostic tests currently in use at Stanford Health Care as well as developing clinical algorithms using large-scale clinical laboratory datasets and patient electronic medical records to predict patient outcomes and aid in therapeutic clinical decision support. Additionally, Dr. Costa served as a co-Investigator in the NIH Clinical Genome Resource (ClinGen) Consortium, and led the engineering and product management teams developing FDA-recognized medical software applications used by healthcare providers, researchers, and biotechnology companies to define the clinical relevance of genes and pathogenicity of mutations identified in patients.

Dr. Costa is the founding director of the Stanford Clinical Data Science Fellowship where post-doctoral fellows engage in interdisciplinary clinical research and embed in health care workflows learning, building and deploying real-world health data solutions in the Stanford Health Care system. He is currently an Attending Medical Geneticist for the Molecular Genetic Pathology Laboratory at Stanford Health Care where he previously served as an Assistant Lab Director.

Dr. Costa received his BS in Genetics from University of California at Davis, his PhD in Genetics from Stanford University School of Medicine, and his ABMGG Clinical Molecular Genetics and Genomics fellowship training from Stanford University School of Medicine.

#### CLINICAL FOCUS

- Molecular Oncology
- Molecular Pathology

- Medical Genetics
- Clinical Pathology

## ACADEMIC APPOINTMENTS

- Member, Maternal & Child Health Research Institute (MCHRI)

## ADMINISTRATIVE APPOINTMENTS

- Attending Medical Geneticist, Molecular Genetic Pathology Laboratory, (2017- present)
- Founding Director, Stanford Clinical Data Science Fellowship, (2018-2021)
- Assistant Lab Director, Molecular Genetic Pathology Laboratory, (2017-2021)

## PROFESSIONAL EDUCATION

- Board Certification: Technologist in Molecular Biology, American Society for Clinical Pathology (2018)
- Fellowship, Stanford University School of Medicine , ABMGG Clinical Molecular Genetics and Genomics (2017)
- Doctor of Philosophy, Stanford University School of Medicine , Genetics (2015)
- Bachelor of Science, University of California, Davis , Genetics (2010)

## LINKS

- LinkedIn: <https://linkedin.com/in/helio-costa>

## Teaching

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### GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Biomedical Informatics (Phd Program)
- Biomedical Informatics (Masters Program)

## Publications

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### PUBLICATIONS

- **Increasing Clinical Trial Accrual via Automated Matching of Biomarker Criteria.** *Pacific Symposium on Biocomputing. Pacific Symposium on Biocomputing*  
Chen, J. W., Kunder, C. A., Bui, N. n., Zehnder, J. L., Costa, H. A., Stehr, H. n.  
2020; 25: 31–42
- **LitGen: Genetic Literature Recommendation Guided by Human Explanations.** *Pacific Symposium on Biocomputing. Pacific Symposium on Biocomputing*  
Nie, A. n., Pineda, A. L., Wright, M. W., Wand, H. n., Wulf, B. n., Costa, H. A., Patel, R. Y., Bustamante, C. D., Zou, J. n.  
2020; 25: 67–78
- **Genomic Evidence for Local Adaptation of Hunter-Gatherers to the African Rainforest.** *Current biology : CB*  
Lopez, M., Choin, J., Sikora, M., Siddle, K., Harmant, C., Costa, H. A., Silvert, M., Mouguiama-Daouda, P., Hombert, J., Froment, A., Le Bomin, S., Perry, G. H., Barreiro, et al  
2019
- **Structural Variation Detection by Proximity Ligation from Formalin-Fixed, Paraffin-Embedded Tumor Tissue.** *The Journal of molecular diagnostics : JMD*  
Troll, C. J., Putnam, N. H., Hartley, P. D., Rice, B., Blanchette, M., Siddiqui, S., Ganbat, J., Powers, M. P., Ramakrishnan, R., Kunder, C. A., Bustamante, C. D., Zehnder, J. L., Green, et al  
2018
- **Gene-specific criteria for PTEN variant curation: Recommendations from the ClinGen PTEN Expert Panel.** *Human mutation*  
Mester, J. L., Ghosh, R., Pesaran, T., Huether, R., Karam, R., Hruska, K. S., Costa, H. A., Lachlan, K., Ngeow, J., Barnholtz-Sloan, J., Sesock, K., Hernandez, F., Zhang, et al  
2018; 39 (11): 1581–92

- **Tumor Molecular Profiling Aids in Determining Tissue of Origin and Therapy for Metastatic Adenocarcinoma in a Patient With Multiple Primary Malignancies** *JCO PRECISION ONCOLOGY*  
Costa, H. A., Reyes, R., Mills, M., Zehnder, J. L., Sledge, G., Curtis, C., Ford, J. M., Suarez, C. J.  
2018; 2
- **Detection and surveillance of bladder cancer using urine tumor DNA.** *Cancer discovery*  
Dudley, J. C., Schroers-Martin, J. n., Lazzareschi, D. V., Shi, W. Y., Chen, S. B., Esfahani, M. S., Trivedi, D. n., Chabon, J. J., Chaudhuri, A. A., Stehr, H. n., Liu, C. L., Lim, H. n., Costa, et al  
2018
- **Promoting appropriate genetic testing: the impact of a combined test review and consultative service.** *Genetics in medicine*  
Suarez, C. J., Yu, L., Downs, N., Costa, H. A., Stevenson, D. A.  
2017
- **Identification of a Novel Somatic Mutation Leading to Allele Dropout for EGFR L858R Genotyping in Non-Small Cell Lung Cancer** *Molecular Diagnosis & Therapy*  
Costa, H. A., Neal, J. W., Bustamante, C. D., Zehnder, J. L.  
2017
- **Discovery and functional characterization of a neomorphic PTEN mutation** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*  
Costa, H. A., Leitner, M. G., Sos, M. L., Mavrantoni, A., Rychkova, A., Johnson, J. R., Newton, B. W., Yee, M., De La Vega, F. M., Ford, J. M., Krogan, N. J., Shokat, K. M., Oliver, et al  
2015; 112 (45): 13976-13981
- **Transcriptome sequencing from diverse human populations reveals differentiated regulatory architecture.** *PLoS genetics*  
Martin, A. R., Costa, H. A., Lappalainen, T., Henn, B. M., Kidd, J. M., Yee, M., Grubert, F., Cann, H. M., Snyder, M., Montgomery, S. B., Bustamante, C. D.  
2014; 10 (8)
- **High-throughput Sequencing of Subcutaneous Panniculitis-like T-Cell Lymphoma Reveals Candidate Pathogenic Mutations.** *Applied immunohistochemistry & molecular morphology : AIMM*  
Fernandez-Pol, S. n., Costa, H. A., Steiner, D. F., Ma, L. n., Merker, J. D., Kim, Y. H., Arber, D. A., Kim, J. n.  
; 27 (10): 740-48