



## Maya M. Kasowski

Assistant Professor of Pathology, of Medicine (Pulmonary, Allergy and Critical Care Medicine) and, by courtesy, of Genetics

### CLINICAL OFFICE (PRIMARY)

- **Department of Pathology**

3375 Hillview Ave

Palo Alto, CA 94304

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

### Bio

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#### BIO

I am a clinical pathologist and assistant professor in the Departments of Medicine, Pathology, and Genetics (by courtesy) at Stanford. I completed my MD-PhD training at Yale University and my residency training and a post-doctoral fellowship in the Department of Genetics at Stanford University. My experiences as a clinical pathologist and genome scientist have made me passionate about applying cutting-edge technologies to primary patient specimens in order to characterize disease pathologies at the molecular level. The core focus of my lab is to study the mechanisms by which genetic variants influence the risk of disease through effects on intermediate molecular phenotypes.

#### CLINICAL FOCUS

- Anatomic and Clinical Pathology

#### ACADEMIC APPOINTMENTS

- Assistant Professor, Pathology
- Assistant Professor, Medicine - Pulmonary, Allergy & Critical Care Medicine
- Assistant Professor (By courtesy), Genetics
- Member, Bio-X
- Member, Maternal & Child Health Research Institute (MCHRI)
- Member, Stanford Cancer Institute

#### PROFESSIONAL EDUCATION

- Residency: Stanford University Pathology Residency (2019) CA
- Medical Education: Yale School Of Medicine (2016) CT

### Teaching

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#### STANFORD ADVISEES

Doctoral Dissertation Reader (AC)

Samson Mataraso

**Doctoral (Program)**

Shouvik Mani, Arpita Singhal

## Publications

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### PUBLICATIONS

- **Chromatin activity identifies differential gene regulation across human ancestries.** *Genome biology*  
Pettie, K. P., Mumbach, M., Lea, A. J., Ayroles, J., Chang, H. Y., Kasowski, M., Fraser, H. B.  
2024; 25 (1): 21
- **Author Correction: Perspectives on ENCODE.** *Nature*  
ENCODE Project Consortium, Snyder, M. P., Gingeras, T. R., Moore, J. E., Weng, Z., Gerstein, M. B., Ren, B., Hardison, R. C., Stamatoyannopoulos, J. A., Graveley, B. R., Feingold, E. A., Pazin, M. J., Pagan, M., et al  
2022
- **Author Correction: Expanded encyclopaedias of DNA elements in the human and mouse genomes.** *Nature*  
ENCODE Project Consortium, Moore, J. E., Purcaro, M. J., Pratt, H. E., Epstein, C. B., Shores, N., Adrian, J., Kawli, T., Davis, C. A., Dobin, A., Kaul, R., Halow, J., Van Nostrand, E. L., et al  
2022
- **Early peanut introduction wins over the HLA-DQA1\*01:02 allele in the interplay between environment and genetics** *JOURNAL OF CLINICAL INVESTIGATION*  
Manohar, M., Nadeau, K., Kasowski, M.  
2022; 132 (1)
- **Chromatin accessibility associates with protein-RNA correlation in human cancer.** *Nature communications*  
Sanghi, A., Gruber, J. J., Metwally, A., Jiang, L., Reynolds, W., Sunwoo, J., Orloff, L., Chang, H. Y., Kasowski, M., Snyder, M. P.  
2021; 12 (1): 5732
- **Detection of cryptogenic malignancies from metagenomic whole genome sequencing of body fluids.** *Genome medicine*  
Gu, W., Talevich, E., Hsu, E., Qi, Z., Urisman, A., Federman, S., Gopez, A., Arevalo, S., Gottschall, M., Liao, L., Tung, J., Chen, L., Lim, et al  
2021; 13 (1): 98
- **Perspectives on ENCODE.** *Nature*  
ENCODE Project Consortium, Snyder, M. P., Gingeras, T. R., Moore, J. E., Weng, Z., Gerstein, M. B., Ren, B., Hardison, R. C., Stamatoyannopoulos, J. A., Graveley, B. R., Feingold, E. A., Pazin, M. J., Pagan, M., et al  
2020; 583 (7818): 693–98
- **Metabolomics in acute myeloid leukemia.** *Molecular genetics and metabolism*  
Wojcicki, A. V., Kasowski, M. M., Sakamoto, K. M., Lacayo, N.  
2020
- **Expanded encyclopaedias of DNA elements in the human and mouse genomes.** *Nature*  
Moore, J. E., Purcaro, M. J., Pratt, H. E., Epstein, C. B., Shores, N. n., Adrian, J. n., Kawli, T. n., Davis, C. A., Dobin, A. n., Kaul, R. n., Halow, J. n., Van Nostrand, E. L., Freese, et al  
2020; 583 (7818): 699–710
- **Remodeling of active endothelial enhancers is associated with aberrant gene-regulatory networks in pulmonary arterial hypertension.** *Nature communications*  
Reyes-Palomares, A. n., Gu, M. n., Grubert, F. n., Berest, I. n., Sa, S. n., Kasowski, M. n., Arnold, C. n., Shuai, M. n., Srivas, R. n., Miao, S. n., Li, D. n., Snyder, M. P., Rabinovitch, et al  
2020; 11 (1): 1673
- **Landscape of cohesin-mediated chromatin loops in the human genome.** *Nature*  
Gruber, F. n., Srivas, R. n., Spacek, D. V., Kasowski, M. n., Ruiz-Velasco, M. n., Sinnott-Armstrong, N. n., Greenside, P. n., Narasimha, A. n., Liu, Q. n., Geller, B. n., Sanghi, A. n., Kulik, M. n., Sa, et al  
2020; 583 (7818): 737–43

- **An improved ATAC-seq protocol reduces background and enables interrogation of frozen tissues.** *Nature methods*  
Corces, M. R., Trevino, A. E., Hamilton, E. G., Greenside, P. G., Sinnott-Armstrong, N. A., Vesuna, S. n., Satpathy, A. T., Rubin, A. J., Montine, K. S., Wu, B. n., Kathiria, A. n., Cho, S. W., Mumbach, et al  
2017
- **Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions** *CELL*  
Grubert, F., Zaugg, J. B., Kasowski, M., Ursu, O., Spacek, D. V., Martin, A. R., Greenside, P., Srivas, R., Phanstiel, D. H., Pekowska, A., Heidari, N., Euskirchen, G., Huber, et al  
2015; 162 (5): 1051-1065
- **Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions.** *Cell*  
Grubert, F., Zaugg, J. B., Kasowski, M., Ursu, O., Spacek, D. V., Martin, A. R., Greenside, P., Srivas, R., Phanstiel, D. H., Pekowska, A., Heidari, N., Euskirchen, G., Huber, et al  
2015; 162 (5): 1051-1065
- **Genome-wide map of regulatory interactions in the human genome** *GENOME RESEARCH*  
Heidari, N., Phanstiel, D. H., He, C., Grubert, F., Jahanbani, F., Kasowski, M., Zhang, M. Q., Snyder, M. P.  
2014; 24 (12): 1905-1917
- **Extensive Variation in Chromatin States Across Humans** *SCIENCE*  
Kasowski, M., Kyriazopoulou-Panagiotopoulou, S., Grubert, F., Zaugg, J. B., Kundaje, A., Liu, Y., Boyle, A. P., Zhang, Q. C., Zakharia, F., Spacek, D. V., Li, J., Xie, D., Olarerin-George, et al  
2013; 342 (6159): 750-752
- **An integrated encyclopedia of DNA elements in the human genome** *NATURE*  
Dunham, I., Kundaje, A., Aldred, S. F., Collins, P. J., Davis, C., Doyle, F., Epstein, C. B., Fietze, S., Harrow, J., Kaul, R., Khatun, J., Lajoie, B. R., Landt, et al  
2012; 489 (7414): 57-74
- **Architecture of the human regulatory network derived from ENCODE data** *NATURE*  
Gerstein, M. B., Kundaje, A., Hariharan, M., Landt, S. G., Yan, K., Cheng, C., Mu, X. J., Khurana, E., Rozowsky, J., Alexander, R., Min, R., Alves, P., Abyzov, et al  
2012; 489 (7414): 91-100
- **Annotation of functional variation in personal genomes using RegulomeDB** *GENOME RESEARCH*  
Boyle, A. P., Hong, E. L., Hariharan, M., Cheng, Y., Schaub, M. A., Kasowski, M., Karczewski, K. J., Park, J., Hitz, B. C., Weng, S., Cherry, J. M., Snyder, M.  
2012; 22 (9): 1790-1797
- **Personal Omics Profiling Reveals Dynamic Molecular and Medical Phenotypes** *CELL*  
Chen, R., Mias, G. I., Li-Pook-Than, J., Jiang, L., Lam, H. Y., Chen, R., Miriami, E., Karczewski, K. J., Hariharan, M., Dewey, F. E., Cheng, Y., Clark, M. J., Im, et al  
2012; 148 (6): 1293-1307
- **A User's Guide to the Encyclopedia of DNA Elements (ENCODE)** *PLOS BIOLOGY*  
Myers, R. M., Stamatoyannopoulos, J., Snyder, M., Dunham, I., Hardison, R. C., Bernstein, B. E., Gingeras, T. R., Kent, W. J., Birney, E., Wold, B., Crawford, G. E., Bernstein, B. E., Epstein, et al  
2011; 9 (4)
- **Variation in Transcription Factor Binding Among Humans** *SCIENCE*  
Kasowski, M., Grubert, F., Heffelfinger, C., Hariharan, M., Asabere, A., Waszak, S. M., Habegger, L., Rozowsky, J., Shi, M., Urban, A. E., Hong, M., Karczewski, K. J., Huber, et al  
2010; 328 (5975): 232-235