Stanford

# Elizabeth Spiteri

Clinical Associate Professor, Pathology


### CLINICAL OFFICE (PRIMARY)

- **Department of Pathology**

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## Bio

### CLINICAL FOCUS

- Clinical Molecular Genetics
- Clinical Cytogenetics
- Clinical Pathology

### ACADEMIC APPOINTMENTS

- Clinical Associate Professor, Pathology
- Member, Maternal & Child Health Research Institute (MCHRI)

### PROFESSIONAL EDUCATION

- Board Certification: Clinical Molecular Genetics and Genomics, American Board of Medical Genetics and Genomics (2018)
- Board Certification: Clinical Cytogenetics and Genomics, American Board of Medical Genetics and Genomics (2005)
- Fellowship: UCLA David Geffen School Of Medicine Registrar (2006) CA
- Fellowship: UCLA David Geffen School Of Medicine Registrar (2005) CA
- PhD Training: Albert Einstein College of Medicine (2002) NY
- Board Certification, Clinical Molecular Genetics , American Board of Medical Genetics and Genomics (2007)

## Publications

### PUBLICATIONS

- **Performance of MYC, BCL2, and BCL6 break-apart FISH in small biopsies with large B-cell lymphoma: a retrospective Cytopathology Hematopathology Interinstitutional Consortium study.** *Frontiers in oncology*

  Menke, J. R., Aypar, U., Bangs, C. D., Cook, S. L., Gupta, S., Hasserjian, R. P., Kong, C. S., Lin, O., Long, S. R., Ly, A., Menke, J. A., Natkunam, Y., Ruiz-Cordero, et al

  2024; 14: 1408238

- **Advancing access to genome sequencing for rare genetic disorders: recent progress and call to action.** *NPJ genomic medicine*

  Jobanputra, V., Schroeder, B., Rehm, H. L., Shen, W., Spiteri, E., Nakouzi, G., Taylor, S., Marshall, C. R., Meng, L., Kingsmore, S. F., Ellsworth, K., Ashley, E., Taft, et al

2024; 9 (1): 23

- **Evidence review and considerations for use of first line genome sequencing to diagnose rare genetic disorders.** *NPJ genomic medicine*
  Wigby, K. M., Brockman, D., Costain, G., Hale, C., Taylor, S. L., Belmont, J., Bick, D., Dimmock, D., Fernbach, S., Greally, J., Jobanputra, V., Kulkarni, S., Spiteri, et al
  2024; 9 (1): 15

- **Section E6.7-6.12 of the American College of Medical Genetics and Genomics (ACMG) Technical Laboratory Standards: Cytogenomic studies of acquired chromosomal abnormalities in solid tumors.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Church, A. J., Akkari, Y., Deeb, K., Kolhe, R., Lin, F., Spiteri, E., Wolff, D. J., Shao, L.
  2024: 101070

- **The clinical, molecular, and prognostic features of the 2022 WHO and ICC classification systems for myelodysplastic neoplasms.** *Leukemia research*
  Khanna, V., Lu, R., Kumar, J., Molina, A., Stehr, H., Spiteri, E., Spinner, M., Silva, O., Fernandez-Pol, S., Tan, B., Greenberg, P. L.
  2023; 136: 107433

- **Two epilepsy-associated variants in KCNA2 (KV1.2) at position H310 oppositely affect channel functional expression.** *The Journal of physiology*
  Minguez-Vinas, T., Prakash, V., Wang, K., Lindstrom, S. H., Pozzi, S., Scott, S. A., Spiteri, E., Stevenson, D. A., Ashley, E. A., Gunnarsson, C., Pantazis, A.
  2023

- **The landscape of reported VUS in multi-gene panel and genomic testing: Time for a change.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Rehm, H. L., Alaimo, J. T., Aradhya, S., Bayrak-Toydemir, P., Best, H., Brandon, R., Buchan, J. G., Chao, E. C., Chen, E., Clifford, J., Cohen, A. S., Conlin, L. K., Das, et al
  2023: 100947

- **Accurate Detection of Clinically Actionable Copy Number Variants in Diverse Hematological Neoplasms By Routine Targeted Sequencing: A Comparative Performance Study**
  Mosquera, A., Hosoya, H., Jin, M. C., Esfahani, M., Schroers-Martin, J., Sworder, B., Liu, C., Spiteri, E., Natkunam, Y., Zehnder, J. L., Stehr, H., Kurtz, D. M., Alizadeh, et al
  AMER SOC HEMATOLOGY.2022: 10712-10713

- **Deconvoluting complex correlates of COVID-19 severity with a multi-omic pandemic tracking strategy.** *Nature communications*
  Parikh, V. N., Ioannidis, A. G., Jimenez-Morales, D., Gorzynski, J. E., De Jong, H. N., Liu, X., Roque, J., Cepeda-Espinoza, V. P., Osoegawa, K., Hughes, C., Sutton, S. C., Youlton, N., Joshi, et al
  2022; 13 (1): 5107

- **Loss-of-function variants in SRRM2 cause a neurodevelopmental disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Cuinat, S., Nizon, M., Isidor, B., Stegmann, A., van Jaarsveld, R. H., van Gassen, K. L., van der Smagt, J. J., Volker-Touw, C. M., Holwerda, S. J., Terhal, P. A., Schuhmann, S., Vasileiou, G., Khalifa, et al
  2022

- **Best practices for the interpretation and reporting of clinical whole genome sequencing.** *NPJ genomic medicine*
  Austin-Tse, C. A., Jobanputra, V., Perry, D. L., Bick, D., Taft, R. J., Venner, E., Gibbs, R. A., Young, T., Barnett, S., Belmont, J. W., Boczek, N., Chowdhury, S., Ellsworth, et al
  2022; 7 (1): 27

- **Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing.** *Nature biotechnology*
  Goenka, S. D., Gorzynski, J. E., Shafin, K., Fisk, D. G., Pesout, T., Jensen, T. D., Monlong, J., Chang, P. C., Baid, G., Bernstein, J. A., Christle, J. W., Dalton, K. P., Garalde, et al
  2022

- **Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock.** *Circulation. Genomic and precision medicine*
  Gorzynski, J. E., Goenka, S. D., Shafin, K., Jensen, T. D., Fisk, D. G., Grove, M. E., Spiteri, E., Pesout, T., Monlong, J., Bernstein, J. A., Ceresnak, S., Chang, P., Christle, et al
  2022: CIRCGEN121003591

- **Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting.** *The New England journal of medicine*
  Gorzynski, J. E., Goenka, S. D., Shafin, K., Jensen, T. D., Fisk, D. G., Grove, M. E., Spiteri, E., Pesout, T., Monlong, J., Baid, G., Bernstein, J. A., Ceresnak, S., Chang, et al
  2022

- **Comparison of the Transcriptomic Signatures in Pediatric and Adult CML.** *Cancers*
  Youn, M., Smith, S. M., Lee, A. G., Chae, H., Spiteri, E., Erdmann, J., Galperin, I., Jones, L. M., Donato, M., Abidi, P., Bittencourt, H., Lacayo, N., Dahl, et al
  1800; 13 (24)

- **Identification of a pathogenic TUBB1 variant in a Chinese family with congenital macrothrombocytopenia through whole genome sequencing.** *Platelets*
  Hou, Y. n., Shao, L. n., Zhou, H. n., Liu, Y. n., Fisk, D. G., Spiteri, E. n., Zehnder, J. L., Peng, J. n., Zhang, B. M., Hou, M. n.
  2021: 1–5

- **Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Melo, U. S., Bonner, D. n., Kent Lloyd, K. C., Moshiri, A. n., Willis, B. n., Lanoue, L. n., Bower, L. n., Leonard, B. C., Martins, D. J., Gomes, F. n., de Souza Leite, F. n., Oliveira, D. n., Kitajima, et al
  2021