Stanford

Elizabeth Spiteri

Clinical Associate Professor, Pathology

CLINICAL OFFICES

- **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

- 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)
- Board Certification: Clinical Cytogenetics, American Board of Medical Genetics and Genomics (2005)

Publications

PUBLICATIONS

- **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
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- **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