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Yael Wilnai Ziskind

Clinical Assistant Professor, Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

- **Center for Academic Medicine- Genetics**

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Bio

CLINICAL FOCUS

- Clinical Genetics and Genomics

ACADEMIC APPOINTMENTS

- Clinical Assistant Professor, Pediatrics - Medical Genetics

PROFESSIONAL EDUCATION

- Fellowship: Stanford University School of Medicine (2012) CA
- Board Certification: Clinical Genetics and Genomics, American Board of Medical Genetics and Genomics (2024)
- Board Certification: Clinical Biochemical Genetics, American Board of Medical Genetics and Genomics (2015)
- Fellowship: Stanford University - Fellowship (2014) CA
- Residency: Tel Aviv Sourasky Medical Center (2008) Israel
- Internship: Wolfson Medical Center (2004) Israel
- Medical Education: Sackler School of Medicine (2003) Israel

Publications

PUBLICATIONS

- **Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner Syndrome**

  Sheppard, S., Campbell, I., Harr, M., Gold, N., Li, D., Bjornsson, H., Cohen, J., Fahrner, J., Fatemi, A., Harris, J., Nowak, C., Stevens, C., Grand, et al

  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S183

- **Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome.** *American journal of medical genetics. Part A*

  Sheppard, S. E., Campbell, I. M., Harr, M. H., Gold, N., Li, D., Bjornsson, H. T., Cohen, J. S., Fahrner, J. A., Fatemi, A., Harris, J. R., Nowak, C., Stevens, C. A., Grand, et al

  2021

- **Prenatal treatment of ornithine transcarbamylase deficiency.** *Molecular genetics and metabolism*

  Wilnai, Y. n., Blumenfeld, Y. J., Cusmano, K. n., Hintz, S. R., Alcorn, D. n., Benitz, W. E., Berquist, W. E., Bernstein, J. A., Castillo, R. O., Concepcion, W. n., Cowan, T. M., Cox, K. L., Lyell, et al

  2018

- **Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations.** *American journal of human genetics*
  Lalani, S. R., Liu, P., Rosenfeld, J. A., Watkin, L. B., Chiang, T., Leduc, M. S., Zhu, W., Ding, Y., Pan, S., Vetrini, F., Miyake, C. Y., Shinawi, M., Gambin, et al
  2016; 98 (2): 347-357

- **The Phenotype of the Musculocontractural Type of Ehlers-Danlos Syndrome due to CHST14 Mutations** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Janecke, A. R., Li, B., Boehm, M., Krabichler, B., Rohrbach, M., Mueller, T., Fuchs, I., Golas, G., Katagiri, Y., Ziegler, S. G., Gahl, W. A., Wilnai, Y., Zoppi, et al
  2016; 170 (1): 103-115

- **The phenotype of the musculocontractural type of Ehlers-Danlos syndrome due to CHST14 mutations.** *American journal of medical genetics. Part A*
  Janecke, A. R., Li, B., Boehm, M., Krabichler, B., Rohrbach, M., Müller, T., Fuchs, I., Golas, G., Katagiri, Y., Ziegler, S. G., Gahl, W. A., Wilnai, Y., Zoppi, et al
  2016; 170A (1): 103-115

- **Abnormal Hepatocellular Mitochondria in Methylmalonic Acidemia** *ULTRASTRUCTURAL PATHOLOGY*
  Wilnai, Y., Enns, G. M., Niemi, A., Higgins, J., Vogel, H.
  2014; 38 (5): 309-314

- **Abnormal hepatocellular mitochondria in methylmalonic acidemia.** *Ultrastructural pathology*
  Wilnai, Y., Enns, G. M., Niemi, A., Higgins, J., Vogel, H.
  2014; 38 (5): 309-314

- **Clinical whole-exome sequencing: are we there yet?** *GENETICS IN MEDICINE*
  Atwal, P. S., Brennan, M., Cox, R., Niaki, M., Platt, J., Homeyer, M., Kwan, A., Parkin, S., Schelley, S., Slattery, L., Wilnai, Y., Bernstein, J. A., Enns, et al
  2014; 16 (9): 717-719

- **Clinical whole-exome sequencing: are we there yet?** *Genetics in medicine*
  Atwal, P. S., Brennan, M., Cox, R., Niaki, M., Platt, J., Homeyer, M., Kwan, A., Parkin, S., Schelley, S., Slattery, L., Wilnai, Y., Bernstein, J. A., Enns, et al
  2014; 16 (9): 717-719

- **PRENATAL TREATMENT OF ORNITHINE TRANSCARBAMYLASE DEFICIENCY**
  Wilnai, Y., Alcorn, D., Benitz, W., Berquist, W., Bernstein, J., Blumenfeld, Y. J., Castillo, R., Concepcion, W., Cowan, T., Cox, K. L., Cusmano, K., Deirdre, L., Esquival, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: 248

- **Severe multi-systemic presentation of COX10 deficiency**
  Wilnai, Y., Cox, R., Bai, R., Enns, G. M.
  ELSEVIER SCI LTD.2013: 926

- **Axial spondylometaphyseal dysplasia with retinitis pigmentosa-a clinical report and diagnostic clues.** *Journal of applied genetics*
  Reinstein, E., Okenfuss, E. B., Wadhawan, I., Wilnai, Y., Manning, M., Rimoin, D. L., Lachman, R. S.
  2013; 54 (2): 231-234

- **Atypical Amyoplasia Congenita in an Infant With Leigh Syndrome: A Mitochondrial Cause of Severe Contractures?** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Wilnai, Y., Seaver, L. H., Enns, G. M.
  2012; 158A (9): 2353-2357