Stanford

# Kristina Cusmano-Ozog

Clinical Associate Professor, Pathology


### CLINICAL OFFICE (PRIMARY)

- **Stanford Healthcare Clinical Microbiology Laboratory**

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## Bio

### CLINICAL FOCUS

- Biochemical Genetics
- Metabolism, Inborn Errors
- Clinical Biochemical Genetics

### ACADEMIC APPOINTMENTS

- Clinical Associate Professor, Pathology

### PROFESSIONAL EDUCATION

- Medical Education: University of South Florida Morsani College of Medicine (2002) FL
- Residency: University of South Florida Pediatric Residency (2005) FL
- Residency: Stanford University Division of Medical Genetics (2007) CA
- Fellowship: Stanford University Clinical Biochemical Genetics Fellowship (2008) CA
- Fellowship: National Institute Of Health Medical Genetics and Genomics Fellowship (2015) MD
- Board Certification: Pediatrics, American Board of Pediatrics (2005)
- Board Certification: Clinical Biochemical Genetics, American Board of Medical Genetics and Genomics (2009)
- Board Certification: Clinical Molecular Genetics and Genomics, American Board of Medical Genetics and Genomics (2015)
- Board Certification: Clinical Genetics and Genomics, American Board of Medical Genetics and Genomics (2018)

## Teaching

### COURSES

**2023-24**

- Human Genetics: GENE 202 (Aut)

**2022-23**

- Human Genetics: GENE 202 (Aut)

## Publications

### PUBLICATIONS

- **A homozygous Gly470Ala variant in PEX6 causes severe Zellweger spectrum disorder.** *American journal of medical genetics. Part A*
  Galarreta, C. I., Wong, K., Carmichael, J., Woods, J., Tise, C. G., Niehaus, A. D., Schildt, A. J., Verscaj, C. P., Cusmano-Ozog, K. P.
  2023

- **MT-ATP6 mitochondrial disease identified by newborn screening reveals a distinct biochemical phenotype.** *American journal of medical genetics. Part A*
  Tise, C. G., Verscaj, C. P., Mendelsohn, B. A., Woods, J., Lee, C. U., Enns, G. M., Stander, Z., Hall, P. L., Cowan, T. M., Cusmano-Ozog, K. P.
  2023

- **Neonatal lupus is a novel cause of positive newborn screening for X-linked adrenoleukodystrophy.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Mendelsohn, B. A., Zimmerman, B., Lee, C. U., Manning, M. A., Cusmano-Ozog, K. P., Tise, C. G.
  2023

- **Creatine Transporter Deficiency Presenting as Failure to Thrive: A Case Report of a Novel SLC6A8 Variant Causing a Treatable but Likely Underdiagnosed Genetic Disorder.** *Journal of investigative medicine high impact case reports*
  Tise, C. G., Palma, M. J., Cusmano-Ozog, K. P., Matalon, D. R.
  2023; 11: 23247096231154438

- **Optimization of the biochemical genetics laboratory rotation using a multidesign approach to curriculum.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Bosfield, K., Albert, J., Cheng, N., Swaringer, T., Cusmano-Ozog, K., Regier, D. S.
  2022

- **Recent tPA administration can cause pseudo-hyperargininemia and may mimic arginase deficiency or arginine supplementation.** *JIMD reports*
  Cusmano-Ozog, K. P., Renck, A. K., Tise, C. G.
  2022; 63 (6): 563-567

- **Genomic and biochemical analysis of repeatedly observed variants in DBT in individuals with maple syrup urine disease of Central American ancestry.** *American journal of medical genetics. Part A*
  Billington, C. J., Chapman, K. A., Leon, E., Meltzer, B. W., Berger, S. I., Olson, M., Figler, R. A., Hoang, S. A., Wanxing, C., Wamhoff, B. R., Collado, M. S., Cusmano-Ozog, K.
  2022; 188 (9): 2738-2749

- **Measurement of lysosomal enzyme activities: A technical standard of the American College of Medical Genetics and Genomics (ACMG).** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Strovel, E. T., Cusmano-Ozog, K., Wood, T., Yu, C.
  2022; 24 (4): 769-783

- **Laboratory analysis of acylcarnitines, 2020 update: a technical standard of the American College of Medical Genetics and Genomics (ACMG).** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Miller, M. J., Cusmano-Ozog, K., Oglesbee, D., Young, S.
  2021; 23 (2): 249-258

- **Aicardi-Goutières syndrome may present with positive newborn screen for X-linked adrenoleukodystrophy.** *American journal of medical genetics. Part A*
  Tise, C. G., Morales, J. A., Lee, A. S., Velez-Bartolomei, F. n., Floyd, B. J., Levy, R. J., Cusmano-Ozog, K. P., Feigenbaum, A. S., Ruzhnikov, M. R., Lee, C. U., Enns, G. M.
  2021

- **Biallelic variants in KYNU cause a multisystemic syndrome with hand hyperphalangism.** *Bone*
  Ehmke, N., Cusmano-Ozog, K., Koenig, R., Holtgrewe, M., Nur, B., Mihci, E., Babcock, H., Gonzaga-Jauregui, C., Overton, J. D., Xiao, J., Martinez, A. F., Muenke, M., Balzer, et al
  2020; 133: 115219

- **Laboratory diagnosis of disorders of peroxisomal biogenesis and function: a technical standard of the American College of Medical Genetics and Genomics (ACMG).** *Genetics in medicine : official journal of the American College of Medical Genetics*
  De Biase, I., Tortorelli, S., Kratz, L., J Steinberg, S., Cusmano-Ozog, K., Braverman, N.

2020; 22 (4): 686-697

- **Growth, development, and phenotypic spectrum of individuals with deletions of 2q33.1 involving SATB2.** *Clinical genetics*
  Zarate, Y. A., Bosanko, K. A., Thomas, M. A., Miller, D. T., Cusmano-Ozog, K. n., Martinez-Monseny, A. n., Curry, C. J., Graham, J. M., Velsher, L. n., Bekheirnia, M. R., Seidel, V. n., Dedousis, D. n., Mitchell, et al
  2020
- **Clinical and molecular spectrum of CHOPS syndrome.** *American journal of medical genetics. Part A*
  Raible, S. E., Mehta, D., Bettale, C., Fiordaliso, S., Kaur, M., Medne, L., Rio, M., Haan, E., White, S. M., Cusmano-Ozog, K., Nishi, E., Guo, Y., Wu, et al
  2019; 179 (7): 1126-1138
- **FGF21 underlies a hormetic response to metabolic stress in methylmalonic acidemia.** *JCI insight*
  Manoli, I., Sysol, J. R., Epping, M. W., Li, L., Wang, C., Sloan, J. L., Pass, A., Gagne, J., Ktena, Y. P., Li, L., Trivedi, N. S., Ouattara, B., Zerfas, et al
  2018; 3 (23)
- **Tissue acylcarnitine status in a mouse model of mitochondrial #-oxidation deficiency during metabolic decompensation due to influenza virus infection.** *Molecular genetics and metabolism*
  Tarasenko, T. N., Cusmano-Ozog, K., McGuire, P. J.
  2018; 125 (1-2): 144-152
- **Extending the phenotypic spectrum of Sengers syndrome: Congenital lactic acidosis with synthetic liver dysfunction.** *Translational science of rare diseases*
  Beck, D. B., Cusmano-Ozog, K., Andescavage, N., Leon, E.
  2018; 3 (1): 45-48
- **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
- **Confirmation that MAT1A p.Ala259Val mutation causes autosomal dominant hypermethioninemia.** *Molecular genetics and metabolism reports*
  Muriello, M. J., Viall, S., Bottiglieri, T., Cusmano-Ozog, K., Ferreira, C. R.
  2017; 13: 9-12
- **Plasma fibroblast growth factor-21 levels in patients with inborn errors of metabolism.** *Molecular genetics and metabolism reports*
  Kirmse, B., Cabrerra-Luque, J., Ayyub, O., Cusmano, K., Chapman, K., Summar, M.
  2017; 13: 52-54
- **Correction of hyperleucinemia in MSUD patients on leucine-free dietary therapy.** *Molecular genetics and metabolism*
  Scott, A. I., Cusmano-Ozog, K., Enns, G. M., Cowan, T. M.
  2017
- **Time-dependent negative bias in plasma ammonia samples in a clinical setting.** *Clinica chimica acta; international journal of clinical chemistry*
  Ayyub, O. B., Hofherr, S., Cusmano-Ozog, K., Ah Mew, N.
  2017; 471: 126-127
- **Spurious Elevation of Multiple Urine Amino Acids by Ion-Exchange Chromatography in Patients with Prolidase Deficiency.** *JIMD reports*
  Ferreira, C. R., Cusmano-Ozog, K.
  2017; 31: 45-49
- **Diagnosis of LCHAD/TFP deficiency in an at risk newborn using umbilical cord blood acylcarnitine analysis.** *Molecular genetics and metabolism reports*
  Raval, D. B., Cusmano-Ozog, K. P., Ayyub, O., Jenevein, C., Kofman, L. H., Lanpher, B., Hauser, N., Regier, D. S.
  2017; 10: 8-10
- **Hereditary fructose intolerance mimicking a biochemical phenotype of mucolipidosis: A review of the literature of secondary causes of lysosomal enzyme activity elevation in serum.** *American journal of medical genetics. Part A*
  Ferreira, C. R., Devaney, J. M., Hofherr, S. E., Pollard, L. M., Cusmano-Ozog, K.
  2017; 173 (2): 501-509
- **The proteome of methylmalonic acidemia (MMA): the elucidation of altered pathways in patient livers.** *Molecular bioSystems*
  Caterino, M., Chandler, R. J., Sloan, J. L., Dorko, K., Cusmano-Ozog, K., Ingenito, L., Strom, S. C., Imperlini, E., Scolamiero, E., Venditti, C. P., Ruoppolo, M.
  2016; 12 (2): 566-74

- **Kupffer cells modulate hepatic fatty acid oxidation during infection with PR8 influenza.** *Biochimica et biophysica acta*
  Tarasenko, T. N., Singh, L. N., Chatterji-Len, M., Zerfas, P. M., Cusmano-Ozog, K., McGuire, P. J.
  2015; 1852 (11): 2391-401

- **What is in the can? The dilemma with dietary supplements.** *Molecular genetics and metabolism*
  D'Aco, K., Mooney, R., Cusmano-Ozog, K., Hofherr, S., Lichter-Konecki, U.
  2014; 113 (4): 239-40

- **Degree of Glutathione Deficiency and Redox Imbalance Depend on Subtype of Mitochondrial Disease and Clinical Status** *PLOS ONE*
  Enns, G. M., Moore, T., Le, A., Atkuri, K., Shah, M. K., Cusmano-Ozog, K., Niemi, A., Cowan, T. M.
  2014; 9 (6)

- **Concurrent non-ketotic hyperglycinemia and propionic acidemia in an eight year old boy.** *Molecular genetics and metabolism reports*
  Kruszka, P. S., Kirmse, B., Zand, D. J., Cusmano-Ozog, K., Spector, E., Van Hove, J. L., Chapman, K. A.
  2014; 1: 237-240

- **Thiamine pyrophosphokinase deficiency causes a Leigh Disease like phenotype in a sibling pair: identification through whole exome sequencing and management strategies.** *Molecular genetics and metabolism reports*
  Fraser, J. L., Vanderver, A., Yang, S., Chang, T., Cramp, L., Vezina, G., Lichter-Konecki, U., Cusmano-Ozog, K. P., Smpokou, P., Chapman, K. A., Zand, D. J.
  2014; 1: 66-70

- **Expansion of the TARP syndrome phenotype associated with de novo mutations and mosaicism.** *American journal of medical genetics. Part A*
  Johnston, J. J., Sapp, J. C., Curry, C., Horton, M., Leon, E., Cusmano-Ozog, K., Dobyns, W. B., Hudgins, L., Zackai, E., Biesecker, L. G.
  2014; 164A (1): 120-128

- **A rapid, sensitive method for quantitative analysis of underivatized amino acids by liquid chromatography-tandem mass spectrometry (LC-MS/MS)** *JOURNAL OF CHROMATOGRAPHY B-ANALYTICAL TECHNOLOGIES IN THE BIOMEDICAL AND LIFE SCIENCES*
  Le, A., Ng, A., Kwan, T., Cusmano-Ozog, K., Cowan, T. M.
  2014; 944: 166-174

- **Liver-directed adeno-associated virus serotype 8 gene transfer rescues a lethal murine model of citrullinemia type 1.** *Gene therapy*
  Chandler, R. J., Tarasenko, T. N., Cusmano-Ozog, K., Sun, Q., Sutton, V. R., Venditti, C. P., McGuire, P. J.
  2013; 20 (12): 1188-91

- **Targeting proximal tubule mitochondrial dysfunction attenuates the renal disease of methylmalonic acidemia** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Manoli, I., Sysol, J. R., Li, L., Houillier, P., Garone, C., Wang, C., Zerfas, P. M., Cusmano-Ozog, K., Young, S., Trivedi, N. S., Cheng, J., Sloan, J. L., Chandler, et al
  2013; 110 (33): 13552-13557

- **A new LC-MS/MS method for the clinical determination of reduced and oxidized glutathione from whole blood.** *Journal of chromatography. B, Analytical technologies in the biomedical and life sciences*
  Moore, T., Le, A., Niemi, A., Kwan, T., Cusmano-Ozog, K., Enns, G. M., Cowan, T. M.
  2013; 929: 51-55

- **Outcome of infants diagnosed with 3-methyl-crotonyl-CoA-carboxylase deficiency by newborn screening** *MOLECULAR GENETICS AND METABOLISM*
  Arnold, G. L., Salazar, D., Neidich, J. A., Suwannarat, P., Graham, B. H., Lichter-Konecki, U., Bosch, A. M., Cusmano-Ozog, K., Enns, G., Wright, E. L., Lanpher, B. C., Owen, N. N., Lipson, et al
  2012; 106 (4): 439-441

- **The role of vanin-1 and oxidative stress-related pathways in distinguishing acute and chronic pediatric ITP** *BLOOD*
  Zhang, B., Lo, C., Shen, L., Sood, R., Jones, C., Cusmano-Ozog, K., Park-Snyder, S., Wong, W., Jeng, M., Cowan, T., Engleman, E. G., Zehnder, J. L.
  2011; 117 (17): 4569-4579

- **Dopa-responsive dystonia presenting as delayed and awkward gait** *PEDIATRIC NEUROLOGY*
  Cheyette, B. N., Cheyette, S. N., Cusmano-Ozog, K., Enns, G. M.
  2008; 38 (4): 273-275

- **Progressive and symmetric supraorbital hyperostosis with bony and soft tissue overgrowth in an Ethiopian female: A newly recognized overgrowth syndrome?** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Adam, M. P., Kobori, J. A., Cusmano-Ozog, K., Chen, K. M., Hoyme, H. E.

2008; 146A (5): 543-547

- **22q13.3 deletion syndrome: A recognizable malformation syndrome associated with marked speech and language delay** *AMERICAN JOURNAL OF MEDICAL GENETICS PART C-SEMINARS IN MEDICAL GENETICS*

  Cusmano-Ozog, K., Manning, M. A., Hoyme, H. E.

  2007; 145C (4): 393-398