

Stanford



Tina Cowan

Professor of Pathology (Clinical) at the Stanford University Medical Center

CLINICAL OFFICES

- **Stanford Clinical Lab at Hillview**

3375 Hillview Ave Ste 2101

MC 5627

Palo Alto, CA 94304

Tel (650) 724-7858 **Fax** (650) 724-1567

Bio

CLINICAL FOCUS

- Pathology
- Pathology and Laboratory Medicine
- Clinical Biochemical Genetics

ACADEMIC APPOINTMENTS

- Professor - Med Center Line, Pathology
- Member, Bio-X
- Member, Maternal & Child Health Research Institute (MCHRI)

ADMINISTRATIVE APPOINTMENTS

- Board of Directors, American College of Medical Genetics and Genomics, (2015- present)
- Board of Directors, Society of Inherited Metabolic Disorders, (2015- present)
- Board of Directors, American Board of Medical Genetics, (2006-2011)
- Faculty Senate, Stanford University School of Medicine, (2005-2008)
- Laboratory Quality Assurance Committee, American College of American Genetics and Genomics, (2003-2007)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Member, American Society of Human Genetics (1986 - present)
- Member, Society for the Study of Inborn Errors of Metabolism (2002 - present)
- Member, Association of Biochemistry Educators (2013 - present)

PROFESSIONAL EDUCATION

- Board Certification: Clinical Molecular Genetics, American Board of Medical Genetics and Genomics (1990)
- PhD Training: University of California Los Angeles School of Medicine (1986) CA

- Board Certification: Medical Genetics, American Board of Medical Genetics and Genomics (1990)
- Fellowship: University of Maryland Baltimore
- B.A., UCLA , Biology (1979)
- Ph.D., UCLA , Genetics (1986)

PATENTS

- Tina Cowan, Anthony Le. "United States Patent 8,945,933 Liquid Chromatography-Mass Spectrometry Methods For Multiplexed Detection and Quantitation of Free Amino Acids", Stanford, Feb 3, 2015

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

Translational research activities are aimed at the development of new laboratory testing approaches for the diagnosis and monitoring of patients with inborn errors of metabolism. In particular, a tandem-mass spectrometry (MS/MS)-based test for glutathione, an important antioxidant, is being applied to the evaluation of patients with various metabolic disorders including mitochondrial myopathies. Results of these studies will further our understanding of the pathophysiology of metabolic diseases, with the ultimate aim of improving patient testing and developing new therapeutic strategies.

Teaching

COURSES

2018-19

- Applied Biochemistry: BIOC 200 (Aut)

2017-18

- Applied Biochemistry: BIOC 200 (Aut)

2016-17

- Applied Biochemistry: BIOC 200 (Aut)

2015-16

- Applied Biochemistry: BIOC 200 (Aut)

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (Phd Program)
- Human Genetics and Genetic Counseling (Masters Program)
- Medical Genetics (Fellowship Program)

Publications

PUBLICATIONS

- **Elevated methylmalonic acidemia (MMA) screening markers in Hispanic and preterm newborns.** *Molecular genetics and metabolism*
Peng, G., de Fontnouvelle, C. A., Enns, G. M., Cowan, T. M., Zhao, H., Scharfe, C.
2018
- **Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder** *AMERICAN JOURNAL OF HUMAN GENETICS*
Olahova, M., Yoon, W., Thompson, K., Jangam, S., Fernandez, L., Davidson, J. M., Kyle, J. E., Grove, M. E., Fisk, D. G., Kohler, J. N., Holmes, M., Dries, A. M., Huang, et al
2018; 102 (3): 494–504
- **Prenatal treatment of ornithine transcarbamylase deficiency.** *Molecular genetics and metabolism*

- Wilnai, Y., Blumenfeld, Y. J., Cusmano, K., Hintz, S. R., Alcorn, D., Benitz, W. E., Berquist, W. E., Bernstein, J. A., Castillo, R. O., Concepcion, W., Cowan, T. M., Cox, K. L., Lyell, et al
2018
- **CAP/ACMG proficiency testing for biochemical genetics laboratories: a summary of performance** *GENETICS IN MEDICINE*
Oglesbee, D., Cowan, T. M., Pasquali, M., Wood, T. C., Weck, K. E., Long, T., Palomaki, G. E., Coll Amer Pathologists, Amer Coll Med Genetics & Genomics, Biochem Mol Genetics Resource Comm
2018; 20 (1): 83–90
 - **Oncogenic KRAS Regulates Amino Acid Homeostasis and Asparagine Biosynthesis via ATF4 and Alters Sensitivity to L-Asparaginase** *CANCER CELL*
Gwinn, D. M., Lee, A. G., Briones-Martin-del-Campo, M., Conn, C. S., Simpson, D. R., Scott, A. I., Le, A., Cowan, T. M., Ruggero, D., Sweet-Cordero, E.
2018; 33 (1): 91–+
 - **Phosphorylation of MCAD selectively rescues PINK1 deficiencies in behavior and metabolism.** *Molecular biology of the cell*
Course, M. M., Scott, A. I., Schoor, C., Hsieh, C. H., Papakyrikos, A. M., Winter, D., Cowan, T. M., Wang, X.
2018
 - **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
 - **Glutathione as a Redox Biomarker in Mitochondrial Disease-Implications for Therapy.** *Journal of clinical medicine*
Enns, G. M., Cowan, T. M.
2017; 6 (5)
 - **Acylcarnitine Profiles Reflect Metabolic Vulnerability for Necrotizing Enterocolitis in Newborns Born Premature.** *journal of pediatrics*
Sylvester, K. G., Kastenberger, Z. J., Moss, R. L., Enns, G. M., Cowan, T. M., Shaw, G. M., Stevenson, D. K., Sinclair, T. J., Scharfe, C., Ryckman, K. K., Jelliffe-Pawlowski, L. L.
2017; 181: 80-85 e1
 - **A gut bacterial pathway metabolizes aromatic amino acids into nine circulating metabolites.** *Nature*
Dodd, D., Spitzer, M. H., Van Treuren, W., Merrill, B. D., Hryckowian, A. J., Higginbottom, S. K., Le, A., Cowan, T. M., Nolan, G. P., Fischbach, M. A., Sonnenburg, J. L.
2017; 551 (7682): 648–52
 - **Expanding the phenotype of hawkinsinuria: new insights from response to N-acetyl-L-cysteine.** *Journal of inherited metabolic disease*
Gomez-Ospina, N., Scott, A. I., Oh, G. J., Potter, D., Goel, V. V., Destino, L., Baugh, N., Enns, G. M., Niemi, A., Cowan, T. M.
2016; 39 (6): 821-829
 - **Treatment of methylmalonic acidemia by liver or combined liver-kidney transplantation.** *journal of pediatrics*
Niemi, A., Kim, I. K., Krueger, C. E., Cowan, T. M., Baugh, N., Farrell, R., Bonham, C. A., Concepcion, W., Esquivel, C. O., Enns, G. M.
2015; 166 (6): 1455-61 e1
 - **Treatment of Methylmalonic Acidemia by Liver or Combined Liver-Kidney Transplantation** *JOURNAL OF PEDIATRICS*
Niemi, A., Kim, I. K., Krueger, C. E., Cowan, T. M., Baugh, N., Farrell, R., Bonham, C. A., Concepcion, W., Esquivel, C. O., Enns, G. M.
2015; 166 (6): 1455-?
 - **Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway** *GENETICS IN MEDICINE*
Enns, G. M., Shashi, V., Bainbridge, M., Gambello, M. J., Zahir, F. R., Bast, T., Crimian, R., Schoch, K., Platt, J., Cox, R., Bernstein, J. A., Scavina, M., Walter, et al
2014; 16 (10): 751-758
 - **Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway.** *Genetics in medicine*
Enns, G. M., Shashi, V., Bainbridge, M., Gambello, M. J., Zahir, F. R., Bast, T., Crimian, R., Schoch, K., Platt, J., Cox, R., Bernstein, J. A., Scavina, M., Walter, et al
2014; 16 (10): 751-758
 - **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)

- **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
- **Evidence of redox imbalance in a patient with succinic semialdehyde dehydrogenase deficiency.** *Molecular genetics and metabolism reports*
Niemi, A., Brown, C., Moore, T., Enns, G. M., Cowan, T. M.
2014; 1: 129-132
- **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)
- **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
- **Liver transplantation for urea cycle disorders in pediatric patients: A single-center experience** *PEDIATRIC TRANSPLANTATION*
Kim, I. K., Niemi, A., Krueger, C., Bonham, C. A., Concepcion, W., Cowan, T. M., Enns, G. M., Esquivel, C. O.
2013; 17 (2): 158-167
- **β -Galactosidosis in Patient with Intermediate GM1 and MBD Phenotype.** *JIMD reports*
Moore, T., Bernstein, J. A., Casson-Parkin, S., Cowan, T. M.
2013; 7: 77-79
- **Increased incidence of profound biotinidase deficiency among Hispanic newborns in California** *MOLECULAR GENETICS AND METABOLISM*
Cowan, T. M., Kazerouni, N. N., Dharajiya, N., Lorey, F., Roberson, M., Hodgkinson, C., Schrijver, I.
2012; 106 (4): 485-487
- **An improved LC-MS/MS method for the detection of classic and low excretor glutaric acidemia type 1** *JOURNAL OF INHERITED METABOLIC DISEASE*
Moore, T., Le, A., Cowan, T. M.
2012; 35 (3): 431-435
- **A Novel Mutation in the HSD17B10 Gene of a 10-Year-Old Boy with Refractory Epilepsy, Choreoathetosis and Learning Disability** *PLOS ONE*
Seaver, L. H., He, X., Abe, K., Cowan, T., Enns, G. M., Sweetman, L., Philipp, M., Lee, S., Malik, M., Yang, S.
2011; 6 (11)
- **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
- **Commentary.** *Clinical chemistry*
Cowan, T. M.
2011; 57 (4): 548-549
- **Technical standards and guidelines for the diagnosis of biotinidase deficiency** *GENETICS IN MEDICINE*
Cowan, T. M., Blitzer, M. G., Wolf, B.
2010; 12 (7): 464-470
- **Mapping Gene Associations in Human Mitochondria using Clinical Disease Phenotypes** *PLOS COMPUTATIONAL BIOLOGY*
Scharfe, C., Lu, H. H., Neuenburg, J. K., Allen, E. A., Li, G., Klopstock, T., Cowan, T. M., Enns, G. M., Davis, R. W.
2009; 5 (4)
- **Clinical and Molecular Heterogeneity in Patients with the CblD Inborn Error of Cobalamin Metabolism** *JOURNAL OF PEDIATRICS*
Miousse, I. R., Watkins, D., Coelho, D., Rugar, T., Crombez, E. A., Vilain, E., Bernstein, J. A., Cowan, T., Lee-Messer, C., Enns, G. M., Fowler, B., Rosenblatt, D. S.
2009; 154 (4): 551-556
- **Inherited disorders affecting mitochondrial function are associated with glutathione deficiency and hypocitrullinemia** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*

Atkuri, K. R., Cowan, T. M., Kwan, T., Ng, A., Herzenberg, L. A., Herzenberg, L. A., Enns, G. M.
2009; 106 (10): 3941-3945

- **Management and quality assurance in the biochemical genetics laboratory.** *Current protocols in human genetics / editorial board, Jonathan L. Haines ... [et al.]*
Cowan, T. M., Strovel, E. T.
2008; Chapter 17: Unit 17 7-?
- **Acylcarnitine profile analysis** *GENETICS IN MEDICINE*
Rinaldo, P., Cowan, T. M., Matern, D.
2008; 10 (2): 151-156
- **Progressive cerebral vascular degeneration with mitochondrial encephalopathy** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
Longo, N., Schrijver, I., Vogel, H., Pique, L. M., Cowan, T. M., Pasquali, M., Steinberg, G. K., Hedlund, G. L., Ernst, S. L., Gallagher, R. C., Enns, G. M.
2008; 146A (3): 361-367
- **Importance of culturing primary lymphocytes at physiological oxygen levels** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
Atkuri, K. R., Herzenberg, L. A., Niemi, A., Cowan, T., Herzenberg, L. A.
2007; 104 (11): 4547-4552
- **Glutaryl-CoA dehydrogenase deficiency and newborn screening: Retrospective analysis of a low excretor provides further evidence that some cases may be missed** *MOLECULAR GENETICS AND METABOLISM*
Gallagher, R. C., Cowan, T. M., Goodman, S. I., Enns, G. A.
2005; 86 (3): 417-420