



Tina Cowan

Professor of Pathology (Clinical) and, by courtesy, of Pediatrics (Genetics)

CLINICAL OFFICE (PRIMARY)

- **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 and Laboratory Medicine
- Pathology
- Clinical Biochemical Genetics

ACADEMIC APPOINTMENTS

- Professor - University Medical Line, Pathology
- Professor - University Medical Line (By courtesy), Pediatrics - Medical Genetics
- Member, Bio-X
- Member, Maternal & Child Health Research Institute (MCHRI)

ADMINISTRATIVE APPOINTMENTS

- Program Director, Clinical Biochemical Genetics Fellowship, (2019- present)
- Board of Directors, Society of Inherited Metabolic Disorders, (2015- present)
- Board of Directors, American College of Medical Genetics and Genomics, (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 Biochemical/ Molecular Genetics, American Board of Medical Genetics and Genomics (1990)
- PhD Training: UCLA David Geffen School Of Medicine Registrar (1986) CA
- Fellowship: University of Maryland Baltimore (1989)
- Board Certification: Medical Genetics, American Board of Medical Genetics and Genomics (1990)
- 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

2024-25

- Applied Biochemistry: BIOC 200 (Aut)

2023-24

- Applied Biochemistry: BIOC 200 (Aut)

2022-23

- Applied Biochemistry: BIOC 200 (Aut)

2021-22

- 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

- **Biochemical genetics decoded: A comparative study of ChatGPT and IEMbase**
Zapata, L., Cowan, T.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2024
- **A Phase 1 Study of Oral Vitamin D3 in Boys and Young Men With X-Linked Adrenoleukodystrophy.** *Neurology. Genetics*

- Van Haren, K. P., Cunanan, K., Awani, A., Gu, M., Peña, D., Chromik, L. C., Považan, M., Rossi, N. C., Goodman, J., Sundaram, V., Winterbottom, J., Raymond, G. V., Cowan, et al
2023; 9 (2): e200061
- **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
 - **Host-microbe co-metabolism via MCAD generates circulating metabolites including hippuric acid.** *Nature communications*
Pruss, K. M., Chen, H., Liu, Y., Van Treuren, W., Higginbottom, S. K., Jarman, J. B., Fischer, C. R., Mak, J., Wong, B., Cowan, T. M., Fischbach, M. A., Sonnenburg, J. L., Dodd, et al
2023; 14 (1): 512
 - **Validation of a targeted metabolomics panel for improved second-tier newborn screening.** *Journal of inherited metabolic disease*
Mak, J., Peng, G., Le, A., Gandotra, N., Enns, G. M., Scharfe, C., Cowan, T. M.
2023
 - **Metabolic diversity in human populations and correlation with genetic and ancestral geographic distances.** *Molecular genetics and metabolism*
Peng, G., Pakstis, A. J., Gandotra, N., Cowan, T. M., Zhao, H., Kidd, K. K., Scharfe, C.
2022; 137 (3): 292-300
 - **MITOCHONDRIAL-ATP6-ASSOCIATED DISEASE PRESENTS WITH DISTINCT PATTERN ON NEWBORN SCREENING: SHOULD IT BE INCLUDED AS A SECONDARY CONDITION?**
Tise, C., Mendelsohn, B., Lee, C., Woods, J., Hall, P., Tang, H., Rinaldo, P., Cowan, T., Cusmano-Ozog, K.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2022: 247-248
 - **Targeted plasma metabolomics combined with machine learning for the diagnosis of severe acute respiratory syndrome virus type 2.** *Frontiers in microbiology*
Le, A. T., Wu, M., Khan, A., Phillips, N., Rajpurkar, P., Garland, M., Magid, K., Sibai, M., Huang, C., Sahoo, M. K., Bowen, R., Cowan, T. M., Pinsky, et al
2022; 13: 1059289
 - **Biochem for the Win! The added value of biochemical genetic testing for diagnosis and variant interpretation in the genomic era**
Tise, C., Grand, K., Corado, J., Gates, R., Graham, J., Enns, G., Gomez-Ospina, N., Mak, J., Cowan, T., Cusmano-Ozog, K.
ELSEVIER SCIENCE INC.2022: S24
 - **GlcNAc-Asn (GNA) is a biomarker for NGLY1 deficiency.** *Journal of biochemistry*
Mueller, W. F., Zhu, L., Tan, B., Dwight, S., Beahm, B., Wilsey, M., Wechsler, T., Mak, J., Cowan, T., Pritchett, J., Taylor, E., Crawford, B. E.
2021
 - **Variable clinical severity in TANGO2 deficiency: Case series and literature review.** *American journal of medical genetics. Part A*
Schymick, J., Leahy, P., Cowan, T., Ruzhnikov, M. R., Gates, R., Fernandez, L., Pramanik, G., Undiagnosed Diseases Network, Yariagadda, V., Wheeler, M., Bernstein, J. A., Enns, G. M., Lee, C.
2021
 - **Detecting lysosomal storage disorders by glycomic profiling using liquid chromatography mass spectrometry.** *Molecular genetics and metabolism*
Mak, J., Cowan, T. M.
2021
 - **Nasopharyngeal metabolomics and machine learning approach for the diagnosis of influenza.** *EBioMedicine*
Hogan, C. A., Rajpurkar, P., Sowrirajan, H., Phillips, N. A., Le, A. T., Wu, M., Garamani, N., Sahoo, M. K., Wood, M. L., Huang, C., Ng, A. Y., Mak, J., Cowan, et al
2021; 71: 103546
 - **Timing of newborn blood collection alters screening performance for metabolic disorders**
Peng, G., Tang, Y., Gandotra, N., Cowan, T., Zhao, H., Scharfe, C.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S26

- **A conserved role for AMP-activated protein kinase in NGLY1 deficiency.** *PLoS genetics*
Han, S. Y., Pandey, A., Moore, T., Galeone, A., Duraine, L., Cowan, T. M., Jafar-Nejad, H.
2020; 16 (12): e1009258
- **A conserved role for AMP-activated protein kinase in NGLY1 deficiency**
Han, S., Pandey, A., Moore, T., Galeone, A., Duraine, L., Cowan, T. M., Hamed, J.
OXFORD UNIV PRESS INC.2020: 1057–58
- **Exploring the mechanisms underlying the NGLY1 loss-of-function phenotypes in Drosophila**
Pandey, A., Han, S., Mueller, W. F., Story, B. A., Moore, T., Galeone, A., Cowan, T. M., Steinmetz, L.
OXFORD UNIV PRESS INC.2020: 1026
- **AAV9-Mediated Gene Therapy for NGLY1 Deficiency and Assessment of GNA Biomarker Changes in a Rat Disease Model**
Mueller, W. F., Zhu, L., Dwight, S. S., Beahm, B., Lee, K. J., Wilsey, M., Mak, J., Pollard, L., Wood, T., Cowan, T., Crawford, B., Wechsler, T.
CELL PRESS.2020: 124–25
- **Whole genome sequencing for mutation discovery in a single case of lysosomal storage disease (MPS type 1) in the dog.** *Scientific reports*
Mansour, T. A., Woolard, K. D., Vernau, K. L., Ancona, D. M., Thomasy, S. M., Sebbag, L., Moore, B. A., Knipe, M. F., Seada, H. A., Cowan, T. M.,
Aguilar, M., Titus Brown, C., Bannasch, et al
2020; 10 (1): 6558
- **Metabolic profiling by reversed-phase/ion-exchange mass spectrometry.** *Journal of chromatography. B, Analytical technologies in the biomedical and life sciences*
Le, A., Mak, J., Cowan, T. M.
2020; 1143: 122072
- **Reducing False-Positive Results in Newborn Screening Using Machine Learning.** *International journal of neonatal screening*
Peng, G., Tang, Y., Cowan, T. M., Enns, G. M., Zhao, H., Scharfe, C.
2020; 6 (1)
- **Ethnic Variability in Newborn Metabolic Screening Markers Associated with False-Positive Outcomes.** *Journal of inherited metabolic disease*
Peng, G. n., Tang, Y. n., Gandotra, N. n., Enns, G. M., Cowan, T. M., Zhao, H. n., Scharfe, C. n.
2020
- **Timing of Newborn Blood Collection Alters Metabolic Disease Screening Performance.** *Frontiers in pediatrics*
Peng, G., Tang, Y., Cowan, T. M., Zhao, H., Scharfe, C.
2020; 8: 623184
- **AMP-independent activator of AMPK for treatment of mitochondrial disorders.** *PloS one*
Moore, T., Yanes, R. E., Calton, M. A., Vollrath, D., Enns, G. M., Cowan, T. M.
2020; 15 (10): e0240517
- **LOCALIZING NEUROLOGIC FEATURES AT PRESENTATION OF VLCAD DEFICIENCY**
Leahy, P., Matalon, D., Ruzhnikov, M., Cowan, T., Enns, G.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2019: 282
- **Biomarkers of oxidative stress, inflammation, and vascular dysfunction in inherited cystathionine -synthase deficient homocystinuria and the impact of taurine treatment in a phase 1/2 human clinical trial** *JOURNAL OF INHERITED METABOLIC DISEASE*
Van Hove, J. K., Freehauf, C. L., Ficicioglu, C., Pena, L. M., Moreau, K. L., Henthorn, T. K., Christians, U., Jiang, H., Cowan, T. M., Young, S. P., Hite, M., Friederich, M. W., Stabler, et al
2019; 42 (3): 424–37
- **Combining newborn metabolic and DNA analysis for second-tier testing of methylmalonic acidemia** *GENETICS IN MEDICINE*
Peng, G., Shen, P., Gandotra, N., Le, A., Fung, E., Jelliffe-Pawlowski, L., Davis, R. W., Enns, G. M., Zhao, H., Cowan, T. M., Scharfe, C.
2019; 21 (4): 896–903
- **Biomarkers of Oxidative Stress, Inflammation, and Vascular Dysfunction in Inherited Cystathionine beta-synthase Deficient Homocystinuria and the Impact of Taurine Treatment in a Phase 1/2 human Clinical Trial.** *Journal of inherited metabolic disease*
Van Hove, J. L., Freehauf, C. L., Ficicioglu, C., Pena, L. D., Moreau, K. L., Henthorn, T. K., Christians, U., Jiang, H., Cowan, T. M., Young, S. P., Hite, M., Friederich, M. W., Stabler, et al

2019

- **LOCALIZING NEUROLOGIC FEATURES AT PRESENTATION OF VLCAD DEFICIENCY**
Leahy, P., Matalon, D., Ruzhnikov, M., Cowan, T., Enns, G.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2019: 311
- **SETD3 is an actin histidine methyltransferase that prevents primary dystocia** *NATURE*
Wilkinson, A. W., Diep, J., Dai, S., Liu, S., Ooi, Y., Song, D., Li, T., Horton, J. R., Zhang, X., Liu, C., Trivedi, D. V., Ruppel, K. M., Vilches-Moure, et al
2019; 565 (7739): 372-+
- **Medical Biochemistry Without Rote Memorization: Multi-Institution Implementation and Student Perceptions of a Nationally Standardized Metabolic Map for Learning and Assessment.** *Medical science educator*
Spicer, D. B., Thompson, K. H., Tong, M. S., Cowan, T. M., Fulton, T. B., Lindsley, J. E.
2019; 29 (1): 87-92
- **Quantitative Analysis of Underivatized Amino Acids by Liquid Chromatography-Tandem Mass Spectrometry** *AMINO ACID ANALYSIS, 2 EDITION*
Mak, J., Cowan, T. M., Le, A., Alterman, M. A.
2019; 2030: 85-109
- **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.
2019; 126 (1): 39-42
- **Quantitative Analysis of Underivatized Amino Acids by Liquid Chromatography-Tandem Mass Spectrometry.** *Methods in molecular biology (Clifton, N.J.)*
Mak, J. n., Cowan, T. M., Le, A. n.
2019; 2030: 85-109
- **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
- **Rapid Underivatized Method for Quantitative Methylmalonic Acid by Liquid Chromatography-Tandem Mass Spectrometry.** *The journal of applied laboratory medicine*
Mak, J., Wong, B. T., Kwan, T., Le, A., Cowan, T. M.
2018; 3 (3): 408-417
- **Rapid Underivatized Method for Quantitative Methylmalonic Acid by Liquid Chromatography-Tandem Mass Spectrometry** *JOURNAL OF APPLIED LABORATORY MEDICINE*
Mak, J., Wong, B. T., Kwan, T., Le, A., Cowan, T. M.
2018; 3 (3): 408-17
- **Combining newborn metabolic and DNA analysis for second-tier testing of methylmalonic acidemia.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Peng, G., Shen, P., Gandotra, N., Le, A., Fung, E., Jelliffe-Pawlowski, L., Davis, R. W., Enns, G. M., Zhao, H., Cowan, T. M., Scharfe, C.
2018
- **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., Papakyrikos, A. M., Winter, D., Cowan, T. M., Wang, X.
2018; 29 (10): 1219-1227
- **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. 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

- **SETD3 is an actin histidine methyltransferase that prevents primary dystocia.** *Nature*
Wilkinson, A. W., Diep, J. n., Dai, S. n., Liu, S. n., Ooi, Y. S., Song, D. n., Li, T. M., Horton, J. R., Zhang, X. n., Liu, C. n., Trivedi, D. V., Ruppel, K. M., Vilches-Moure, 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. n., Hsieh, C. H., Papakyrikos, A. M., Winter, D. n., Cowan, T. M., Wang, X. n.
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; 122 (4): 156–59
- **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. n., Spitzer, M. H., Van Treuren, W. n., Merrill, B. D., Hryckowian, A. J., Higginbottom, S. K., Le, A. n., Cowan, T. M., Nolan, G. P., Fischbach, M. A., Sonnenburg, J. L.
2017; 551 (7682): 648–52
- **Laboratory diagnosis of biotinidase deficiency, 2017 update: a technical standard and guideline of the American College of Medical Genetics and Genomics.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Strovel, E. T., Cowan, T. M., Scott, A. I., Wolf, B.
2017; 19 (10)
- **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
- **The antibiotic cefepime interferes with amino acid analysis by ion-exchange chromatography** *CLINICA CHIMICA ACTA*
Scott, A. I., Le, A., Cowan, T. M., Mendelsohn, B. A.
2016; 456: 149–50
- **RATE OF LEUCINE CLEARANCE IN MSUD**
Scott, A. I., Cusmano-Ozog, K., Enns, G. M., Cowan, T. M.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2016: 284

- **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-?
- **Analysis of 20 years of urinary mps proficiency data from the college of American pathogists (CAP)**
Wood, T., Cowan, T., Pasquali, M., Oglesbee, D.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2015: 346
- **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
- **Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway (vol 111, pg 236, 2014)** *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 (7): 568
- **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)
- **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., Esquivel, et al
ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: 248
- **CORRELATION OF GLUTATHIONE REDOX POTENTIAL WITH MITOCHONDRIAL DISEASE ETIOLOGY AND CLINICAL SEVERITY**
Enns, G. M., Moore, T., Le, A., Atkuri, K., Shah, M. K., Cusmano-Ozog, K., Niemi, A., Cowan, T. M.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: 268
- **MUTATIONS IN NGLY1 CAUSE AN INHERITED DISORDER OF THE ENDOPLASMIC RETICULUM-ASSOCIATED DEGRADATION (ERAD) PATHWAY**
Enns, G. M., Shashi, V., Zahir, F., Gambello, M. J., Bainbridge, M. N., Bast, T., Crimian, R., Schoch, K., Zoghbi, H. Y., Platt, J., Cox, R., Bernstein, J., Scavina, et al
ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: 236-37
- **QUANTIFICATION OF URINE KERATAN SULFATE LEVELS IN MUCOLIPIDOSIS II AND III PATIENTS BY UPLC-MS/MS**
Niemi, A., Gomez-Ospina, N., Goel, V., Destino, L., Cowan, T. M.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: 253-54
- **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
 - **The Role of Oxidative Stress in Pediatric Immune Thrombocytopenia**
Lo, C., Zhang, B., Cusmano-Ozog, K., Wong, W., Jeng, M., Cowan, T., Zehnder, J. L.
AMER SOC HEMATOLOGY.2012
 - **Improved redox status after liver transplantation in a patient with MMA mut(0) subtype; functional evidence for EPI-743 therapy**
Niemi, A., Niemi, A., Moore, T., Cowan, T., Kheifets, V., Enns, G. M.
ELSEVIER SCI LTD.2012: 557–57
 - **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
 - **Low glutathione levels in a patient with succinic semialdehyde dehydrogenase (SSADH) deficiency** *35th Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD)*
Niemi, A., Brown, C., Moore, T., Enns, G. M., Cowan, T. M.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2012: 345–45
 - **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
 - **Biomarkers of redox abnormalities in mitochondrial disorders and organic acidemias**
Atkuri, K. R., Cowan, T. M., Procaccio, V., Herzenberg, L. A., Enns, G. M.
ELSEVIER SCI LTD.2010: 206–7

- **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
- **Redox Abnormalities in Inherited Mitochondrial Diseases**
Atkuri, K. R., Moore, T., Cowan, T. M., Herzenberg, L. A., Herzenberg, L. A., Procaccio, V., Enns, G. M.
ELSEVIER SCIENCE INC.2009: S85
- **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
- **An unusual case of Pompe disease presenting as muscular dystrophy**
Cusmano-Ozog, K., Vogel, H., Cowan, T., Enns, G.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2008: S18
- **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