Stanford

Tina Cowan

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

CLINICAL OFFICE (PRIMARY)

- **Stanford Clinical Lab at Hillview**

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  Palo Alto, CA 94304

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Bio

CLINICAL FOCUS

- Pathology and Laboratory Medicine
- Pathology
- Clinical Biochemical Genetics

ACADEMIC APPOINTMENTS

- Professor-Univ Med Line, Pathology
- Professor-Univ Med 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

- PhD Training: UCLA David Geffen School Of Medicine (1986) CA
- Board Certification: Clinical Biochemical/ Molecular Genetics, American Board of Medical Genetics and Genomics (1990)
- 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 theraputic strategies.

Teaching

COURSES

2026-27

- Foundations of Medicine I: INDE 243 (Aut)
- Foundations of Medicine II: INDE 244 (Win)
- Foundations of Medicine III: INDE 245 (Spr)

2025-26

- Applied Biochemistry: BIOC 200 (Aut)

2024-25

- Applied Biochemistry: BIOC 200 (Aut)

2023-24

- 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

- **A newborn with glycogen storage disease type Ib presenting with hypoglycemia and evidence of mitochondrial dysfunction.** *Molecular genetics and metabolism reports*

Fuchs, M., Keehan, L., Ky, T., Enns, G. M., Cowan, T. M.
2026; 49: 101364

- **A newborn with glycogen storage disease type Ib presenting with hypoglycemia and evidence of mitochondrial dysfunction** *MOLECULAR GENETICS AND METABOLISM REPORTS*
  Fuchs, M., Keehan, L., Ky, T., Enns, G. M., Cowan, T. M.
  2026; 49
- **An encyclopedia of the cord blood metabolome reveals maternal-fetal interactions and disease risk.** *Cell reports. Medicine*
  Lancaster, S., Mataraso, S., Reiss, J. D., Contrepois, K., Trowbridge, C. A., Michael, B., Simms, I., Ellenberger, M., Gibson, M., Clary, M., McGuire, L., Wong, F., Canfield, et al
  2026: 102548
- **Improving newborn screening accuracy through genome sequencing, targeted metabolomics, and machine learning.** *BMC medical genomics*
  Xie, Y., Peng, G., Tikhonova, I., Enns, G., Zhao, H., Cowan, T., Scharfe, C.
  2025; 18 (1): 187
- **Comprehensive metabolomics combined with machine learning for the identification of SARS-CoV-2 and other viruses directly from upper respiratory samples.** *Journal of clinical microbiology*
  Hogan, C. A., Le, A. T., Khan, A., Su, L. D., Huang, C., Sahoo, M. K., Lo, C. -., Karim, M., Stein, K. A., Einav, S., Cowan, T. M., Pinsky, B. A.
  2025: e0204224
- **Urine organic acid analysis as a tool in evaluation for Zellweger Spectrum disorder: A retrospective study.** *Molecular genetics and metabolism*
  Chilakamarri, L., Neu, M. B., Barrick, R., Huguenin, S. M., Abdenur, J. E., Cowan, T. M., Cusmano-Ozog, K. P., Tise, C. G.
  2025; 146 (1-2): 109225
- **The emerging role of metabolomics analysis in genetic and genomic testing: A points to consider statement 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., Duque Lasio, M. L., Kennedy, A. D., McClellan, R., Oglesbee, D., Smith, W. E., Sutton, V. R., Cowan, T., ACMG Laboratory Quality Assurance Committee. Electronic address: documents@acmg.net,,
  2025: 101493
- **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, Yarlagadda, 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., Panday, 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. L. K., Freehauf, C. L., Ficicioglu, C., Pena, L. D. 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.
  edited by 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

- **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 rescuesPINK1deficiencies 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

- **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 rescuesPINK1deficiencies 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., Kastenberg, 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., Esquival, 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., Nierni, 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.
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