Stanford

Tina Cowan

Professor of Pathology (Clinical) and, by courtesy, of Pediatrics (Genetics) at the Stanford University Medical Center

CLINICAL OFFICES

- **Stanford Clinical Lab at Hillview**

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  Palo Alto, CA 94304

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

Bio

CLINICAL FOCUS

- Pathology and Laboratory Medicine
- Pathology

ACADEMIC APPOINTMENTS

- Professor - Med Center Line, Pathology
- Professor - Med Center Line (By courtesy), Pediatrics - Medical Genetics
- 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 theraputic strategies.

Teaching

COURSES

2020-21

- Applied Biochemistry: BIOC 200 (Aut)

2019-20

- Applied Biochemistry: BIOC 200 (Aut)

2018-19

- Applied Biochemistry: BIOC 200 (Aut)

2017-18

- 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

- **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., Tang, Y., Gandotra, N., Enns, G. M., Cowan, T. M., Zhao, H., Scharfe, C.
  2020

- **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

- **Quantitative Analysis of Underivatized Amino Acids by Liquid Chromatography-Tandem Mass Spectrometry.** *Methods in molecular biology (Clifton, N.J.)*
  Mak, J., Cowan, T. M., Le, 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.
  2018

- **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

- **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

- **SETD3 is an actin histidine methyltransferase that prevents primary dystocia.** *Nature*
  Wilkinson, A. W., Diep, J., Dai, S., Liu, S., Ooi, Y. S., Song, D., Li, T. M., Horton, J. R., Zhang, X., Liu, C., 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 rescuesPINK1deficiencies 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., 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., 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

- **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.
  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

- **ß-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., Rupar, 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