Stanford

Gregory Enns

Professor of Pediatrics (Genetics) at the Lucile Salter Packard Children's Hospital

Pediatrics - Medical Genetics

CLINICAL OFFICES

- **Pediatric Genetics**

  730 Welch Rd 2A

  Palo Alto, CA 94304

  Tel (650) 721-5804 **Fax** (650) 498-4555

Bio

CLINICAL FOCUS

- Biochemical Genetics
- Mitochondrial Diseases
- Clinical Genetics and Genomics

ACADEMIC APPOINTMENTS

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

PROFESSIONAL EDUCATION

- Board Certification: Clinical Genetics and Genomics, American Board of Medical Genetics and Genomics (1999)
- Residency: Children's Hospital Los Angeles Pediatric Residency (1995) CA
- Internship: Children's Hospital Los Angeles Pediatric Residency (1992) CA
- Board Certification: Clinical Biochemical Genetics, American Board of Medical Genetics and Genomics (1999)
- Medical Education: University of St Andrews (1990) Scotland
- Fellowship: UCSF Medical Center (1998) CA
- MB, ChB, University of Glasgow , Medicine (1990)
- Diploma, Medical Science, University of St. Andrews , Medicine (1987)
- BA, Pomona College , Biology (1984)

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

Research interests include novel means of diagnosing and treating mitochondrial disorders, with an emphasis on antioxidant therapy, lysosomal disorders, and newborn screening by tandem mass spectrometry. Current pursuits include the analysis of glutathione and antioxidant status in patients who have mitochondrial disorders and the development of new techniques for diagnosing these conditions.

CLINICAL TRIALS

- A Phase 1/2 Study of AEB1102 in Patients With Arginase I Deficiency, Recruiting
- Acid Lipase Replacement Investigating Safety and Efficacy (ARISE) in Participants With Lysosomal Acid Lipase Deficiency, Recruiting
- International Collaborative Gaucher Group (ICGG) Gaucher Disease Registry & Pregnancy Sub-registry, Recruiting
- Mucopolysaccharidosis I (MPS I) Registry, Recruiting
- North American Mitochondrial Disease Consortium Patient Registry and Biorepository (NAMDC), Recruiting
- Safety and Efficacy Study of EPI-743 in Children With Leigh Syndrome, Recruiting
- EPI-743 for Mitochondrial Respiratory Chain Diseases, Not Recruiting
- Responses to Influenza Vaccine in Patients With Mitochondrial Disorders (MELAS), Not Recruiting
- Short-Term Outcome of N-Carbamylglutamate in the Treatment of Acute Hyperammonemia, Not Recruiting

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (Phd Program)

Publications

PUBLICATIONS

- **Mitochondrial diseases in North America: An analysis of the NAMDC Registry.** *Neurology. Genetics*
  Barca, E., Long, Y., Cooley, V., Schoenaker, R., Emmanuele, V., DiMauro, S., Cohen, B. H., Karaa, A., Vladutiu, G. D., Haas, R., Van Hove, J. L., Scaglia, F., Parikh, et al
  2020; 6 (2): e402

- **Successful liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE).** *Molecular genetics and metabolism*
  Kripps, K., Nakayuenyongsuk, W., Shayota, B. J., Berquist, W., Gomez-Ospina, N., Esquivel, C. O., Concepcion, W., Sampson, J. B., Cristin, D. J., Jackson, W. E., Gilliland, S., Pomfret, E. A., Kueht, et al
  2020

- **Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy** *PLOS ONE*
  Kahn-Kirby, A. H., Amagata, A., Maeder, C. I., Mei, J. J., Sideris, S., Kosaka, Y., Hinman, A., Malone, S. A., Bruegger, J. J., Wang, L., Kim, V., Shrader, W. D., Hoff, et al
  2019; 14 (3)

- **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

- **Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society** *GENETICS IN MEDICINE*
  Parikh, S., Goldstein, A., Karaa, A., Koenig, M., Anselm, I., Brunel-Guitton, C., Christodoulou, J., Cohen, B. H., Dimmock, D., Enns, G. M., Falk, M. J., Feigenbaum, A., Frye, et al
  2017; 19 (12)

- **Glutathione as a Redox Biomarker in Mitochondrial Disease-Implications for Therapy.** *Journal of clinical medicine*
  Enns, G. M., Cowan, T. M.
  2017; 6 (5)

- **Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis.** *Nature communications*
  Gomez-Ospina, N., Potter, C. J., Xiao, R., Manickam, K., Kim, M., Kim, K. H., Shneider, B. L., Picarsic, J. L., Jacobson, T. A., Zhang, J., He, W., Liu, P., Knisely, et al
  2016; 7: 10713-?

- **A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency** *NEW ENGLAND JOURNAL OF MEDICINE*
  Burton, B. K., Balwani, M., Feillet, F., Baric, I., Burrow, T. A., Camarena Grande, C., Coker, M., Consuelo-Sanchez, A., Deegan, P., Di Rocco, M., Enns, G. M., Erbe, R., Ezgu, et al
  2015; 373 (11): 1010-1020

- **Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society.** *Genetics in medicine*
  Parikh, S., Goldstein, A., Koenig, M. K., Scaglia, F., Enns, G. M., Saneto, R., Anselm, I., Cohen, B. H., Falk, M. J., Greene, C., Gropman, A. L., Haas, R., Hirano, et al
  2015; 17 (9): 689-701

- **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

- **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)

- **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

- **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

- **Brain uptake of Tc99m-HMPAO correlates with clinical response to the novel redox modulating agent EPI-743 in patients with mitochondrial disease** *MOLECULAR GENETICS AND METABOLISM*
  Blankenberg, F. G., Kinsman, S. L., Cohen, B. H., Goris, M. L., Spicer, K. M., Perlman, S. L., Krane, E. J., Kheifets, V., Thoolen, M., Miller, G., Enns, G. M.
  2012; 107 (4): 690-699

- **Leigh syndrome caused by a novel m.4296G > A mutation in mitochondrial tRNA isoleucine** *MITOCHONDRION*
  Cox, R., Platt, J., Chen, L. C., Tang, S., Wong, L., Enns, G. M.
  2012; 12 (2): 258-261

- **Initial experience in the treatment of inherited mitochondrial disease with EPI-743** *MOLECULAR GENETICS AND METABOLISM*

Enns, G. M., Kinsman, S. L., Perlman, S. L., Spicer, K. M., Abdenur, J. E., Cohen, B. H., Amagata, A., Barnes, A., Kheifets, V., Shrader, W. D., Thoolen, M., Blankenberg, F., Miller, et al
2012; 105 (1): 91-102

- **High-quality DNA sequence capture of 524 disease candidate genes** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Shen, P., Wang, W., Krishnakumar, S., Palm, C., Chi, A., Enns, G. M., Davis, R. W., Speed, T. P., Mindrinos, M. N., Scharfe, C.
  2011; 108 (16): 6549-6554
- **Novel Deoxyguanosine Kinase Gene Mutations and Viral Infection Predispose Apparently Healthy Children to Fulminant Liver Failure** *JOURNAL OF PEDIATRIC GASTROENTEROLOGY AND NUTRITION*
  Shieh, J. T., Berquist, W. E., Zhang, Q., Chou, P., Wong, L. C., Enns, G. M.
  2009; 49 (1): 130-132
- **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)
- **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
- **Survival after treatment with phenylacetate and benzoate for urea-cycle disorders** *NEW ENGLAND JOURNAL OF MEDICINE*
  Enns, G. M., Berry, S. A., Berry, G. T., Rhead, W. J., Brusilow, S. W., Hamosh, A.
  2007; 356 (22): 2282-2292
- **Molecular-clinical correlations in a family with variable tissue mitochondrial DNA T8993G mutant load** *MOLECULAR GENETICS AND METABOLISM*
  Enns, G. M., Bai, R., Beck, A. E., Wong, L.
  2006; 88 (4): 364-371
- **Relationship of primary mitochondrial respiratory chain dysfunction to fiber type abnormalities in skeletal muscle** *CLINICAL GENETICS*
  Enns, G. M., Hoppel, C. L., DeArmond, S. J., Schelley, S., Bass, N., Weisiger, K., Horoupian, D., Packman, S.
  2005; 68 (4): 337-348
- **The contribution of mitochondria to common disorders** *MOLECULAR GENETICS AND METABOLISM*
  Enns, G. M.
  2003; 80 (1-2): 11-26
- **Mitochondrial respiratory chain complex I deficiency with clinical and biochemical features of long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency** *JOURNAL OF PEDIATRICS*
  Enns, G. M., Bennett, M. J., Hoppel, C. L., Goodman, S. I., Weisiger, K., Ohnstad, C., Golabi, M., Packman, S.
  2000; 136 (2): 251-254
- **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
- **Hypoglycemia in CDG patients due to PMM2 mutations: Follow up on hyperinsulinemic patients.** *JIMD reports*
  Moravej, H., Altassan, R., Jaeken, J., Enns, G. M., Ellaway, C., Balasubramaniam, S., De Lonlay, P., Coman, D., Mercimek-Andrews, S., Witters, P., Morava, E.
  2020; 51 (1): 76–81
- **Clinical Spectrum and Functional Consequences Associated with Bi-Allelic Pathogenic PNPT1 Variants.** *Journal of clinical medicine*
  Rius, R., Van Bergen, N. J., Compton, A. G., Riley, L. G., Kava, M. P., Balasubramaniam, S., Amor, D. J., Fanjul-Fernandez, M., Cowley, M. J., Fahey, M. C., Koenig, M. K., Enns, G. M., Sadedin, et al
  2019; 8 (11)

- **De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Mirzaa, G. M., Chong, J. X., Piton, A., Popp, B., Foss, K., Guo, H., Harripaul, R., Xia, K., Scheck, J., Aldinger, K. A., Sajan, S. A., Tang, S., Bonneau, et al
  2019
- **Case series of sebelipase alfa hypersensitivity reactions and successful sebelipase alfa rapid desensitization.** *JIMD reports*
  Huffaker, M. F., Liu, A. Y., Enns, G. M., Vijay, S., Amor, A. J., Adkinson, N. F.
  2019; 49 (1): 30–36
- **Assessing the strength of evidence for genes implicated in fatty acid oxidation disorders using the ClinGen clinical validity framework.** *Molecular genetics and metabolism*
  McGlaughon, J. L., Pasquali, M., Wallace, K., Ross, J., Senol-Cosar, O., Shen, W., Weaver, M. A., Feigenbaum, A., Lyon, E., Enns, G. M., Mao, R., Baudet, H. G.
  2019
- **STANDARDIZING METABOLIC DISEASE GENE AND VARIANT CURATION: THE CLINGEN INBORN ERRORS OF METABOLISM WORKING GROUP**
  Zastrow, D., Baudet, H., Thomas, A., Shen, W., Si, C., Weaver, M., Liu, J., Goldstein, J., Thaxton, C., Ross, J., Crowley, S., Kurtz, C., McGlaughon, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2019: 313
- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Kumar, A., Zastrow, D. B., Kravets, E. J., Beleford, D., Ruzhnikov, M. Z., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Mackenzie, K. M., Eng, et al
  2019; 179 (6): 966–77
- **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
- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing** *JOURNAL OF GENETIC COUNSELING*
  Zastrow, D. B., Kohler, J. N., Bonner, D., Reuter, C. M., Fernandez, L., Grove, M. E., Fisk, D. G., Yang, Y., Eng, C. M., Ward, P. A., Bick, D., Worthey, E. A., Fisher, et al
  2019; 28 (2): 213–28
- **STANDARDIZING METABOLIC DISEASE GENE AND VARIANT CURATION: THE CLINGEN INBORN ERRORS OF METABOLISM WORKING GROUP**
  Zastrow, D., Baudet, H., Thomas, A., Shen, W., Si, C., Weaver, M., Liu, J., Goldstein, J., Thaxton, C., Ross, J., Crowley, S., Kurtz, C., McGlaughon, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2019: 336–40
- **Perspectives on urea cycle disorder management: Results of a clinician survey.** *Molecular genetics and metabolism*
  Enns, G. M., Porter, M. H., Francis-Sedlak, M., Burdett, A., Vockley, J.
  2019
- **Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy.** *PloS one*
  Kahn-Kirby, A. H., Amagata, A., Maeder, C. I., Mei, J. J., Sideris, S., Kosaka, Y., Hinman, A., Malone, S. A., Bruegger, J. J., Wang, L., Kim, V., Shrader, W. D., Hoff, et al
  2019; 14 (3): e0214250
- **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
- **Conducting an investigator-initiated randomized double-blinded intervention trial in acute decompensation of inborn errors of metabolism: Lessons from the N-Carbamylglutamate Consortium.** *Translational science of rare diseases*
  Ah Mew, N., Cnaan, A., McCarter, R., Choi, H., Glass, P., Rice, K., Scavo, L., Gillespie, C. W., Diaz, G. A., Berry, G. T., Wong, D., Konczal, L., McCandless, et al
  2018; 3 (3-4): 157–70
- **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)

- **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
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- **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.
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- **Unique aspects of sequence variant interpretation for inborn errors of metabolism (IEM): The ClinGen IEM Working Group and the Phenylalanine Hydroxylase Gene** *HUMAN MUTATION*
  Zastrow, D. B., Baudet, H., Shen, W., Thomas, A., Si, Y., Weaver, M. A., Lager, A. M., Liu, J., Mangels, R., Dwight, S. S., Wright, M. W., Dobrowolski, S. F., Eilbeck, et al
  2018; 39 (11): 1569–80

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  Zastrow, D. B., Baudet, H., Shen, W., Thomas, A., Si, Y., Weaver, M. A., Lager, A. M., Liu, J., Mangels, R., Dwight, S. S., Wright, M. W., Dobrowolski, S. F., Eilbeck, et al
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  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

- **Current clinical evidence does not support a link between TBL1XR1 and Rett syndrome: Description of one patient with Rett features and a novel mutation in TBL1XR1, and a review of TBL1XR1 phenotypes** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Zaghlula, M., Glaze, D. G., Enns, G. M., Potocki, L., Schwabe, A. L., Suter, B.
  2018; 176 (7): 1683–87

- **Mitochondrial disease phenotypes of 999 patients in the North American Mitochondrial Disease Consortium (NAMDC)**
  Barca, E., Cooley, V., Schoenaker, R., Emmanuele, V., DiMauro, S., Cohen, B., Karaa, A., Vladutiu, G., Haas, R., Haas, R., Van Hove, J., Scaglia, F., Parikh, et al
  LIPPINCOTT WILLIAMS & WILKINS.2018

- **PERSPECTIVES ON UREA CYCLE DISORDER MANAGEMENT: RESULTS OF A CLINICIAN SURVEY**
  Burdett, A., Francis-Sedlak, M., Vockley, J., Enns, G. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2018: 221–22

- **A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network.** *The Journal of pediatrics*
  Reuter, C. M., Brimble, E., DeFilippo, C., Dries, A. M., Enns, G. M., Ashley, E. A., Bernstein, J. A., Fisher, P. G., Wheeler, M. T.
  2018

- **Triheptanoin: A Rescue Therapy for Cardiogenic Shock in Carnitine-acylcarnitine Translocase Deficiency** *JIMD REPORTS, VOL 39*
  Mahapatra, S., Ananth, A., Baugh, N., Damian, M., Enns, G. M., Morava, E., Baumgartner, M., Patterson, M., Rahman, S., Zschocke, J., Peters
  2018; 39: 19–23

- **Triheptanoin: A Rescue Therapy for Cardiogenic Shock in Carnitine-acylcarnitine Translocase Deficiency.** *JIMD reports*
  Mahapatra, S., Ananth, A., Baugh, N., Damian, M., Enns, G. M.
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- **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
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- **Management of ophthalmologic manifestations of mitochondrial diseases Response** *GENETICS IN MEDICINE*
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- **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

- **Pediatric mitochondrial diseases and the heart** *CURRENT OPINION IN PEDIATRICS*
  Enns, G. M.
  2017; 29 (5): 541–51

- **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

- **Pathogenic variants in HTRA2 cause an early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria** *JOURNAL OF INHERITED METABOLIC DISEASE*
  Olahova, M., Thompson, K., Hardy, S. A., Barbosa, I. A., Besse, A., Anagnostou, M., White, K., Davey, T., Simpson, M. A., Champion, M., Enns, G., Schelley, S., Lightowlers, et al
  2017; 40 (1): 121-130

- **Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society.**
  Parikh, S., Goldstein, A., Karaa, A., Koenig, M. K., Anselm, I., Brunel-Guitton, C., Christodoulou, J., Cohen, B. H., Dimmock, D., Enns, G. M., Falk, M. J., Feigenbaum, A., Frye, et al
  2017

- **De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability.** *American journal of human genetics*
  Küry, S., van Woerden, G. M., Besnard, T., Proietti Onori, M., Latypova, X., Towne, M. C., Cho, M. T., Prescott, T. E., Ploeg, M. A., Sanders, S., Stessman, H. A., Pujol, A., Distel, et al
  2017; 101 (5): 768–88

- **An Open-Label, Dose-Escalation Study to Assess the Safety, Tolerability, Efficacy, Pharmacokinetics and Pharmacodynamics of Cysteamine Bitartrate Delayed-release Capsules (RP103) for Treatment of Children with Inherited Mitochondrial Disease (RP103-MITO-001)**
  Cohen, B. H., Enns, G. M., Haas, R., Longo, N., Scaglia, F., Lang, W., Sile, S.
  ELSEVIER SCI LTD.2016: 117

- **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.
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- **Triheptanoin treatment in patients with pediatric cardiomyopathy associated with long chain-fatty acid oxidation disorders.** *Molecular genetics and metabolism*
  Vockley, J., Charrow, J., Ganesh, J., Eswara, M., DIAZ, G. A., McCracken, E., Conway, R., Enns, G. M., Starr, J., Wang, R., Abdenur, J. E., Sanchez-de-Toledo, J., Marsden, et al
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