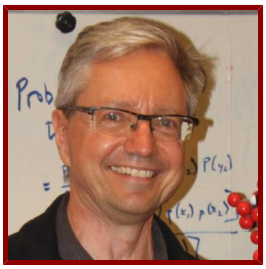
Stanford



Gregory Enns

Professor of Pediatrics (Genetics)

Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

- **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
- Medical Biochemical Genetics

ACADEMIC APPOINTMENTS

- Professor-Univ Med 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 (2020)
- Residency: Children's Hospital Los Angeles Pediatric Residency (1995) 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

- 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
- Acid Lipase Replacement Investigating Safety and Efficacy (ARISE) in Participants With Lysosomal Acid Lipase Deficiency, Not Recruiting
- EPI-743 for Mitochondrial Respiratory Chain Diseases, Not Recruiting
- Responses to Influenza Vaccine in Patients With Mitochondrial Disorders (MELAS), Not Recruiting
- Safety and Efficacy Study of EPI-743 in Children With Leigh Syndrome, Not Recruiting

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (PhD Program)

Publications

PUBLICATIONS

- **Consensus Guidelines for Diagnosis and Management of Pyruvate Dehydrogenase Complex Deficiency.** *Journal of inherited metabolic disease*
  Keshavan, N., Neugebauer, J., Bellusci, M., Bertini, E., Brown, G., Darin, N., DeBrosse, S., Drexler, L., Drexler, S., Enns, G. M., Ganetzky, R. D., Gilbert, K., Larson, et al
  2026; 49 (6): e70226

- **Phase III trial of sodium dichloroacetate for pyruvate dehydrogenase complex deficiency in children.** *JCI insight*
  Stacpoole, P. W., Abdenur, J. E., Bedoyan, J. K., Botto, L., Enns, G. M., Falk, M. J., Ganetzky, R., Garganta, C., Glinton, K., Gropman, A., Hamm, S., Henry, E., Longo, et al
  2026; 11 (18)

- **Comprehensive functional testing in fibroblasts has strong utility to diagnose mitochondrial disease.** *EMBO molecular medicine*
  Van Hove, J. L., Friederich, M. W., Van Hove, R. A., Lee, J. C., Knight, K. M., Donovan, T. E., Silveira, L., Ganetzky, R. D., Hirano, M., Abdenur, J., Butler, M. G., Cassiman, D., Cohen, et al
  2026

- **Efficacy and safety of pegzilarginase in arginase 1 deficiency (PEACE): a phase 3, randomized, double-blind, placebo-controlled, multi-centre trial.** *EClinicalMedicine*
  Russo, R. S., Gasperini, S., Bubb, G., Neuman, L., Sloan, L. S., Diaz, G. A., Enns, G. M.
  2024; 68: 102405

- **Efficacy and Safety of Elamipretide in Individuals With Primary Mitochondrial Myopathy: The MMPOWER-3 Randomized Clinical Trial.** *Neurology*
  Karaa, A., Bertini, E., Carelli, V., Cohen, B. H., Enns, G. M., Falk, M. J., Goldstein, A., Gorman, G. S., Haas, R., Hirano, M., Klopstock, T., Koenig, M. K., Kornblum, et al
  2023

- **The Present and Future of Mitochondrial-Based Therapeutics for Eye Disease.** *Translational vision science & technology*
  Ji, M. H., Kreymerman, A., Belle, K., Ghiam, B. K., Muscat, S. P., Mahajan, V. B., Enns, G. M., Mercola, M., Wood, E. H.
  2021; 10 (8): 4

- **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

- **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

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

- **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

- **RNU4ATAC-opathy: Clinical, molecular and transcriptomic insights from a large cohort.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Matalon, D. R., Duker, A. L., Arriaga, T. M., Russell, K., Mendez, H. R., Bonner, D. E., Harley, M. E., Singer-Berk, M., Wojcik, M. H., Pais, L., DiTroia, S., O'Leary, M., Cassini, et al
  2026: 102633

- **Refractory lactic acidosis in methylmalonic acidemia unmasking thiamine deficiency: A case report and call for routine thiamine assessment**
  Chilakamarri, L., Enns, G., Wright, J., Lee, C.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2026

- **A multi-center retrospective chart analysis identifying clinical, biochemical, and radiographic signatures of short-chain Enoyl CoA hydratase (ECHS1) deficiency**
  Kong, C., Alves, C., Anselm, I., Berry, G. T., Cabello, J., Cornejo, V., Fernandez, A., Enns, G., Gomez, M., Haldeman-Englert, C., Hollander, S., Innes, M., Jannotta, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2026

- **Revisiting LSDMCA: male lethality escape and genotype-phenotype correlations** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  D'Alessio, A., Indrieri, A., Vitiello, G., Morleo, M., Schelley, S., Enns, G. M., Passarelli, C., Tammaro, R., Tiranti, V., Peron, C., Deb, W., Novelli, A., Isidor, et al
  2026

- **Emerging neurological and cognitive symptoms in patients with late-onset ornithine transcarbamylase deficiency: a narrative review.** *Metabolic brain disease*
  Konczal, L., Enns, G. M., Gropman, A. L., Garcia, D., Merritt Ii, J. L., Wilkening, G.
  2026; 41 (1)

- **Clinical Characteristics of Arginase 1 Deficiency: Natural History Insights From International Clinical Trials.** *Journal of inherited metabolic disease*
  Rudebeck, M., Braverman, N., Chang, R., Enns, G. M., Ghosh, A., Gorce, M., Karall, D., Sharma, R., Shelkowitz, E., Zori, R., McNutt, M.
  2026; 49 (2): e70156

- **Clinical and Genotypic Spectrum of Twinkle-Related Disorders: Insights From a Multinational Cohort Study.** *Neurology*
  Lopriore, P., Ünlütürk, Z., Klopstock, T., Karaa, A., Rouzier, C., Domínguez-González, C., Lamperti, C., Mancuso, M., Cecchi, G., Montano, V., Siciliano, G., Nicoletta, V., Maioli, et al
  2026; 106 (3): e214401

- **Carbonic anhydrase VA deficiency due to a novel CA5A variant** *MOLECULAR GENETICS AND METABOLISM REPORTS*
  Keehan, L., Null, E., Chilakamarri, L., Carter, C., Lee, C., Enns, G. M., Matalon, D. R.
  2025; 45

- **Carbonic anhydrase VA deficiency due to a novel CA5A variant.** *Molecular genetics and metabolism reports*
  Keehan, L., Null, E., Chilakamarri, L., Carter, C., Lee, C., Enns, G. M., Matalon, D. R.

2025; 45: 101259

- **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

- **Long-Term Efficacy and Tolerability of Pegzilarginase in Arginase 1 Deficiency: Results of Two International Multicentre Open-Label Extension Studies.** *Journal of inherited metabolic disease*
  McNutt, M., Rutsch, F., Russo, R. S., Gasperini, S., Batzios, S., Teles, E. L., Brassier, A., Ganesh, J., Schulze, A., Enns, G. M., Rudebeck, M.
  2025; 48 (4): e70066

- **Genetic analysis and multimodal imaging confirm m.12148 T > C mitochondrial variant pathogenicity leading to multisystem dysfunction.** *Molecular genetics and metabolism*
  Belle, K., Kreymerman, A., Young, J. L., Vadgama, N., Ji, M. H., Randhawa, S., Caicedo, J., Wong, M., Muscat, S. P., Gifford, C. A., Lee, R. T., Nasir, J., Enns, et al
  2025; 144 (3): 109049

- **Newborn Screening for X-Linked Adrenoleukodystrophy (X-ALD): Biochemical, Molecular, and Clinical Characteristics of Other Genetic Conditions.** *Genes*
  Mares Beltran, C. F., Tise, C. G., Barrick, R., Niehaus, A. D., Sponberg, R., Chang, R., Enns, G. M., Abdenur, J. E.
  2024; 15 (7)

- **High prevalence of hepatic fibrosis in liver tissue from individuals with argininosuccinate lyase deficiency**
  Burrage, L., Ali, S., Nisar, A., Quan, T., Aceves, N., Shneider, B., Leung, D., Nagamani, S., Enns, G., Jain, S., Goss, J., Hazard, F., Schady, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2024

- **Cardiac transplantation in pediatric patients with inborn errors of metabolism: Outcomes from a single center**
  Null, E., Tolani, D., Rosenthal, D. N., Enns, G. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2024

- **Characteristics and management of US pediatric patients with Gaucher disease from the Gaucher Outcome Survey**
  Goker-Alpan, O., Burrow, A., Enns, G. M., Ficicioglu, C., Kishnani, P. S., Ortiz, D., Prada, C. E., Vats, D., Weinreb, N. J., Robinson, I., Pathak, R. R., Wright, E., Botha, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2024

- **Specifications of the ACMG/AMP guidelines for ACADVL variant interpretation.** *Molecular genetics and metabolism*
  Flowers, M., Dickson, A., Miller, M. J., Spector, E., Enns, G. M., Baudet, H., Pasquali, M., Racacho, L., Sadre-Bazzaz, K., Wen, T., Fogarty, M., Fernandez, R., Weaver, et al
  2023; 140 (3): 107668

- **Contributions from medical geneticists in clinical trials of genetic therapies: 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*
  Peña, L. D., Burrage, L. C., Enns, G. M., Esplin, E. D., Harding, C., Mendell, J. R., Niu, Z. N., Scharfe, C., Yu, T., Koeberl, D. D.
  2023: 100831

- **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

- **Outcomes after liver transplantation in MPV17 deficiency: A rebuttal.** *Pediatric transplantation*
  Huang, A. C., Ebel, N. H., Romero, D., Enns, G. M., Esquivel, C. O., Bonham, C.
  2023: e14472

- **BIOCHEMICAL, MOLECULAR, AND CLINICAL CHARACTERISTICS OF PEROXISOMAL DISORDERS DETECTED BY CALIFORNIA NEWBORN SCREENING (NBS) PROGRAM**

Beltran, C., Abdenur, J., Chang, R., Barrick, R., Spongberg, R., Tise, C. G., Niehaus, A. D., Enns, G.
ACADEMIC PRESS INC ELSEVIER SCIENCE.2023: 72

- **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
- **Fractionated plasma N-glycan profiling of novel cohort of ATP6AP1-CDG subjects identifies phenotypic association.** *Journal of inherited metabolic disease*
  Alharbi, H., Daniel, E. J., Thies, J., Chang, I., Goldner, D. L., Ng, B. G., Witters, P., Aqul, A., Velez-Bartolomei, F., Enns, G. M., Hsu, E., Kichula, E., Lee, et al
  2023
- **Outcomes after liver transplantation in MPV17 deficiency (Navajo neurohepatopathy): A single-center case series.** *Pediatric transplantation*
  Huang, A. C., Ebel, N. H., Romero, D., Martin, B., Jhun, I., Brown, M., Enns, G. M., Esquivel, C., Bonham, C.
  2022: e14274
- **Response to triheptanoin therapy in critically ill patients with LC-FAOD: Report of patients treated through an expanded access program.** *Molecular genetics and metabolism*
  Vockley, J., Enns, G. M., Ramirez, A. N., Bedrosian, C. L., Reineking, B., Lu, X., Ray, K., Rahman, S., Marsden, D.
  2022
- **SUCCESSFUL CARDIAC TRANSPLANTATION AND LONG-TERM FOLLOW-UP IN DNAJC19-ASSOCIATED DILATED CARDIOMYOPATHY WITH ATAXIA**
  Tahata, S., Tise, C., Floyd, B., Cusmano-Ozog, K., Ruzhnikov, M., Enns, G.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2022: 302
- **PEGZILARGINASE IN ARGINASE 1 DEFICIENCY: RESULTS OF THE PEACE PIVOTAL PHASE 3 CLINICAL TRIAL**
  Enns, G. M., Russo, R., Bradford, E., Bubb, G., Sloan, L. S., Gasperini, S., Diaz, G. A.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2022: 269
- **NEUROIMAGING FINDINGS IN INBORN ERRORS OF METABOLISM: ATYPICAL FINDINGS FROM FIVE CASES AFFECTED BY SMALL MOLECULE DISORDERS**
  Morales, J., Dahmoush, H., Enns, G.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2022: 251-252
- **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
- **Clinical characterization of a new individual with mild SC4MOL deficiency: diagnostic and therapeutic implications** *JOURNAL OF TRANSLATIONAL GENETICS AND GENOMICS*
  Morales, J., Curry, C. J., Tise, C. G., Kratz, L., Enns, G. M.
  2022; 6 (2): 257-265
- **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
- **Diagnostic challenges and disease management in patients with a mild Zellweger spectrum disorder phenotype.** *Molecular genetics and metabolism*
  Enns, G. M., Ammous, Z., Himes, R. W., Nogueira, J., Palle, S., Sullivan, M., Ramirez, C.
  2021
- **Profound neonatal lactic acidosis and renal tubulopathy in a patient with glycogen storage disease type IXα2 secondary to a de novo pathogenic variant in PHKA2.** *Molecular genetics and metabolism reports*
  Morales, J. A., Tise, C. G., Narang, A., Grimm, P. C., Enns, G. M., Lee, C. U.
  2021; 27: 100765
- **Profound neonatal lactic acidosis and renal tubulopathy in a patient with glycogen storage disease type IXa2 secondary to a de novo pathogenic variant in PHKA2** *MOLECULAR GENETICS AND METABOLISM REPORTS*

Morales, J., Tise, C. G., Narang, A., Grimm, P. C., Enns, G. M., Lee, C. U.
2021; 27

- **Compound heterozygous KCTD7 variants in progressive myoclonus epilepsy.** *Journal of neurogenetics*
  Burke, E. A., Sturgeon, M., Zastrow, D. B., Fernandez, L., Prybol, C., Marwaha, S., Frothingham, E. P., Ward, P. A., Eng, C. M., Fresard, L., Montgomery, S. B., Enns, G. M., Fisher, et al
  2021: 1–10

- **A resource of lipidomics and metabolomics data from individuals with undiagnosed diseases** *SCIENTIFIC DATA*
  Kyle, J. E., Stratton, K. G., Zink, E. M., Kim, Y., Bloodsworth, K. J., Monroe, M. E., Bacino, C. A., Bacino, C. A., Hanchard, N. A., Lewis, R. A., Rosenfeld, J. A., Scott, D. A., Tran, et al
  2021; 8 (1): 114

- **Unexpected diagnoses in patients with abnormal newborn screening**
  Tise, C., Velez-Bartolomei, F., Morales, J., Lee, C., Bernstein, J., Enns, G.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S354

- **Clinical outcomes of major clinical events and emergency triheptanoin use in critically ill patients with long-chain fatty acid oxidation disorders**
  Enns, G., Marsden, D., Ramirez, A., Bedrosian, C., Reineking, B., Lu, X., Rahman, S., Vockley, J.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S86-S87

- **Tailoring the ACMG/AMP sequence variant interpretation guidelines to the unique aspects of germline ACADVL variants**
  Flowers, M., Weaver, M., Baudet, H., Pasquali, M., Enns, G., Feigenbaum, A., Lyon, E., Miller, M., Graham, B., Spector, E., Racacho, L., McLachlan, M., Sadre-Bazzaz, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S254

- **Retrospective study of the disease course in pediatric patients with severe MMA caused by MMUT mutations: design and baseline characteristics**
  Enns, G., Baker, P., Li, H., Morgan, T., Vockley, J.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S23-S25

- **Refining ClinGen loss of function variant recommendations for the phenylalanine hydroxylase (PAH) gene: the PAH variant curation expert panel's experience**
  Groopman, E., Weaver, M., Zastrow, D., Enns, G., Feigenbaum, A., Lichter-Konecki, U., Lyon, E., Pasquali, M., Caldovic, L., Ross, J., Craigen, W.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S231

- **Aicardi-Goutières syndrome may present with positive newborn screen for X-linked adrenoleukodystrophy.** *American journal of medical genetics. Part A*
  Tise, C. G., Morales, J. A., Lee, A. S., Velez-Bartolomei, F. n., Floyd, B. J., Levy, R. J., Cusmano-Ozog, K. P., Feigenbaum, A. S., Ruzhnikov, M. R., Lee, C. U., Enns, G. M.
  2021

- **PERIOPERATIVE MANAGEMENT AND POST-LIVER TRANSPLANTATION OUTCOMES IN UREA CYCLE DISORDERS, MAPLE SYRUP URINE DISEASE AND ORGANIC ACIDEMIAS: 20 YEARS' EXPERIENCE**
  Ebel, N., Vuong, P., Baker, C., Brubaker, A., Than, P., Enns, G., Esquivel, C. O.
  WILEY.2020: 881A–882A

- **PERIOPERATIVE MANAGEMENT AND POST-LIVER TRANSPLANTATION OUTCOMES IN METHYLMALONIC ACIDEMIA, PROPIONIC ACIDEMIA AND UREA CYCLE DISORDERS: OUR 20 YEAR EXPERIENCE**
  Vuong, P., Ebel, N., Baker, C. V., Brubaker, A., Than, P., Enns, G., Esquivel, C. O.
  LIPPINCOTT WILLIAMS & WILKINS.2020: S547

- **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

- **Clinical effect and safety profile of pegzilarginase in patients with arginase 1 deficiency.** *Journal of inherited metabolic disease*
  Diaz, G. A., Schulze, A. n., McNutt, M. C., Leão-Teles, E. n., Merritt, J. L., Enns, G. M., Batzios, S. n., Bannick, A. n., Zori, R. T., Sloan, L. S., Potts, S. L., Bubb, G. n., Quinn, et al
  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. R. 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

- **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

- **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. n., Burdett, A. n., Vockley, J. n.
  2019
- **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
  2018; 3 (23)
- **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
- **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
- **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
- **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
- **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. n., DeFilippo, C. n., 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.
  edited by 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.
  2018; 39: 19–23

- **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

- **Management of ophthalmologic manifestations of mitochondrial diseases Response** *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)

- **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. n., van Woerden, G. M., Besnard, T. n., Proietti Onori, M. n., Latypova, X. n., Towne, M. C., Cho, M. T., Prescott, T. E., Ploeg, M. A., Sanders, S. n., Stessman, H. A., Pujol, A. n., Distel, et al
  2017; 101 (5): 768–88

- **Nutritional interventions in primary mitochondrial disorders: Developing an evidence base.** *Molecular genetics and metabolism*
  Camp, K. M., Krotoski, D., Parisi, M. A., Gwinn, K. A., Cohen, B. H., Cox, C. S., Enns, G. M., Falk, M. J., Goldstein, A. C., Gopal-Srivastava, R., Gorman, G. S., Hersh, S. P., Hirano, et al
  2016; 119 (3): 187-206

- **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.
  2016; 39 (6): 821-829

- **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
  2016; 119 (3): 223-231

- **De Novo Mutations in CHD4, an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Weiss, K., Terhal, P. A., Cohen, L., Bruccoleri, M., Irving, M., Martinez, A. F., Rosenfeld, J. A., Machol, K., Yang, Y., Liu, P., Walkiewicz, M., Beuten, J., Gomez-Ospina, et al
  2016; 99 (4): 934-941

- **Impaired Health-Related Quality of Life in Children and Families Affected by Methylmalonic Acidemia.** *Journal of genetic counseling*
  Splinter, K., Niemi, A., Cox, R., Platt, J., Shah, M., Enns, G. M., Kasahara, M., Bernstein, J. A.
  2016; 25 (5): 936-944

- **Living donor liver transplantation for inborn errors of metabolism - An underutilized resource in the United States.** *Pediatric transplantation*
  Pham, T. A., Enns, G. M., Esquivel, C. O.
  2016; 20 (6): 770-773

- **The M405V allele of the glutaryl-CoA dehydrogenase gene is an important marker for glutaric aciduria type I (GA-I) low excretors.** *Molecular genetics and metabolism*
  Schillaci, L. P., Greene, C. L., Strovel, E., Rispoli-Joines, J., Spector, E., Woontner, M., Scharer, G., Enns, G. M., Gallagher, R., Zinn, A. B., McCandless, S. E., Hoppel, C. L., Goodman, et al
  2016; 119 (1-2): 50-56

- **Prenatally Diagnosed Cases of Binder Phenotype Complicated by Respiratory Distress in the Immediate Postnatal Period.** *Journal of ultrasound in medicine*
  Blumenfeld, Y. J., Davis, A. S., Hintz, S. R., Milan, K., Messner, A. H., Barth, R. A., Hudgins, L., Chueh, J., Homeyer, M., Bernstein, J. A., Enns, G., Atwal, P., Manning, et al
  2016; 35 (6): 1353-1358

- **Clinical Course of Six Children With GNAO1 Mutations Causing a Severe and Distinctive Movement Disorder** *PEDIATRIC NEUROLOGY*
  Ananth, A. L., Robichaux-Viehoever, A., Kim, Y., Hanson-Kahn, A., Cox, R., Enns, G. M., Strober, J., Willing, M., Schlaggar, B. L., Wu, Y. W., Bernstein, J. A.
  2016; 59: 81-84

- **Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Petrovski, S., Kury, S., Myers, C. T., Anyane-Yeboa, K., Cogne, B., Bialer, M., Xia, F., Hemati, P., Riviello, J., Mehaffey, M., Besnard, T., Becraft, E., Wadley, et al
  2016; 98 (5): 1001-1010

- **Rapid progression and mortality of lysosomal acid lipase deficiency presenting in infants** *GENETICS IN MEDICINE*
  Jones, S. A., Valayannopoulos, V., Schneider, E., Eckert, S., Banikazemi, M., Bialer, M., Cederbaum, S., Chan, A., Dhawan, A., Di Rocco, M., Domm, J., Enns, G. M., Finegold, et al
  2016; 18 (5): 452-458

- **Line intensities and temperature-dependent line broadening coefficients of Q-branch transitions in the v2 band of ammonia near 10.4 μm.** *Journal of quantitative spectroscopy & radiative transfer*
  Sur, R., Spearrin, R. M., Peng, W. Y., Strand, C. L., Jeffries, J. B., Enns, G. M., Hanson, R. K.
  2016; 175: 90-99

- **Line intensities and temperature-dependent line broadening coefficients of Q-branch transitions in the v(2) band of ammonia near 10.4 mu m** *JOURNAL OF QUANTITATIVE SPECTROSCOPY & RADIATIVE TRANSFER*

Sur, R., Spearrin, R. M., Peng, W. Y., Strand, C. L., Jeffries, J. B., Enns, G. M., Hanson, R. K.
2016; 175: 90-99

- **THE M405V ALLELE OF THE GLUTARYL-CoA DEHYDROGENASE GENE IS COMMON AMONG MANY GLUTARIC ACIDURIA TYPE I (GA-I) LOW EXCRETORS**
  Schillaci, L., Greene, C., Strovel, E., Rispoli-Joines, J., Spector, E., Woontner, M., Scharer, G., Enns, G., Gallagher, R., Zinn, A., McCandless, S., Hoppel, C., Goodman, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2016: 283

- **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

- **OPA1-PLUS SYNDROMES MAY INCLUDE CARDIOMYOPATHY - EXPANDING THE PHENOTYPE**
  Myers, A., Goyal, N., Vagelos, R., Zei, P. C., Enns, G. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2016: 273–74

- **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., Shneider, B. L., Picarsic, J. L., Jacobson, T. A., Zhang, J., He, W., Liu, P., Knisely, et al
  2016; 7

- **Clinical Features of Lysosomal Acid Lipase Deficiency.** *Journal of pediatric gastroenterology and nutrition*
  Burton, B. K., Deegan, P. B., Enns, G. M., Guardamagna, O., Horslen, S., Hovingh, G. K., Lobritto, S. J., Malinova, V., Mclin, V. A., Raiman, J., Di Rocco, M., Santra, S., Sharma, et al
  2015; 61 (6): 619-625

- **Reply.** *journal of pediatrics*
  Niemi, A., Enns, G. M.
  2015; 167 (5): 1173-1174

- **De novo truncating variants in the AHDC1 gene encoding the AT-hook DNA-binding motif-containing protein 1 are associated with intellectual disability and developmental delay.** *Cold Spring Harbor molecular case studies*
  Yang, H., Douglas, G., Monaghan, K. G., Retterer, K., Cho, M. T., Escobar, L. F., Tucker, M. E., Stoler, J., Rodan, L. H., Stein, D., Marks, W., Enns, G. M., Platt, et al
  2015; 1 (1)

- **A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency.** *The New England journal of medicine*
  Burton, B. K., Balwani, M., Feillet, F., Barić, I., Burrow, T. A., Camarena Grande, C., Coker, M., Consuelo-Sánchez, A., Deegan, P., Di Rocco, M., Enns, G. M., Erbe, R., Ezgu, et al
  2015; 373 (11): 1010-20

- **A congenital disorder of deglycosylation: Biochemical characterization of N-glycanase 1 deficiency in patient fibroblasts.** *Glycobiology*
  He, P., Grotzke, J. E., Ng, B. G., Gunel, M., Jafar-Nejad, H., Cresswell, P., Enns, G. M., Freeze, H. H.
  2015; 25 (8): 836-844

- **ClinGen - The Clinical Genome Resource** *NEW ENGLAND JOURNAL OF MEDICINE*
  Rehm, H. L., Berg, J. S., Brooks, L. D., Bustamante, C. D., Evans, J. P., Landrum, M. J., Ledbetter, D. H., Maglott, D. R., Martin, C. L., Nussbaum, R. L., Plon, S. E., Ramos, E. M., Sherry, et al
  2015; 372 (23): 2235-2242

- **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-?

- **Correlation of HMPAO uptake with live single cell synchrotron mid-Infrared spectromicroscopy in inherited mitochondrial disease**
  Holman, E., Chen, L., Holman, H., Enns, G., Blankenberg, F.
  SOC NUCLEAR MEDICINE INC.2015

- **Long-term safety and efficacy of sapropterin: The PKUDOS registry experience** *MOLECULAR GENETICS AND METABOLISM*
  Longo, N., Arnold, G. L., Pridjian, G., Enns, G. M., Ficicioglu, C., Parker, S., Cohen-Pfeffer, J. L.

2015; 114 (4): 557-563

- **Identification of HIBCH Gene Mutations Causing Autosomal Recessive Leigh Syndrome: A Gene Involved in Valine Metabolism** *PEDIATRIC NEUROLOGY*
  Soler-Alfonso, C., Enns, G. M., Koenig, M. K., Saavedra, H., Bonfante-Mejia, E., Northrup, H.
  2015; 52 (3): 361-365

- **Results of a Global Phase 3, Randomized, Double-Blind, Placebo-Controlled Trial Evaluating the Efficacy and Safety of Sebelipase Alfa as an Enzyme Replacement Therapy in Children and Adults with Lysosomal Acid Lipase Deficiency**
  Balwani, M., Burton, B., Baric, I., Bialer, M., Burrow, T. A., Camarena Grande, C., Coker, M., Consuelo Sanchez, A., Deegan, P., Di Rocco, M., Enns, G. M., Erbe, R. W., Ezgu, et al
  WILEY-BLACKWELL.2014: 1270A

- **Sebelipase alfa over 52 weeks reduces serum transaminases, liver volume and improves serum lipids in patients with lysosomal acid lipase deficiency** *JOURNAL OF HEPATOLOGY*
  Valayannopoulos, V., Malinova, V., Honzik, T., Balwani, M., Breen, C., Deegan, P. B., Enns, G. M., Jones, S. A., Kane, J. P., Stock, E. O., Tripuraneni, R., Eckert, S., Schneider, et al
  2014; 61 (5): 1135-1142

- **Sebelipase alfa over 52 weeks reduces serum transaminases, liver volume and improves serum lipids in patients with lysosomal acid lipase deficiency.** *Journal of hepatology*
  Valayannopoulos, V., Malinova, V., Honzík, T., Balwani, M., Breen, C., Deegan, P. B., Enns, G. M., Jones, S. A., Kane, J. P., Stock, E. O., Tripuraneni, R., Eckert, S., Schneider, et al
  2014; 61 (5): 1135-42

- **Abnormal Hepatocellular Mitochondria in Methylmalonic Acidemia** *ULTRASTRUCTURAL PATHOLOGY*
  Wilnai, Y., Enns, G. M., Niemi, A., Higgins, J., Vogel, H.
  2014; 38 (5): 309-314

- **Abnormal hepatocellular mitochondria in methylmalonic acidemia.** *Ultrastructural pathology*
  Wilnai, Y., Enns, G. M., Niemi, A., Higgins, J., Vogel, H.
  2014; 38 (5): 309-314

- **Clinical whole-exome sequencing: are we there yet?** *GENETICS IN MEDICINE*
  Atwal, P. S., Brennan, M., Cox, R., Niaki, M., Platt, J., Homeyer, M., Kwan, A., Parkin, S., Schelley, S., Slattery, L., Wilnai, Y., Bernstein, J. A., Enns, et al
  2014; 16 (9): 717-719

- **Clinical whole-exome sequencing: are we there yet?** *Genetics in medicine*
  Atwal, P. S., Brennan, M., Cox, R., Niaki, M., Platt, J., Homeyer, M., Kwan, A., Parkin, S., Schelley, S., Slattery, L., Wilnai, Y., Bernstein, J. A., Enns, et al
  2014; 16 (9): 717-719

- **Treatment of Mitochondrial Disorders: Antioxidants and Beyond** *JOURNAL OF CHILD NEUROLOGY*
  Enns, G. M.
  2014; 29 (9): 1235-1240

- **Points to Consider in the Clinical Use of NGS Panels for Mitochondrial Disease: An Analysis of Gene Inclusion and Consent Forms** *JOURNAL OF GENETIC COUNSELING*
  Platt, J., Cox, R., Enns, G. M.
  2014; 23 (4): 594-603

- **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

- **Monitoring mitochondrial disease related nephropathy and subclinical redox stress of the brain in the kd/kd murine model**
  Blankenberg, F., Enns, G.
  SOC NUCLEAR MEDICINE INC.2014

- **HMPAO uptake assay with t-BHP or irradiation of primary patient SURF-1 fibroblast cultures**
  Holman, E., Enns, G., Blankenberg, F.
  SOC NUCLEAR MEDICINE INC.2014

- **Clinical interpretation and implications of whole-genome sequencing.** *JAMA : the journal of the American Medical Association*
  Dewey, F. E., Grove, M. E., Pan, C., Goldstein, B. A., Bernstein, J. A., Chaib, H., Merker, J. D., Goldfeder, R. L., Enns, G. M., David, S. P., Pakdaman, N., Ormond, K. E., Caleshu, et al
  2014; 311 (10): 1035-1045

- **Clinical interpretation and implications of whole-genome sequencing.** *JAMA*
  Dewey, F. E., Grove, M. E., Pan, C., Goldstein, B. A., Bernstein, J. A., Chaib, H., Merker, J. D., Goldfeder, R. L., Enns, G. M., David, S. P., Pakdaman, N., Ormond, K. E., Caleshu, et al
  2014; 311 (10): 1035-1045

- **MITOCHONDRIAL COMPLEX CARE CLINIC MODEL - EMPOWERING FAMILIES AS PART OF THE CARE TEAM**
  Niemi, A., Cox, R., Platt, J., Wayman, K., Mark, J., Enns, G. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: 254

- **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

- **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

- **NEED FOR EVIDENCE-BASED RESEARCH TO ASSESS BENEFITS OF NUTRITION TREATMENTS FOR RARE INBORN ERRORS OF METABOLISM**
  Frazier, D. M., VanCalcar, S., Enns, G. M., McGuire, P. J.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: 295–96

- **Severe and rapid disease course in the natural history of infants with lysosomal acid lipase deficiency**
  Jones, S. A., Bernstein, D., Bialer, M., Dhawan, A., Hendriksz, C., Whitley, C. B., Banikazemi, M., Chang, A., Guardamagna, O., Raiman, J., Gamal, I., Selim, L., Cederbaum, et al
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2014: S57–S58

- **Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges.** *Mitochondrion*
  Parikh, S., Goldstein, A., Koenig, M. K., Scaglia, F., Enns, G. M., Saneto, R.
  2014; 14 (1): 26-33

- **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

- **Practice patterns of mitochondrial disease physicians in North America. Part 2: treatment, care and management** *MITOCHONDRION*
  Parikh, S., Goldstein, A., Koenig, M. K., Scaglia, F., Enns, G. M., Saneto, R.
  2013; 13 (6): 681-687

- **First-line genetic testing for mitochondrial disorders in the next-generation sequencing era: Comprehensive known disease gene panel or exome sequencing?**
  Bai, R., Haverfield, E., Higgs, J., Suchy, S. F., Arjona, D., Retterer, K., Smaoui, N., Richard, G., Bale, S., Kendall, F. D., Parikh, S., Gropman, A. L., Haas, et al
  ELSEVIER SCI LTD.2013: 934

- **Severe multi-systemic presentation of COX10 deficiency**
  Wilnai, Y., Cox, R., Bai, R., Enns, G. M.
  ELSEVIER SCI LTD.2013: 926

- **Mitochondrial NGS panels: Is more better?**
  Platt, J., Cox, R., Enns, G. M.
  ELSEVIER SCI LTD.2013: 919

- **Novel c.316G > C mutation in PDHA1 causes PDH deficiency**
  Khouzam, A., Platt, J., Cox, R., Enns, G. M.
  ELSEVIER SCI LTD.2013: 925

- **Molecular and clinical characterization of the myopathic form of mitochondrial DNA depletion syndrome caused by mutations in the thymidine kinase (TK2) gene** *MOLECULAR GENETICS AND METABOLISM*
  Chanprasert, S., Wang, J., Weng, S., Enns, G. M., Boue, D. R., Wong, B. L., Mendell, J. R., Perry, D. A., Sahenk, Z., Craigen, W. J., Climent Alcala, F. J., Pascual, J. M., Melancon, et al
  2013; 110 (1-2): 153-161

- **Clinical Effect and Safety Profile of Recombinant Human Lysosomal Acid Lipase in Patients With Cholesteryl Ester Storage Disease** *HEPATOLOGY*
  Balwani, M., Breen, C., Enns, G. M., Deegan, P. B., Honzik, T., Jones, S., Kane, J. P., Malinova, V., Sharma, R., Stock, E. O., Valayannopoulos, V., Wraith, J. E., Burg, et al
  2013; 58 (3): 950-957

- **Molecular and clinical characterization of the myopathic form of mitochondrial DNA depletion syndrome caused by mutations in the thymidine kinase (TK2) gene.** *Molecular genetics and metabolism*
  Chanprasert, S., Wang, J., Weng, S., Enns, G. M., Boué, D. R., Wong, B. L., Mendell, J. R., Perry, D. A., Sahenk, Z., Craigen, W. J., Alcala, F. J., Pascual, J. M., Melancon, et al
  2013; 110 (1-2): 153-161

- **Targeting proximal tubule mitochondrial dysfunction attenuates the renal disease of methylmalonic acidemia** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Manoli, I., Sysol, J. R., Li, L., Houillier, P., Garone, C., Wang, C., Zerfas, P. M., Cusmano-Ozog, K., Young, S., Trivedi, N. S., Cheng, J., Sloan, J. L., Chandler, et al
  2013; 110 (33): 13552-13557

- **Expanding research to provide an evidence base for nutritional interventions for the management of inborn errors of metabolism** *MOLECULAR GENETICS AND METABOLISM*
  Camp, K. M., Lloyd-Puryear, M. A., Yao, L., Groft, S. C., Parisi, M. A., Mulberg, A., Gopal-Srivastava, R., Cederbaum, S., Enns, G. M., Ershow, A. G., Frazier, D. M., Gohagan, J., Harding, et al
  2013; 109 (4): 319-328

- **Glutathione: A redox signature in monitoring EPI-743 therapy in children with mitochondrial encephalomyopathies** *MOLECULAR GENETICS AND METABOLISM*
  Pastore, A., Petrillo, S., Tozzi, G., Carrozzo, R., Martinelli, D., Dionisi-Vici, C., Di Giovamberardino, G., Ceravolo, F., Klein, M. B., Miller, G., Enns, G. M., Bertini, E., Piemonte, et al
  2013; 109 (2): 208-214

- **Comprehensive next-generation sequence analyses of the entire mitochondrial genome reveal new insights into the molecular diagnosis of mitochondrial DNA disorders** *GENETICS IN MEDICINE*
  Cui, H., Li, F., Chen, D., Wang, G., Truong, C. K., Enns, G. M., Graham, B., Milone, M., Landsverk, M. L., Wang, J., Zhang, W., Wong, L. C.
  2013; 15 (5): 388-394

- **Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of alpha-Dystroglycan** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Stevens, E., Carss, K. J., Cirak, S., Foley, R., Torelli, S., Willer, T., Tambunan, D. E., Yau, S., Brodd, L., Sewry, C. A., Feng, L., Haliloglu, G., Orhan, et al
  2013; 92 (3): 354-365

- **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

- **Long term safety and clinical activity of SBC-102, a recombinant human lysosomal acid lipase (rhLAL), in patients with late onset LAL deficiency** *9th Annual Research Meeting of the Lysosomal-Disease-Network (LDN)*

Balwani, M., Malinova, V., Sharma, R., Valayannopoulos, V., Stock, E. O., Boyadjiev, S. A., Kessler, B., Deegan, P., Enns, G. M., Breen, C., Kane, J. P., Schneider, E., Quinn, et al

ACADEMIC PRESS INC ELSEVIER SCIENCE.2013: S22–S23

- **ARTHROGRYPOSIS, RENAL DYSFUNCTION AND CHOLESTASIS (ARC) SYNDROME: A NEW PATIENT CASE REPORT** *Western Regional Meeting of the American-Federation-for-Medical-Research*

  Brennan, M., SLATTERY, L., Esplin, E., Enns, G. M., Hudgins, L., Manning, M.

  LIPPINCOTT WILLIAMS & WILKINS.2013: 188–88

- **Pediatric Liver Transplantation as Definitive Therapy for Urea Cycle Disorders** *13th Annual State of the Art Winter Symposium of the American-Society-of-Transplant-Surgeons (ASTS) Held in Conjunction with the NATCO Symposium for Advanced Transplant Professionals*

  Kim, I. K., Niemi, A., Krueger, C., Enns, G., Esquivel, C. O.

  WILEY-BLACKWELL.2013: 86–86

- **Enzyme Replacement with Recombinant Human Lysosomal Acid Lipase (rhLAL) in Patients with Cholesteryl Ester Storage Disease, the Late Onset Form of LAL Deficiency, Produces Sustained Decreases in Transaminases and Reduction in Liver Fat Content** *63rd Annual Meeting of the American-Association-for-the-Study-of-Liver-Diseases (AASLD)*

  Jones, S., Enns, G. M., Balwani, M., Breen, C., Sharma, R., Deegan, P., Malinova, V., Honzik, T., Valayannopoulos, V., Schneider, E., Burg, J., Quinn, A. G.

  WILEY-BLACKWELL.2012: 824A–825A

- **Atypical Amyoplasia Congenita in an Infant With Leigh Syndrome: A Mitochondrial Cause of Severe Contractures?** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*

  Wilnai, Y., Seaver, L. H., Enns, G. M.

  2012; 158A (9): 2353-2357

- **RECOMBINANT HUMAN LYSOSOMAL ACID LIPASE (LAL) DEMONSTRATES PHARMACODYNAMIC ACTIVITY IN CHOLESTERYL ESTER STORAGE DISEASE (CESD), THE LATE ONSET FORM OF LAL DEFICIENCY**

  Jones, S., Enns, G., Balwani, M., Breen, C., Sharma, R., Deegan, P., Malinova, V., Honzik, T., Valayannopoulos, V., Schneider, E., Burg, J., Quinn, A. G.

  SPRINGER.2012: S11–S11

- **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

- **Outcome of infants diagnosed with 3-methyl-crotonyl-CoA-carboxylase deficiency by newborn screening** *MOLECULAR GENETICS AND METABOLISM*

  Arnold, G. L., Salazar, D., Neidich, J. A., Suwannarat, P., Graham, B. H., Lichter-Konecki, U., Bosch, A. M., Cusmano-Ozog, K., Enns, G., Wright, E. L., Lanpher, B. C., Owen, N. N., Lipson, et al

  2012; 106 (4): 439-441

- **Propionic acidemia: To liver transplant or not to liver transplant?** *PEDIATRIC TRANSPLANTATION*

  Chapman, K. A., Summar, M. L., Enns, G. M.

  2012; 16 (3): 209-210

- **Pediatric Liver Transplantation as Definitive Therapy for Urea Cycle Disorders** *24th Annual Meeting of the European-Association-for-Cardiothoracic-Surgery*

  Kim, I., Niemi, A., Enns, G., Esquivel, C.

  WILEY-BLACKWELL.2012: 344–344

- **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

- **Mutations in KAT6B, Encoding a Histone Acetyltransferase, Cause Genitopatellar Syndrome** *AMERICAN JOURNAL OF HUMAN GENETICS*

  Campeau, P. M., Kim, J. C., Lu, J. T., Schwartzentruber, J. A., Abdul-Rahman, O. A., Schlaubitz, S., Murdock, D. M., Jiang, M., Lammer, E. J., Enns, G. M., Rhead, W. J., Rowland, J., Robertson, et al

  2012; 90 (2): 282-289

- **Initial Human Experience with SBC-102, a Recombinant Enzyme Replacement Therapy in Adults with Lysosomal Acid Lipase Deficiency** *8th Annual World Symposium of the Lysosomal Disease Network*
  Enns, G., Balwani, M., Deegan, P., Malinova, V. R., Honzik, H., Sharma, R., Valayannopoulos, V., Wraith, E., Schneider, E., Burg, J., Quinn, A.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2012: S29–S29

- **Natural history of propionic acidemia** *MOLECULAR GENETICS AND METABOLISM*
  Pena, L., Franks, J., Chapman, K. A., Gropman, A., Mew, N. A., Chakrapani, A., Island, E., MacLeod, E., Matern, D., Smith, B., Stagni, K., Sutton, V. R., Ueda, et al
  2012; 105 (1): 5-9

- **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)

- **Length of prenatal exposure to selective serotonin reuptake inhibitor (SSRI) antidepressants: effects on neonatal adaptation and psychomotor development** *PSYCHOPHARMACOLOGY*
  Casper, R. C., Gilles, A. A., Fleisher, B. E., Baran, J., Enns, G., Lazzeroni, L. C.
  2011; 217 (2): 211-219

- **alpha-Tocotrienol quinone modulates oxidative stress response and the biochemistry of aging** *BIOORGANIC & MEDICINAL CHEMISTRY LETTERS*
  Shrader, W. D., Amagata, A., Barnes, A., Enns, G. M., Hinman, A., Jankowski, O., Kheifets, V., Komatsuzaki, R., Lee, E., Mollard, P., Murase, K., Sadun, A. A., Thoolen, et al
  2011; 21 (12): 3693-3698

- **Clinical Report-Health Supervision for Children With Fragile X Syndrome** *PEDIATRICS*
  Hersh, J. H., Saul, R. A., Saal, H. M., Braddock, S. R., Enns, G. M., Gruen, J. R., Perrin, J. M., Tarini, B. A., Hanson, J. W., Lloyd-Puryear, M. A., Musci, T. J., Rasmussen, S. A., Spire, et al
  2011; 127 (5): 994-1006

- **Long-term follow-up of a patient with early onset CBLG disease** *34th Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD 2011)*
  Niemi, A. K., Cusmano-Ozog, K., Rosenblatt, D. S., Enns, G. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2011: 306–7

- **Baseline characteristics of PKU patients enrolled in the PKUDOS registry** *34th Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD 2011)*
  Burton, B. K., Longo, N., Arnold, G. L., Enns, G. M., Andersson, H. C., Mofidi, S., Peck, D., Prado, B. C., Pridjian, G., Waisbren, S., White, D.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2011: 273–73

- **MITOCHONDRIAL PATHOLOGY IN ANGELMAN SYNDROME** *Western Regional Meeting of the American-Federation-for-Medical-Research*
  Niemi, A., Cox, R., Scharfe, C., Cherry, A., Enns, G.
  LIPPINCOTT WILLIAMS & WILKINS.2011: 165–66

- **Suboptimal outcomes in patients with PKU treated early with diet alone: Revisiting the evidence** *MOLECULAR GENETICS AND METABOLISM*
  Enns, G. M., Koch, R., Brumm, V., Blakely, E., Suter, R., Jurecki, E.
  2010; 101 (2-3): 99-109

- **Long-term outcome following pediatric liver transplantation for metabolic disorders** *PEDIATRIC TRANSPLANTATION*
  Stevenson, T., Millan, M. T., Wayman, K., Berquist, W. E., Sarwal, M., Johnston, E. E., Esquivel, C. O., Enns, G. M.
  2010; 14 (2): 268-275

- **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

- **EVIDENCE OF REDOX IMBALANCE IN A PATIENT WITH METHYLMALONIC ACIDEMIA (mut0)** *33rd Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD 2010)*
  Cusmano-Ozog, K., Moore, T., Niemi, A., Zadeh, N., Cowan, T. M., Enns, G. M.

ACADEMIC PRESS INC ELSEVIER SCIENCE.2010: 210–11

- **Nitrogen sparing therapy revisited 2009** *3rd International Satellite on Urea Cycle Disorders*
  Enns, G. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2010: S65–S71
- **Future treatment strategies in phenylketonuria** *MOLECULAR GENETICS AND METABOLISM*
  van Spronsen, F. J., Enns, G. M.
  2010; 99: S90-S95
- **Pathological evidence of Wolman's disease following hematopoietic stem cell transplantation despite correction of lysosomal acid lipase activity** *BONE MARROW TRANSPLANTATION*
  Gramatges, M. M., Dvorak, C. C., Regula, D. P., Enns, G. M., Weinberg, K., Agarwal, R.
  2009; 44 (7): 449-450
- **Hypoplastic Glomerulocystic Kidney Disease and Hepatoblastoma A Potential Association not Caused by Mutations in Hepatocyte Nuclear Factor 1 beta** *JOURNAL OF PEDIATRIC HEMATOLOGY ONCOLOGY*
  Abdul-Rahman, O. A., Edghill, E. L., Kwan, A., Enns, G. M., Hattersley, A. T.
  2009; 31 (7): 527-529
- **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
- **A novel homozygous SCO2 mutation, p.G193S, causing fatal infantile cardioencephalomyopathy** *CLINICAL NEUROPATHOLOGY*
  Mobley, B. C., Enns, G. M., Wong, L., Vogel, H.
  2009; 28 (2): 143-149
- **Successful pregnancy and cesarean delivery via noninvasive ventilation in mitochondrial myopathy** *JOURNAL OF PERINATOLOGY*
  Yuan, N., El-Sayed, Y. Y., Ruoss, S. J., Riley, E., Enns, G. M., Robinson, T. E.
  2009; 29 (2): 166-167
- **Neurologic Damage and Neurocognitive Dysfunction in Urea Cycle Disorders** *SEMINARS IN PEDIATRIC NEUROLOGY*
  Enns, G. M.
  2008; 15 (3): 132-139
- **Cell-based therapies for metabolic liver disease** *MOLECULAR GENETICS AND METABOLISM*
  Enns, G. M., Millan, M. T.
  2008; 95 (1-2): 3-10
- **Maternal phenylketonuria** *PEDIATRICS*
  Saal, H. M., Braddock, S. R., Bull, M. J., Enns, G., Gruen, J. R., Mendelsohn, N. J., Saul, R. A.
  2008; 122 (2): 445-449
- **Dopa-responsive dystonia presenting as delayed and awkward gait** *PEDIATRIC NEUROLOGY*
  Cheyette, B. N., Cheyette, S. N., Cusmano-Ozog, K., Enns, G. M.
  2008; 38 (4): 273-275
- **Central nervous system therapy for lysosomal storage disorders** *NEUROSURGICAL FOCUS*
  Enns, G. M., Huhn, S. L.
  2008; 24 (3-4)
- **Health supervision for children with neurofibromatosis** *PEDIATRICS*
  Schaefer, G. B., Bull, M. J., Enns, G. M., Gruen, J. R., Hersh, J. H., Mendelsohn, N. J., Saal, H. M.
  2008; 121 (3): 633-642
- **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

- **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

- **Clinical and molecular features of mitochondrial DNA depletion due to mutations in deoxyguanosine kinase** *HUMAN MUTATION*
  Dimmock, D. P., Zhang, Q., Dionisi-Vici, C., Carrozzo, R., Shieh, J., Tang, L. Y., Truong, C., Schmitt, E., Sifry-Platt, M., Lucioli, S., Santorelli, F. M., Ficicioglu, C. H., Rodriguez, et al
  2008; 29 (2): 330-331

- **Inborn error of metabolism presenting as malformation syndrome: Hydrocephalus as a manifestation of cobalamin D disease** *Western Regional Meeting of the American-Federation-for-Medical-Research*
  Bernstein, J. A., Lee-Messer, C., Cusmano, K., Rosenblatt, D. S., Cowan, T., Enns, G. M.
  LIPPINCOTT WILLIAMS & WILKINS.2008: 196–96

- **Glutaric acidemia type I: a neurosurgical perspective** *JOURNAL OF NEUROSURGERY*
  Hou, L. C., Veeravagu, A., Hsu, A. R., Enns, G. M., Huhn, S. L.
  2007; 107 (2): 167-172

- **Cobalamin C disease identified by expanded newborn screening: The California experience.** *Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD 2007)*
  Cusmano-Ozog, K., Lorey, F., LEVINE, S., Martin, M., Nicholas, E., Packman, S., Rosenblatt, D. S., Cederbaum, S. D., Cowan, T. M., Enns, G. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2007: 240–40

- **Early identification and aggressive treatment of cobalamin C disease.** *Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD 2007)*
  Cusmano-Ozog, K., Martin, M., Nicholas, E., Packman, S., Rosenblatt, D. S., Cowan, T. M., Enns, G. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2007: 249–50

- **Cobalamin c disease and expanded newborn screening: The California experience.** *8th Conference of the Western Student Medical Research Forum/Western Section of the American-Federation-for-Medical-Research/Western Association-of-Physicians/Western-Society-for-Pediatric-Research/Western-Society-for-Clinical-Investigation*
  Cusmano-Ozog, K., Lorey, F., LEVINE, S., Martin, M., Nicholas, E., Packman, S., Rosenblatt, D. S., Cowan, T. M., Enns, G. M.
  LIPPINCOTT WILLIAMS & WILKINS.2007: S90–S90

- **Systemic hyalinosis: A distinctive early childhood-onset disorder characterized by mutations in the anthrax toxin receptor 2 gene (ANTRX2)** *PEDIATRICS*
  Shieh, J. T., Swidler, P., Martignetti, J. A., Ramirez, M. C., Balboni, I., Kaplan, J., Kennedy, J., Abdul-Rahman, O., Enns, G. M., Sandborg, C., Slavotinek, A., Hoyme, H. E.
  2006; 118 (5): E1485-E1492

- **Introduction to the newborn screening fact sheets** *PEDIATRICS*
  Kaye, C. I., Schaefer, G. B., Bull, M. J., Enns, G. M., Gruen, J. R., Hersh, J. H., Mendelsohn, N. J., Saal, H. M., Goldberg, J. D., Hanson, J. W., Lloyd-Puryear, M. A., Rasmussen, S. A., Spire, et al
  2006; 118 (3): 1304-1312

- **Immunohistochemical analysis of cytochrome oxidase deficiency using fixed tissues**
  Vogel, H., Masek, M., Gallagher, R., Enns, G.
  BLACKWELL PUBLISHING.2006: S167

- **Genitopatellar syndrome: expanding the phenotype and excluding mutations in LMX1B and TBX4.** *American journal of medical genetics. Part A*
  Abdul-Rahman, O. A., La, T. H., Kwan, A., Schlaubitz, S., Barsh, G. S., Enns, G. M., Hudgins, L.
  2006; 140 (14): 1567-1572

- **Genitopatellar syndrome: Expanding the phenotype and excluding mutations in LMX1B and TBX4** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Abdul-Rahman, O. A., La, T. H., Kwan, A., Schlaubitz, S., Barsh, G. S., Enns, G. M., Hudgins, L.
  2006; 140A (14): 1567-1572

- **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

- **Identification of three novel mutations in the dihydropyrimidine dehydrogenase gene associated with altered pre-mRNA splicing or protein function (vol 386, pg 319, 2005)** *BIOLOGICAL CHEMISTRY*
  Van Kuilenburg, A. B., Meinsma, R., Beke, E., Bobba, B., Boffi, P., Enns, G. M., Witt, D. R., Dobritzsch, D.
  2005; 386 (10): 1075

- **Management of methylmalonic acidaemia by combined liver-kidney transplantation** *JOURNAL OF INHERITED METABOLIC DISEASE*
  Nagarajan, S., Enns, G. M., Millan, M. T., Winter, S., Sarwal, M. M.
  2005; 28 (4): 517-524

- **Postpartum "psychosis" in mild argininosuccinate synthetase deficiency** *OBSTETRICS AND GYNECOLOGY*
  Enns, G. M., O'Brien, W. E., Kobayashi, K., Shinzawa, H., Pellegrino, J. E.
  2005; 105 (5): 1244-1246

- **Identification of three novel mutations in the dihydropyrimidine dehydrogenase gene associated with altered pre-mRNA splicing or protein function** *BIOLOGICAL CHEMISTRY*
  van Kuilenburg, A. B., Meinsma, R., Beke, E., Bobba, B., Boffi, P., Enns, G. M., Witt, D. R., Dobritzsch, D.
  2005; 386 (4): 319-324

- **Two cases of transient 5-oxoprolinuria resolving with antioxidant therapy.** *Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD 2005)*
  Huguenin, S. M., Gallagher, R. C., Lyons, M. J., Kwan, T., Enns, G. M., Cowan, T. M.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2005: 223–24

- **Mutations in POMT1 are found in a minority of patients with Walker-Warburg syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Currier, S. C., Lee, C. K., Chang, B. S., Bodell, A. L., Pai, G. S., Job, L., Lagae, L. G., Al-Gazali, L. I., Eyaid, W. M., Enns, G., Dobyns, W. B., Walsh, C. A.
  2005; 133A (1): 53-57

- **Mild developmental delay in terminal chromosome 6p deletion** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Chen, K. M., Cherry, A. M., Hahn, J. S., Enns, G. M.
  2004; 129A (2): 201-205

- **Terminal 22q deletion syndrome: A newly recognized cause of speech and language disability in the autism spectrum** *PEDIATRICS*
  Manning, M. A., Cassidy, S. B., Clericuzio, C., Cherry, A. M., Schwartz, S., Hudgins, L., Enns, G. M., Hoyme, H. E.
  2004; 114 (2): 451-457

- **Head imaging abnormalities in dihydropyrimidine dehydrogenase deficiency** *JOURNAL OF INHERITED METABOLIC DISEASE*
  Enns, G. M., Barkovich, A. J., van Kuilenburg, A. B., Manning, M., Sanger, T., Witt, D. R., Van Gennip, A. H.
  2004; 27 (4): 513-522

- **Methotrexate/misoprostol embryopathy: Report of four cases resulting from failed medical abortion** *Bryan D Hall Festschrift 2003*
  Adam, M. P., Manning, M. A., Beck, A. E., Kwan, A., Enns, G. M., Clericuzio, C., Hoyme, H. E.
  WILEY-LISS.2003: 72–78

- **Compensatory amplification of mtDNA in a patient with a novel deletion/duplication and high mutant load** *JOURNAL OF MEDICAL GENETICS*
  Wong, L. J., Perng, C. L., Hsu, C. H., Bai, R. K., Schelley, S., Vladutiu, G. D., Vogel, H., Enns, G. M.
  2003; 40 (11)

- **Congenital disorder of glycosylation Ic in patients of Indian origin** *MOLECULAR GENETICS AND METABOLISM*
  Newell, J. W., Seo, N. S., Enns, G. M., McCraken, M., Mantovani, J. F., Freeze, H. H.
  2003; 79 (3): 221-228

- **Severe liver disease in urea cycle disorders.** *Western Regional Meeting of the American-Federation-for-Medical-Research*
  Traynor, J. D., Tuchman, M., Manning, M. A., Goodman, S. I., Enns, G. M.
  LIPPINCOTT WILLIAMS & WILKINS.2003: S118–S118

- **Bilateral cystic lung disease in a monoamnionic twin of an anencephalic: A case of glial heterotopia in the lungs.** *Western Regional Meeting of the American-Federation-for-Medical-Research*
  Anderson, J. M., Adams, M. L., Morgan, T., Robinson, T., Enns, G. M., Keller, K. A., Sylvester, K. G., Hintz, S. R.
  LIPPINCOTT WILLIAMS & WILKINS.2003: S119–S120

- **Clinical and molecular features of congenital disorder of glycosylation in patients with type 1 sialotransferrin pattern and diverse ethnic origins** *JOURNAL OF PEDIATRICS*
  Enns, G. M., Steiner, R. D., Buist, N., Cowan, C., Leppig, K. A., McCracken, M. F., Westphal, V., Freeze, H. H., O'Brien, J. F., Jaeken, J., Matthijs, G., Behera, S., Hudgins, et al
  2002; 141 (5): 695-700

- **Post-partum "psychosis" in mild argininosuccinate synthetase deficiency.** *52nd Annual Meeting of the American-Society-of-Human-Genetics*
  Enns, G. M., O'Brien, W. E., Kobayashi, K., Shinzawa, H., Hart, K., Gao, H. Z., Tabata, A., Saheki, T.
  CELL PRESS.2002: 425–25

- **The adolescent with an inborn error of metabolism: medical issues and transition to adulthood.** *Adolescent medicine (Philadelphia, Pa.)*
  Enns, G. M., Packman, W.
  2002; 13 (2): 315-?

- **Early neonatal diagnosis of long-chain 3-hydroxyacyl coenzyme A dehydrogenase and mitochondrial trifunctional protein deficiencies** *MOLECULAR GENETICS AND METABOLISM*
  Hintz, S. R., Matern, D., Strauss, A., Bennett, M. J., Hoyme, H. E., Schelley, S., Kobori, J., Colby, C., Lehman, N. L., Enns, G. M.
  2002; 75 (2): 120-127

- **Redefining a case of Jarcho-Levin syndrome as Oculo-Auriculo-Vertebral Spectrum.**
  Cheng, S. F., Enns, G. M., Golabi, M., Blumberg, B., Kostiner, D. R.
  CELL PRESS.2001: 300–300

- **Functional analysis of novel mutations in a congenital disorder of glycosylation Ia patient with mixed Asian ancestry** *MOLECULAR GENETICS AND METABOLISM*
  Westphal, V., Enns, G. M., McCracken, M. F., Freeze, H. H.
  2001; 73 (1): 71-76

- **Clinical course and biochemistry of sialuria** *JOURNAL OF INHERITED METABOLIC DISEASE*
  Enns, G. M., Seppala, R., Musci, T. J., Weisiger, K., Ferrell, L. D., Wenger, D. A., Gahl, W. A., Packman, S.
  2001; 24 (3): 328-336

- **Natural history of branchio-oto-renal (BOR) syndrome.**
  Hudgins, L., Jones, M. C., Olney, R. S., Enns, G. M., Schelley, S. L.
  CELL PRESS.2000: 56–56

- **Molecular correlations in phenylketonuria: Mutation patterns and corresponding biochemical and clinical phenotypes in a heterogeneous California population** *PEDIATRIC RESEARCH*
  Enns, G. M., Martinez, D. R., Kuzmin, A. I., Koch, R., Wakeem, C. K., Woo, S. L., Eisensmith, R. C., Packman, S.
  1999; 46 (5): 594-602

- **Apparent cyclophosphamide (cytoxan) embryopathy: A distinct phenotype?** *AMERICAN JOURNAL OF MEDICAL GENETICS*
  Enns, G. M., Roeder, E., Chan, R. T., Catts, Z. A., Cox, V. A., Golabi, M.
  1999; 86 (3): 237-241

- **Progressive neurological deterioration and MRI changes in cblC methylmalonic acidaemia treated with hydroxocobalamin** *JOURNAL OF INHERITED METABOLIC DISEASE*
  Enns, G. M., Barkovich, A. J., Rosenblatt, D. S., Fredrick, D. R., Weisiger, K., Ohnstad, C., Packman, S.
  1999; 22 (5): 599-607

- **Severe congenital anomalies requiring transplantation in children with Kabuki syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS*
  Ewart-Toland, A., Enns, G. M., Cox, V. A., Mohan, G. C., Rosenthal, P., Golabi, M.
  1998; 80 (4): 362-367

- **Congenital diaphragmatic defects and associated syndromes, malformations, and chromosome anomalies: A retrospective study of 60 patients and literature review** *AMERICAN JOURNAL OF MEDICAL GENETICS*

Enns, G. M., Cox, V. A., Goldstein, R. B., Gibbs, D. L., Harrison, M. R., Golabi, M.
1998; 79 (3): 215-225