



Gregory Enns

Professor of Pediatrics (Genetics)
Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

- **Pediatric Genetics**

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Bio

CLINICAL FOCUS

- Biochemical Genetics
- Mitochondrial Diseases
- Medical Biochemical Genetics

ACADEMIC APPOINTMENTS

- Professor - University Medical 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

- **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
- **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
- **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
- **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., Niemi, 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
 - **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., Tammara, 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 li, 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., Dahmouh, 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 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
 - **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. A., Tise, C. G., Narang, A., Grimm, P. C., Enns, G. M., Lee, C. U.
2021; 27: 100765
 - **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., Bloodworth, 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**
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