Stanford

Gregory Enns

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

Pediatrics - Medical Genetics

CLINICAL OFFICES

- **Pediatric Genetics**

  730 Welch Rd 2A

  Palo Alto, CA 94304

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

Bio

CLINICAL FOCUS

- Clinical Genetics
- Biochemical Genetics
- Mitochondrial Diseases

ACADEMIC APPOINTMENTS

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

PROFESSIONAL EDUCATION

- Board Certification: Clinical Genetics, American Board of Medical Genetics and Genomics (1999)
- Board Certification: Clinical Biochemical Genetics, American Board of Medical Genetics and Genomics (1999)
- Residency: Children's Hospital Los Angeles (1995) CA
- Medical Education: University of St Andrews (1990) Scotland
- Fellowship: UCSF Medical Center (1998) CA

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Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

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

CLINICAL TRIALS

- A Multicenter Study of SBC-102 (Sebelipase Alfa) in Patients With Lysosomal Acid Lipase Deficiency/ ARISE (Acid Lipase Replacement Investigating Safety and Efficacy), Recruiting
- A Phase 1/2 Study of AEB1102 in Patients With Arginase I Deficiency, Recruiting
- International Collaborative Gaucher Group (ICGG) Gaucher Registry, Recruiting
- Mucopolysaccharidosis I (MPS I) Registry, Recruiting
- North American Mitochondrial Disease Consortium Patient Registry and Biorepository (NAMDC), Recruiting
- Safety and Efficacy Study of EPI-743 in Children With Leigh Syndrome, Recruiting

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Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (Phd Program)

Publications

PUBLICATIONS

- **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
- **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

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